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syn-drome (sin'drōm) [Gr. *syndromē* concurrence]

a set of symptoms that occur together; the sum of signs of any morbid state; a symptom complex. In genetics, a pattern of multiple malformations thought to be pathogenetically related. See also [disease](#).

Syndrome

For terms not found here, see also under disease.

22q11 deletion syndrome, hemizygous deletion of a 1.5 to 3.0 Mb region of chromosome 22q11.2 as a result of defective recombination in meiosis. It has a highly variable phenotype, partly as a function of the extent and precise location of the deletion, and includes various named syndromes with different presentations that have been shown to have the same genetic basis, including [DiGeorge syndrome](#) and [velocardiofacial syndrome](#) (qq.v.).

Agenaes syndrome, an autosomal recessive syndrome consisting of intrahepatic cholestasis and edema of the lower limbs.

Aarskog syndrome, **Aarskog-Scott syndrome**, an X-linked syndrome characterized by ocular hypertelorism, anteverted nostrils, broad upper lip, peculiar scrotal "shawl" above the penis, and small hands. Called also [faciogenital dysplasia](#) and [faciodigitogenital s.](#)

Aase syndrome, a familial syndrome characterized by mild growth retardation, hypoplastic anemia, variable leukocytopenia, triphalangeal thumbs, narrow shoulders, and late closure of fontanels, and occasionally by cleft lip, cleft palate, retinopathy, and web neck. A recessive mode of inheritance has been suggested.

abdominal muscle deficiency syndrome, [prune-belly s.](#)

abstinence syndrome, [substance withdrawal](#).

achalasia-addisonian syndrome, [Allgrove s.](#)

Achard syndrome, arachnodactyly associated with receding mandible and joint laxity limited to the hands and feet.

Achard-Thiers syndrome, [masculinization](#) with hirsutism and [adult-onset diabetes mellitus](#) in postmenopausal women resulting from overproduction of adrenocortical androgens.

acquired immune deficiency syndrome, **acquired immunodeficiency syndrome**, the most severe manifestation of disease due to infection with [human immunodeficiency virus](#) (HIV). The criteria established by the Centers for Disease Control and Prevention for the diagnosis of AIDS include (1) presence of certain [opportunistic infections](#) indicating an underlying defect in [cell-mediated immunity](#) in the absence of known causes of underlying [immunodeficiency](#) or other host defense defects; or (2) [CD4+ cell](#) count of less than 200/mL; or (3) [CD4+ cell](#) percentage of less than 14 per cent. See also [human immunodeficiency virus infection](#), under infection.

actinic reticuloid syndrome, [chronic actinic dermatitis](#).

acute brain syndrome, [delirium](#).

acute chest syndrome, a complex of symptoms seen in patients with [sickle cell disease](#), often due to a bacterial infection or to infarction of lung tissue; characteristics include severe chest pain, dyspnea, tachypnea, fever, excessive leukocytosis, pulmonary edema, and sometimes petechiae on the chest or conjunctivae as well as fat emboli. Death may result from severe pulmonary complications.

acute coronary syndrome, a classification encompassing clinical presentations ranging from [unstable angina](#) through [non-Q wave infarction](#), sometimes also including [Q wave infarction](#).

acute nephritic syndrome, the sudden onset of [hematuria](#), [proteinuria](#), diminished urine production, [azotemia](#), [hypertension](#), and [edema](#); the clinical manifestation of [acute glomerulonephritis](#).

acute organic brain syndrome, [delirium](#), occasionally, a term used to denote the acute form of an organic mental syndrome.

acute radiation syndrome, a syndrome caused by exposure to a whole-body dose of over 1 gray of ionizing radiation. Symptoms, whose severity and time of onset depend on the size of the dose, include erythema, nausea and vomiting, fatigue, diarrhea, fever, petechiae, bleeding from the mucous membranes, reduction in the number of lymphocytes, granulocytes, and platelets, gastrointestinal hemorrhage, epilation, hypotension, tachycardia, and dehydration; death may occur within hours or weeks of exposure.

acute respiratory distress syndrome, fulminant pulmonary interstitial and alveolar edema, which usually develops within a few days after an initiating trauma; it is thought to result from alveolar injury that has led to increased capillary permeability. Called also [adult respiratory distress s.](#) and [shock lung](#).

acute retinal necrosis syndrome, [acute retinal necrosis](#).

Adams-Stokes syndrome, episodic cardiac arrest and syncope due to failure of normal and escape pacemakers, with or without ventricular fibrillation; it is the principal clinical manifestation of severe heart block. Called also [Adams-Stokes syncope](#), [Morgagni-Adams-Stokes s.](#), and [Stokes-Adams s.](#) or [syncope](#).

addisonian syndrome, the complex of symptoms resulting from adrenocortical insufficiency; see [Addison disease](#), under disease.

addisonian-achalasia syndrome, [Allgrove s.](#)

Adie syndrome, a syndrome consisting of a pathological pupil reaction (tonic pupil) with a myotonic condition on accommodation; the pupil on the affected side contracts (for near vision) and dilates more slowly than the pupil on the opposite side. In direct or indirect light, the affected pupil either does not react at all or reacts in an abnormal fashion. Certain tendon reflexes are absent or diminished, usually the patellar reflexes, but there are no motor or sensory disturbances or other demonstrable changes indicating disease of the nervous system. Called also [Holmes-Adie s.](#)

adiposogenital syndrome, see under [dystrophy](#).

adrenogenital syndrome, a general term for the group of syndromes in which inappropriate [masculinization](#) or [feminization](#), sometimes with [precocious puberty](#), results from disorders of adrenal function that also affect gonadal steroidogenesis; it includes congenital adrenal hyperplasia and tumors of the adrenal cortex.

adult respiratory distress syndrome, [acute respiratory distress s.](#)

AEC syndrome, [Hay-Wells s.](#)

afferent loop syndrome, chronic partial obstruction of the proximal loop of duodenum and jejunum after partial gastrectomy and gastrojejunostomy, resulting in duodenal distention, pain, and nausea after the person ingests food.

aglossia-actactylia syndrome, [hypoglossia-hypodactyly s.](#)

Ahumada-del Castillo syndrome, [galactorrhea-amenorrhea syndrome](#) with low [gonadotropin](#) secretion.

Aicardi syndrome, a syndrome affecting female infants, characterized by agenesis of the corpus callosum, large discrete areas of chorioretinopathy, spasms and tonic seizures, and mental retardation.

akinetic-rigid syndrome, muscular rigidity with varying degrees of slowness of movement; seen in parkinsonism and disorders of the basal

ganglia.

Alagille syndrome, an autosomal dominant syndrome of neonatal jaundice, cholestasis with peripheral pulmonic stenosis, and occasionally septal defects or patent ductus arteriosus, due to paucity or absence of intrahepatic bile ducts; it is characterized by unusual facies and ocular, vertebral, and nervous system abnormalities.

Alajouanine syndrome, symmetric lesions of the sixth and seventh cranial nerves with bilateral facial paralysis and bilateral lateral rectus palsy of the eyeball, associated with bilateral clubfoot. Cf. [Möbius s.](#)

albinism-deafness syndrome, an X-linked syndrome consisting of [deafness](#) of varying degrees with [piebaldism](#) or [albinism](#); some authorities consider it an allelic variant of [Wardenburg syndrome type 2](#).

Albright syndrome, **Albright-McCune-Sternberg syndrome**, [polyostotic fibrous dysplasia](#), patchy dermal pigmentation, and endocrine dysfunction. Called also [McCune-Albright s.](#)

Aldrich syndrome, [Wiskott-Aldrich s.](#)

Alezzandrini syndrome, a rare disorder of unknown pathogenesis, characterized by unilateral [tapetoretinal degeneration](#) followed by facial [vitiligo](#) and [poliosis](#) on the same side, sometimes associated with deafness.

"Alice in Wonderland" syndrome, a delusional state manifested by depersonalization, alteration in the sense of the passage of time, distorted perception of objects, hallucinations, and other delusions or illusions. It may be associated with schizophrenia, epilepsy, migraine, diseases of the parietal lobe, hypnagogic states, or the use of hallucinogenic drugs.

alien hand syndrome, **alien limb syndrome**, involuntary but apparently purposeful movements of a hand or a limb, which is perceived by the patient to be controlled by an outside force; seen with lesions of the corpus callosum or frontal lobe and with corticobasal degeneration.

Allan-Herndon-Dudley syndrome, an X-linked syndrome caused by mutations in the SLC16A2 gene (locus: Xq13.2), which encodes a thyroid hormone transporter, characterized by severe mental retardation, dysarthria, athetoid movements, muscle hypoplasia, and spastic paraplegia.

Allemann syndrome, the association of double kidney and clubbed fingers, sometimes associated with facial asymmetry and degeneration of various motor nerves.

Allgrove syndrome, glucocorticoid deficiency with achalasia and alacrima; inherited as an autosomal recessive trait; called also [achalasia-addisonian s.](#), [addisonian-achalasia s.](#), and [triple-A s.](#)

Alport syndrome, a hereditary disorder characterized by progressive [sensorineural hearing loss](#), progressive [pyelonephritis](#) or [glomerulonephritis](#), and occasionally ocular defects. There are various subtypes, some with autosomal dominant inheritance, some autosomal recessive, and others X-linked.

Alström syndrome, an autosomal recessive syndrome of retinitis pigmentosa with nystagmus and early loss of central vision, deafness, obesity, and diabetes mellitus.

amnesic syndrome, **amnesic syndrome**, **amnesic-confabulatory syndrome**, a mental disorder characterized by impaired memory with anterograde and sometimes retrograde amnesia in a normal state of consciousness; i.e., the syndrome does not include the impaired memory seen in dementia or delirium. There may be disorientation, confabulation, and lack of insight into the memory deficit. The most common cause is thiamine deficiency from chronic alcohol abuse ([Wernicke-Korsakoff syndrome](#)), but it may also result from any pathological process causing bilateral damage to parts of the medial temporal lobe or diencephalon, such as the hippocampal formations, mammillary bodies, or dorsal medial nuclei of the thalamus. Other causes include head trauma, brain tumors, infarction, cerebral hypoxia, carbon monoxide poisoning, and herpes simplex encephalitis. Called also [dysmnestic s.](#)

amniotic band syndrome, early rupture of the amnion with formation of fibrous strands of amnion that may adhere to or compress parts of the fetus, resulting in a wide variety of abnormalities, including craniofacial defects, limb distortions, amputation, and abdominal or thoracic evisceration. Called also [amniotic band sequence](#), [amniotic band disruption complex](#), and [constriction band s.](#)

amniotic infection syndrome of Blane, a syndrome in which fetal sepsis follows swallowing and at times aspiration of contaminated amniotic fluid.

amyostatic syndrome, [Wilson disease](#).

Andersen syndrome, a syndrome consisting of [bronchiectasis](#) with [cystic fibrosis](#) and vitamin A deficiency. [Andersen-Tawil s.](#)

Andersen-Tawil syndrome, an autosomal dominant form of long QT syndrome characterized by periodic paralysis, ventricular arrhythmias, and dysmorphism of facial and skeletal bones. Called also [Andersen s.](#)

androgen insensitivity syndrome, resistance of target organs in males to the action of [androgens](#); the result is any of a spectrum from normal-appearing male phenotypes to female phenotype; see [complete androgen insensitivity s.](#), [partial androgen insensitivity s.](#), and [mild androgen insensitivity s.](#)

Angelman syndrome, an autosomal recessive syndrome characterized by jerky puppetlike movements, frequent laughter, mental and motor retardation, peculiar open-mouthed facies, and seizures. It is caused by loss of expression of genes on maternally derived chromosome 15q11-13, usually as a result of a deletion; the same loss of gene function inherited from the father causes [Prader-Willi syndrome](#).

Angelucci syndrome, excitable temperament, palpitation, and vasomotor disturbance in patients with vernal conjunctivitis.

angry back syndrome, [excited skin s.](#)

angular gyrus syndrome, a syndrome resulting from an infarction or other lesion of the angular gyrus on the dominant side; symptoms may include alexia or agraphia or may feature the symptoms of [Gerstmann syndrome](#).

ankyloblepharon-ectodermal dysplasia-clefting syndrome, [Hay-Wells s.](#)

anorexia-cachexia syndrome, a systemic response to conditions such as cancer or the [acquired immunodeficiency syndrome](#), resulting from a poorly understood relationship between [anorexia](#) and [cachexia](#), manifested by malnutrition, weight loss, muscular weakness, acidosis, and toxemia. The anorexia may be caused by a severe metabolic disturbance that contributes to development of cachectic wasting, which in turn reinforces anorexia by release from a tumor of a humoral product that stimulates the [satiety center](#) in the hypothalamus.

anterior chamber cleavage syndrome, a term for several types of mesenchymal dysgenesis affecting neural crest derivatives in the iris, trabecula, and cornea. In ascending severity these disorders are: [Axenfeld anomaly](#), [Axenfeld syndrome](#), [Rieger anomaly](#), and [Rieger syndrome](#).

anterior cord syndrome, [anterior spinal artery s.](#)

anterior cornual syndrome, muscular atrophy due to lesions of the anterior horns of the spinal cord. Cf. [spinal muscular atrophy](#).

anterior interosseous syndrome, **anterior interosseous nerve syndrome**, a complex of symptoms caused by a lesion of the anterior interosseous nerve, resulting usually from fracture or laceration but sometimes from an entrapment neuropathy; symptoms include pain in the proximal forearm and weakness of the muscles innervated by the nerve. Called also Kiloh-Nevin s.

anterior opercular syndrome, **anterior operculum syndrome**, intermittent or persistent loss of voluntary movements of the muscles of the lower face, throat, jaw, and tongue, characterized by dysarthria, sialorrhoea, and dysphagia, and caused by bilateral lesions of the frontoparietal operculum. It most commonly results from sequential bilateral opercular strokes; other causes include congenital anomalies, epilepsy, and infection. Called also [Foix-Chavany-Marie s.](#)

anterior scalene syndrome, [scalenus anterior s.](#)

anterior spinal artery syndrome, injury to the ventral spinal cord caused by blockage of the anterior spinal artery and infarction of the areas it supplies. Below the level of the lesion complete paralysis, hypalgesia, and hypesthesia occur but there is relative preservation of the posterior sensations of touch, position, and vibration.

anterior tibial compartment syndrome, rapid swelling, increased tension, pain, and ischemic necrosis of the muscles of the anterior tibial compartment of the leg; the skin becomes glossy, erythematous, and edematous as the necrosis occurs. The cause is unknown, but usually there is a history of excessive exertion.

anticholinergic syndrome, the central and peripheral effects produced by overdosage or abnormal reaction to clinical dosage of anticholinergic drugs, e.g., atropine, phenothiazines, antihistamines, and tricyclic antidepressants; signs and symptoms include anxiety, delirium, disorientation, hallucinations, seizures, tachycardia, hyperpyrexia, mydriasis, vasodilation, gastric and urinary retention, and decreased salivary, sweat, bronchial, and nasopharyngeal secretions.

anticonvulsant hypersensitivity syndrome, a life-threatening, multiorgan [hypersensitivity reaction](#) to [anticonvulsant](#) medications, especially [phenytoin](#); manifestations include rash, [arthralgia](#), [eosinophilia](#), [fever](#), [lymphadenopathy](#), and liver dysfunction.

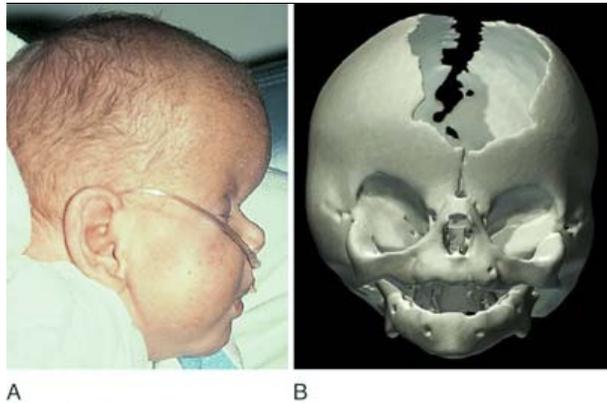
antiphospholipid syndrome, antiphospholipid antibody syndrome, a multisystem inflammatory disorder characterized by the presence of circulating [antiphospholipid antibodies](#) with thrombosis (including [thrombotic microangiopathy](#)), [spontaneous abortion](#), [thrombocytopenia](#), valvular heart disease, and other less frequent symptoms. A severe type called catastrophic antiphospholipid syndrome is characterized by infarctions of several different organs and is often fatal.

Anton syndrome, Anton-Babinski syndrome, a form of [anosognosia](#) in which the patient denies, and often is unaware of, the existence of clinically demonstrable blindness and may resort to confabulation to hide it; it may be the result of [denial](#) (q.v.) or of bilateral infarctions of the occipital lobes.

anxiety syndrome, the physical symptoms accompanying anxiety, such as palpitation of the heart, rapid and shallow respiration, sweating, pallor, and a feeling of panic.

aortic arch syndrome, any of a group of disorders leading to occlusion of the arteries arising from the aortic arch; causes include atherosclerosis, arterial embolism, syphilitic or tuberculous arteritis, and other conditions. See also [Takayasu arteritis](#), under arteritis.

Apert syndrome, an autosomal dominant disorder characterized by oxycephaly and syndactyly, often with other skeletal deformities and usually with mental retardation. Called also [acrocephalosyndactyly, type I](#) or, occasionally, [acrocephalosyndactyly](#).



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Apert syndrome in clinical view (A) and CT construction of the craniofacial skeleton (B).

syndrome of apparent mineralocorticoid excess, apparent mineralocorticoid excess.

ARN syndrome, acute retinal necrosis.

Arnold-Chiari syndrome, see under [malformation](#).

Arnold nerve reflex cough syndrome, a reflex cough due to irritation of the area supplied by Arnold nerve (the auricular branch of the vagus nerve); this area is the posterior and inferior portion of the external auditory canal and the posterior half of the tympanic membrane.

arthropathy-camptodactyly syndrome, a rare autosomal recessive disorder characterized by arthropathy associated with congenital flexion contractures of the fingers and synovial and tendon abnormalities, and by constrictive pericarditis.

Ascher syndrome, blepharochalasis occurring with goiter (adenoma of the thyroid) and redundancy of the mucous membrane and submucous tissue of the upper lip.

Asherman syndrome, persistent [amenorrhea](#) and secondary [sterility](#) due to intrauterine adhesions and synechiae, usually as a result of uterine [curettage](#).

Asherson syndrome, a syndrome of [dysphagia](#) due to neuromuscular incoordination and achalasia of the [cricopharyngeus muscle](#) during the third stage of [swallowing](#). It causes diversion of liquids into the air passages, precipitating paroxysms of coughing. Called also [cricopharyngeal achalasia s.](#)

Asperger syndrome, a pervasive developmental disorder resembling autistic disorder, being characterized by severe impairment of social interactions and by restricted interests and behaviors, but lacking the delays in development of language, cognitive function, and self-help skills that additionally define autistic disorder. It may be equivalent to a high-functioning form of autistic disorder.

asplenia syndrome, Ivemark s.

ataxia-telangiectasia syndrome, ataxia-telangiectasia; see under ataxia.

atypical mole syndrome, atypical nevus syndrome, dysplastic nevus s.

auriculotemporal syndrome, redness and sweating on the cheek in connection with eating; many persons have mild cases, especially after eating spicy or sharp-tasting foods. Some individuals have more severe forms, such as after surgery or other damage to the [parotid gland](#) or [auriculotemporal nerve](#), or in disease states such as [syringomyelia](#) or some types of [encephalitis](#). Called also [Frey s.](#), [gustatory hyperhidrosis](#) or [sweating](#), and [gustatory sweating s.](#)

autoerythrocyte sensitization syndrome, painful bruising s.

autoimmune lymphoproliferative syndrome, a hereditary disorder of lymphocyte apoptosis that results in the accumulation of large numbers of mature lymphocytes in the lymph nodes and spleen, appearing during childhood and characterized by massive lymphadenopathy, splenomegaly, and autoimmune hemolytic anemia and other cytopenias.

autoimmune polyendocrine syndromes, autoimmune polyglandular syndromes, polyglandular autoimmune s's.

Avellis syndrome, a syndrome in which a brainstem lesion limits vagal innervation unilaterally, resulting in ipsilateral paralysis of the vocal cord and soft palate and loss of sensitivity to pain and temperature in the contralateral leg, trunk, arm, and neck, and in the skin over the scalp; called also [Avellis paralysis](#).

Axenfeld syndrome, Axenfeld anomaly accompanied by glaucoma and defective development of the corneoscleral trabecular meshwork and other angle structures. See also [anterior chamber cleavage s.](#)

Ayerza syndrome, pulmonary hypertension with dilatation of the pulmonary arteries, related to disease of the lungs; cf. [plexogenic pulmonary arteriopathy](#).

Baastrup syndrome, kissing spines.

Babinski syndrome, the association of cardiac and arterial disorders with chronic syphilitic meningitis, tabes dorsalis, paralytic dementia, and other late syphilitic manifestations.

Babinski-Fröhlich syndrome, adiposogenital dystrophy.

Babinski-Nageotte syndrome, a syndrome due to multiple lesions affecting the medullary pyramid and sensory tracts, the cerebellar peduncle, and the reticular formation, and marked by contralateral hemiplegia and hemianesthesia (usually only of the pain and temperature senses), ipsilateral hemiasynergia, hemiataxia, and Horner syndrome.

Babinski-Vaquez syndrome, [Babinski s.](#)

bacterial overgrowth syndrome, [stasis s.](#)

BADS syndrome, a rare congenital syndrome of unknown heritability characterized by black locks, oculocutaneous albinism, and deafness of the sensorineural type; see [oculocutaneous albinism](#).

Balint syndrome, [gaze paralysis](#), [ataxia](#) of eye movements, and other disturbances of visual attention, with preservation of spontaneous and reflex eye movements, usually caused by bilateral lesions in the parietooccipital region.

Baller-Gerold syndrome, an autosomal recessive syndrome characterized by craniosynostosis and radial aplasia. Called also [craniosynostosis-radial aplasia s.](#)

ballooning mitral valve syndrome, ballooning posterior leaflet syndrome, [mitral valve prolapse s.](#)

Bannayan-Zonana syndrome, a rare autosomal dominant syndrome characterized by hemangiomas of the trunk, cutaneous lipomas, macrocephaly, and swelling of the abdomen with angiomas.

Bannwarth syndrome, the European term for the [meningopolyneuritis](#) that may occur in [Lyme disease](#).

Bardet-Biedl syndrome, an autosomal recessive disorder characterized by mental retardation, pigmentary retinopathy, obesity, polydactyly, and hypogonadism; cf. [Laurence-Moon s.](#) and [Biemond s., II.](#)

Barlow syndrome, [mitral valve prolapse s.](#)

Barraquer-Simons syndrome, [partial lipodystrophy](#).

Barrett syndrome, [peptic ulcer](#) of the lower esophagus, often with stricture, due to the presence of columnar-lined epithelium in the esophagus (sometimes containing functional mucous cells, parietal cells, or chief cells) instead of the normal squamous cell epithelium. It is sometimes premalignant, followed by esophageal adenocarcinoma. Called also [Barrett esophagus](#).

Bart syndrome, a form of [epidermolysis bullosa dystrophica](#) inherited as an autosomal dominant trait, characterized by congenital localized absence of the skin, blister formation resulting from mechanical trauma, and nail dystrophy.

Barth syndrome, an X-linked disorder caused by mutation in the TAZ gene (locus: q28), which encodes a protein of unknown function that is highly expressed in cardiac and skeletal muscle, characterized by dilated cardiomyopathy, hypotonia, neutropenia, and growth retardation, with moderately increased urinary excretion of 3-methylglutaconic and 3-methylglutaric acids. Called also [3-methylglutaconicaciduria type II](#).

Barter syndrome, hypertrophy and hyperplasia of the [juxtaglomerular cells](#), producing [hypokalemic alkalosis](#) and [hyperaldosteronism](#), characterized by absence of hypertension in spite of markedly increased plasma [renin](#) concentrations, and by insensitivity to the pressor effects of [angiotensin](#). It usually affects children, may be hereditary, and may be associated with other anomalies such as mental retardation and short stature. See also [Gitelman s.](#) Called also [juxtaglomerular cell hyperplasia](#).

basal cell nevus syndrome, [nevoid basal cell carcinoma s.](#)

basilar artery syndrome, [vertebrobasilar insufficiency](#).

BASM syndrome, [biliary atresia splenic malformation s.](#)

Bassen-Kornzweig syndrome, [abetalipoproteinemia](#).

battered child syndrome, unexplained or inappropriately explained physical trauma and other manifestations of severe, repeated physical abuse of children, usually by a parent or other caretaker.

Bazex syndrome, eczematous and psoriasiform lesions on the ears, nose, cheeks, hands, feet, and knees in patients with carcinomas of the upper respiratory and digestive tracts. Called also [paraneoplastic acrokeratosis](#).

Beals syndrome, [congenital contractural arachnodactyly](#).

Bean syndrome, [blue rubber bleb nevus s.](#)

Bearn-Kunkel syndrome, Bearn-Kunkel-Slater syndrome, [chronic active hepatitis](#).

Beckwith syndrome, Beckwith-Wiedemann syndrome, a congenital autosomal dominant syndrome with variable expressivity characterized by exomphalos, macroglossia, and gigantism, often associated with [organomegaly](#), adrenocortical [cytomegaly](#), and dysplasia of the renal medulla. Called also [EMG s.](#) and [exomphalos-macroglossia-gigantism s.](#)

Behçet syndrome, a variant of [neutrophilic dermatosis](#) of unknown etiology, involving the small blood vessels, characterized by recurrent aphthous ulceration of oral and pharyngeal mucous membranes and genitalia, with skin lesions, severe uveitis, retinal vasculitis, optic atrophy, and often involvement of the joints, gastrointestinal system, and central nervous system.

Benedikt syndrome, a syndrome consisting of ipsilateral oculomotor paralysis, contralateral hyperkinesia, contralateral tremor and paresis of the arm and leg, and ipsilateral ataxia; caused by lesions that damage the third nerve and involve the nucleus ruber and corticospinal tract. Called also [tegmental mesencephalic paralysis](#) and [tegmental s.](#)

benign hypermobility syndrome, benign joint hypermobility syndrome, Ehlers-Danlos syndrome, type III, inherited as an autosomal dominant trait and characterized by [hypermobility](#) of the joints with minimal abnormalities of the skin.

Berardinelli-Seip syndrome, [total lipodystrophy](#).

Berdon syndrome, [megacystis-microcolon-intestinal hypoperistalsis s.](#)

Bernard syndrome, Bernard-Horner syndrome, Horner s.

Bernard-Sergent syndrome, [addisonian crisis](#).

Bernard-Soulier syndrome, an autosomal recessive disorder characterized by giant platelets with membranes lacking glycoprotein Ib, the probable receptor for plasma [von Willebrand factor](#); this keeps the platelets from binding the factor, which is necessary for their adhesion to the subendothelial surfaces of blood vessels. Symptoms include mild to moderate mucocutaneous and visceral hemorrhaging, purpura, and prolonged bleeding time. Called also [giant platelet s.](#) See also [thrombasthenia](#) and [von Willebrand disease](#).

Bernheim syndrome, [right ventricular heart failure](#) due to left [ventricular hypertrophy](#) with bulging of the [interventricular septum](#) that causes obstruction to flow from the [right atrium](#) to [ventricle](#), altering ventricular filling and capacity.

Bertolotti syndrome, sacralization of the fifth lumbar vertebra together with sciatica and scoliosis.

bi syndrome, in traditional Chinese medicine, a disease with pain caused by an obstruction in the body.

Biemond syndrome, II, an autosomal recessive disorder characterized by iris coloboma, obesity, mental retardation, hypogonadism, and postaxial polydactyly; cf. [Bardet-Biedl s.](#) and [Laurence-Moon s.](#)

biliary atresia splenic malformation syndrome, a rare congenital condition of biliary atresia together with polysplenia or some other anomaly of the spleen. It may be associated with other anomalies, including defects of the portal vein, heart, lungs, and liver. Called also [BASM s.](#)

billowing mitral valve syndrome, billowing posterior leaflet syndrome, [mitral valve prolapse s.](#)

Bing-Neel syndrome, the central nervous system manifestations of Waldenström macroglobulinemia; symptoms may include encephalopathy, hemorrhage, stroke, convulsions, delirium, and coma.

Birt-Hogg-Dube syndrome, an autosomal dominant disorder of proliferation of ectodermal and mesodermal components, caused by a mutation on chromosome 17p and characterized by multiple [acrochordons](#), [trichodiscomas](#), and [fibrofolliculomas](#) on the head, chest, back, and arms; kidney tumors; spontaneous [pneumothorax](#); and [intestinal polyposis](#).

Björnstad syndrome, an autosomal recessive disorder characterized by congenital sensorineural deafness and pili torti.

Blackfan-Diamond syndrome, [congenital hypoplastic anemia](#) (def. 1).

Blau syndrome, a rare, autosomal dominant type of [autoinflammatory disease](#) (q.v.) characterized by granulomatous arthritis, skin rash, and ocular symptoms that may include iritis or anterior uveitis.

blind loop syndrome, [stasis s.](#)

Bloch-Sulzberger syndrome, [incontinentia pigmenti](#).

Bloom syndrome, an autosomal recessive syndrome consisting of [erythema](#) and [telangiectasia](#) in a butterfly distribution on the face, [photosensitivity](#), and well-proportioned dwarfism of prenatal onset. Immunoglobulins show abnormalities and there is a high incidence of malignancy, especially leukemia. Many patients are of Ashkenazi Jewish ancestry, due to a founder effect. The genome is unusually unstable in somatic cells, with an excess of [sister chromatid exchange](#), and is characterized by chromosome abnormalities including breaks, gaps, rearrangements, and other mutations; it is caused by mutations in a gene (BLM) that encodes a specific DNA [helicase](#).

blue diaper syndrome, a defect of [tryptophan](#) absorption in which, because of intestinal bacterial action on the [tryptophan](#), the urine contains abnormal [indoles](#), giving it a blue color. It is similar to [Hartnup disease](#) and is thought to have autosomal recessive inheritance.

blue rubber bleb nevus syndrome, a rare syndrome, sometimes autosomal dominant, of multiple [blue rubber bleb nevi](#) associated with hemangiomas of the gastrointestinal tract, which bleed readily and cause chronic [iron deficiency anemia](#); most cases present in infancy or childhood, but some do not appear until later. Called also [Bean s.](#)

blue toe syndrome, a blue color of the toes, sometimes bilateral, with skin necrosis and ischemic gangrene, resulting from arterial occlusion by emboli, thrombi, or injury.

body of Luys syndrome, [hemiballismus](#).

Boerhaave syndrome, spontaneous rupture of the esophagus.

Bonnet-Dechaume-Blanc syndrome, [Wyburn-Mason s.](#)

Böök syndrome, [PHC s.](#)

BOR syndrome, [branchio-oto-renal s.](#)

Börjeson syndrome, **Börjeson-Forsman-Lehmann syndrome**, an X-linked syndrome characterized by severe mental retardation, epilepsy, hypogonadism, hypometabolism, marked obesity, swelling of the subcutaneous tissues of the face, and large ears.

Bouillaud syndrome, pericarditis and endocarditis accompanying rheumatic fever.

Bourneville-Pringle syndrome, [tuberous sclerosis complex](#).

Bouveret syndrome, [paroxysmal supraventricular tachycardia](#). obstruction of the gastric outlet by a gallstone passed into the duodenal bulb through a cholecystoduodenal or choledochoduodenal fistula.

bowel bypass syndrome, a type of [neutrophilic dermatosis](#) with [arthritis](#) that may occur one to six years after [jejunoileal bypass](#), characterized by rash, malaise, [myalgia](#), joint pain, sterile skin pustules, and a flulike illness; it is probably caused by circulating [immune complexes](#) that include bacterial antigens resulting from overgrowth in the bypassed part of the intestine.

brachial syndrome, see under [plexopathy](#).

Brachmann-de Lange syndrome, [de Lange s.](#)

Bradbury-Eggleston syndrome, a syndrome of dysautonomia with orthostatic hypotension, visual disturbances, hypohidrosis, impotence, lowered basal metabolic rate, dizziness, syncope, presyncope, and a slow, unchanging pulse. It occurs predominantly in older males in the early morning hours during the summer and is due to impaired peripheral vasoconstriction; it usually has a progressive course. Called also [primary](#) or [pure autonomic failure](#).

bradycardia-tachycardia syndrome, **brady-tachy syndrome**, a clinical manifestation of the sick sinus syndrome characterized by periods of atrial and ventricular bradycardia alternating with periods of rapid regular or irregular atrial or ventricular tachyarrhythmias.

branchio-oto-renal syndrome, [branchial arch](#) anomalies (preauricular pits, branchial fistulas or pits) associated with [Mondini deafness](#) and renal dysplasia, inherited as an autosomal dominant trait with high penetrance and variable expression. Called also [BOR s.](#) and [Melnick-Fraser s.](#)

Brennemann syndrome, mesenteric and retroperitoneal lymphadenitis as a sequel of throat infections.

Briquet syndrome, [somatization disorder](#).

Brissaud-Sicard syndrome, spasmodic hemiplegia caused by lesions of the pons.

Bristowe syndrome, a series of symptoms caused by a tumor of the corpus callosum, including hemiplegia and apraxia.

brittle cornea syndrome, a rare autosomal recessive syndrome characterized by brittle corneas, blue sclerae, skin hyperelasticity, and joint hypermobility.

Brock syndrome, [middle lobe s.](#)

bronze baby syndrome, brown discoloration of the skin in an infant undergoing phototherapy for neonatal hyperbilirubinemia, caused by failure to excrete bilirubin products properly.

Brooke-Spiegler syndrome, an autosomal dominant syndrome (gene locus: 16q12-q13) characterized by multiple skin tumors such as cylindromas and trichoepitheliomas, usually in the head and neck region; some authorities consider this and multiple familial trichoepithelioma to be variants of the same condition. Called also [familial cylindromatosis](#).

Brooks-Wisniewski-Brown syndrome, an X-linked syndrome of characteristic facies, [static encephalopathy](#), mental and growth retardation, ocular abnormalities, [spastic diplegia](#), and behavioral problems; a mitochondrial defect has been suggested as the cause.

Brown vertical retraction syndrome, adhesion of the muscles of the eye in the fetus.

Brown-Séquard syndrome, a syndrome due to damage of one half of the spinal cord, resulting in ipsilateral paralysis and loss of discriminatory and joint sensation, and contralateral loss of pain and temperature sensation. Called also [Brown-Séquard paralysis](#) or [sign](#).

Brown-Vialetto-van Laere syndrome, an autosomal recessive syndrome consisting of progressive bulbar palsy with any of several cranial nerve disorders, including nerve deafness, facial weakness, dysarthria, and dysphagia.

Brueghel syndrome, [Meige s.](#)

Brugada syndrome, a type of [ion channelopathy](#) characterized by sudden, idiopathic [ventricular fibrillation](#) in an apparently healthy person, often resulting in death ([sudden adult](#) or [sudden arrhythmia death syndrome](#)). It is autosomal dominant and occurs most often in males of Southeast Asian origin.

Bruns syndrome, intermittent headache, vertigo, vomiting, and visual disturbances on sudden movement of the head, characteristic of cysticercus infection of the fourth ventricle, lesion of the fourth ventricle, or tumors of the midline of the cerebellum and third or lateral ventricles. Called also [Bruns sign](#).

Bruns-Garland syndrome, [diabetic amyotrophy](#).

Brunsting syndrome, **Brunsting-Perry syndrome**, a variant of [cicatrical pemphigoid](#) with lesions on the skin of the head and neck that heal and leave superficial scarring; if it is on the scalp, it causes [cicatrical alopecia](#). Called also [Brunsting-Perry pemphigoid](#).

Brushfield-Wyatt syndrome, a congenital syndrome consisting of extensive unilateral nevus flammeus, [homonymous hemianopia](#) of both eyes, contralateral hemiplegia, cerebral angioma, and mental retardation; it may be related to the [Sturge-Weber syndrome](#).

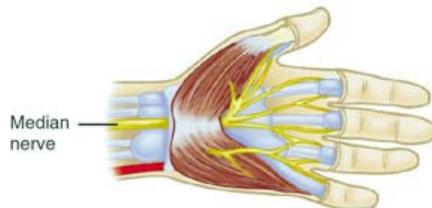
Buckley syndrome, [hyperimmunoglobulinemia E s.](#)

Budd-Chiari syndrome, symptomatic obstruction or occlusion of the hepatic veins, causing [hepatomegaly](#), abdominal pain and tenderness, intractable ascites, mild [jaundice](#), and, eventually, [portal hypertension](#) and liver failure. The obstruction is caused by thrombi or fibrous obliteration of the veins and has been associated with coagulation disorders; [myeloproliferative disorders](#); invasion of hepatic veins by hepatic, renal, or adrenal carcinoma; and abdominal trauma. Onset may be acute with death in a few days in cases of complete occlusion, but more often there is a chronic course with survival for months or years. Called also [Chiari s.](#) and [endophlebitis hepatica obliterans](#). Cf. [veno-occlusive disease of liver](#).

bulbar syndrome, any syndrome caused by a lesion in the medulla and its nuclei, with paralysis of the cranial nerves originating there; cf. [progressive bulbar palsy](#). Called also [Dejerine s.](#)

Bürger-Grütz syndrome, familial hyperlipoproteinemia, type I.

Burnett syndrome, [milk-alkali s.](#)
burning feet syndrome, [Gopalan s.](#)
burning mouth syndrome, any of various conditions of burning sensations and pain in the mouth ([stomatalgia](#) or [glossalgia](#)) having unknown etiologies; they occur most often in middle-aged, perimenopausal women or persons with [candidiasis](#) who have used antibiotics for prolonged periods.
Buschke-Ollendorff syndrome, an autosomal dominant syndrome, present at birth or appearing before puberty, characterized by often symmetric [connective tissue nevi](#) of the elastic type on the limbs and lower trunk ([dermatofibrosis lenticularis disseminata](#)) in association with [osteopoikilosis](#).
Butler-Albright syndrome, a type of [distal renal tubular acidosis](#) occurring later than infancy and having autosomal dominant inheritance.
Bywaters syndrome, [crush s.](#)
CADASIL syndrome, a rare, autosomal dominant, adult-onset vascular condition affecting mainly the centrum semiovale of the cerebral hemispheres; characteristics include migraines and other neurologic symptoms, stroke and related conditions, and eventually [subcortical dementia](#) and death.
Caffey syndrome, **Caffey-Silverman syndrome**, [infantile cortical hyperostosis](#).
camptomelic syndrome, osteochondrodysplasia associated with flat facies, bowed tibiae with skin dimpling, hypoplastic scapulae, and short vertebrae.
Canada-Cronkhite syndrome, [Cronkhite-Canada s.](#)
Canale-Smith syndrome, [autoimmune lymphoproliferative s.](#)
cancer family syndrome, type 2 [hereditary nonpolyposis colorectal cancer](#).
Capgras syndrome, a form of delusional misidentification in which the patient believes that other persons in the environment are not their real selves but doubles. Cf. [Frégoli phenomenon](#).
capillary leak syndrome, extravasation of plasma fluid and proteins into the extravascular space, resulting in sometimes fatal hypotension and reduced organ perfusion; an adverse effect of aldesleukin (interleukin-2) therapy.
Caplan syndrome, pneumoconiosis associated with rheumatoid arthritis. Radiographically, multiple spherical nodular lesions with clearly demarcated borders are found throughout both lungs. Called also [rheumatoid pneumoconiosis](#).
carcinoid syndrome, a symptom complex associated with carcinoid tumors, characterized by attacks of severe cyanotic flushing of the skin and by watery diarrhea, bronchoconstrictive attacks, lesions of the heart valves, edema, ascites, and increased urinary excretion of 5-hydroxyindoleacetic acid. Symptoms are caused by secretion by the tumor of [serotonin](#), [prostaglandins](#), and other biologically active substances.
cardiac syndrome X, a relatively benign syndrome of [angina pectoris](#) or anginalike chest pain associated with normal arteriographic appearance of the coronary arteries. Called also [s. X](#).
Carney syndrome, see under [complex](#).
carotid sinus syndrome, syncope sometimes associated with convulsive seizures due to overactivity of the [carotid sinus reflex](#) (q.v.) when pressure is applied to one or both carotid sinuses. Called also [carotid sinus syncope](#) and [Charcot-Weiss-Baker s.](#)
carpal tunnel syndrome, an entrapment neuropathy characterized by pain and burning or tingling paresthesias in the fingers and hand, sometimes extending to the elbow. Symptoms result from compression of the median nerve in the carpal tunnel.



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 Median nerve entrapped in carpal tunnel in carpal tunnel syndrome.

Carpenter syndrome, an autosomal recessive disorder characterized by acrocephaly with polysyndactyly, brachydactyly, mental retardation, mild obesity, hypogonadism, and other anomalies. It is believed to encompass disorders formerly distinguished as [Goodman syndrome](#) (q.v.). Called also [acrocephalopolysyndactyly, type II](#).
cast syndrome, superior mesenteric artery syndrome caused by enclosure in a body cast.
cat-eye syndrome, an association of coloboma of the iris and anal atresia; there may also be many other anomalies, including preauricular skin tags or fistulas, hypertelorism, congenital heart disease, skeletal abnormalities, and renal malformations. It is associated with partial trisomy 22, i.e., the presence of a partial additional copy of chromosome 22.
cat's cry syndrome, [cri du chat s.](#)
cat's eye syndrome, [cat-eye s.](#)
cauda equina syndrome, a group of symptoms caused by compression of the spinal nerve roots, including dull, aching pain of the perineum, bladder, and sacrum that generally radiates in a sciatic fashion and is associated with paresthesias and areflexic paralysis. see under [neuritis](#).
caudal dysplasia syndrome, **caudal regression syndrome**, failure of formation of part or all of the coccygeal, sacral, and occasionally lumbar vertebral units and the corresponding segments of the caudal spinal cord, with resulting neurogenic dysfunction of bowel and bladder; called also [sacral agenesis](#).
cavernous sinus syndrome, edema of the conjunctiva, proptosis, edema of upper lid and root of the nose, together with paralysis of the third, fourth, and sixth cranial nerves and the ophthalmic branch of the fifth; it is usually due to thrombosis or tumor of the cavernous sinus. Cf. [Tolosa-Hunt s.](#) Called also [Foix s.](#)
celiac artery compression syndrome, **celiac axis compression syndrome**, compression of the celiac axis or trunk by crura of the diaphragm, which can interfere with the blood supply to the liver and spleen, resulting in intermittent postprandial pain in the upper abdomen.
central alveolar hypoventilation syndrome, [primary alveolar hypoventilation](#).
central cord syndrome, a syndrome associated with injury to the cervical or upper thoracic portions of the spinal cord that results in damage to the central cord with sparing of the more external fibers that supply the lower extremity. It is characterized by disproportionately more weakness or paralysis in the upper extremity than in the lower, with varying sensory deficits below the level of the lesion, sometimes with bladder and bowel dysfunction.
central sleep apnea syndrome, see under [apnea](#).
centroposterior syndrome, [syringomyelia](#).
cerebellar syndrome, see under [ataxia](#).
cerebellopontine angle syndrome, a syndrome caused by a tumor of the cerebellopontine angle or an acoustic tumor, characterized by hearing loss, subjective noises, ipsilateral cerebellar ataxia, and eventually ipsilateral impairment of function of the sixth and seventh cranial nerves accompanied by elevated intracranial pressure.
cerebral hyperperfusion syndrome, [hyperperfusion s.](#)

cerebrocardiac syndrome, [Krishaber disease](#).

cerebrocostomandibular syndrome, an autosomal recessive syndrome of severe micrognathia and costovertebral abnormalities, including small bell-shaped thorax, incompletely ossified aberrant rib structure, and abnormal rib attachment to vertebrae. Also present are palatal defects, glossoptosis, prenatal and postnatal growth deficiencies, and mental retardation, the last perhaps due to the neonatal respiratory distress which is frequently the presenting sign of the disorder.

cerebrohepatorenal syndrome, an autosomal recessive disorder characterized by craniofacial abnormalities, hypotonia, hepatomegaly, polycystic kidneys, jaundice, and death in early infancy, and associated with absence of peroxisomes in the liver and kidneys; called also [Zellweger s.](#)

cervical syndrome, **cervical disk syndrome**, a condition caused by irritation or compression of the cervical nerve roots by a protruding disk; symptoms include neck pain radiating into the shoulder, arm, or forearm, paresthesias, and muscle weakness or spasm.

cervical rib syndrome, a [thoracic outlet syndrome](#) caused by a cervical rib.

cervicobrachial syndrome, [brachial plexopathy](#).

Cestan syndrome, **Cestan-Chenais syndrome**, an association of [contralateral hemiplegia](#) and [hemianesthesia](#), ipsilateral [lateropulsion](#) and [hemiasynergia](#), [Horner syndrome](#), and ipsilateral [laryngoplegia](#), due to scattered lesions of the [pyramid](#), [sensory tract](#), [inferior cerebellar peduncle](#), and [nucleus ambiguus](#).

Cestan-Raymond syndrome, [Raymond-Cestan s.](#)

Charcot syndrome, [amyotrophic lateral sclerosis](#), [intermittent claudication](#).

Charcot-Marie syndrome, [Charcot-Marie-Tooth disease](#).

Charcot-Weiss-Baker syndrome, [carotid sinus s.](#)

CHARGE syndrome, a syndrome of associated defects caused by mutations in either of two genes that are important in embryonic development: CHD7 (locus: 8q12.1) and SEMA3E (locus: 7q21.11). Defects include coloboma of the eye, heart anomaly, choanal atresia, retardation, and genital and ear anomalies. Facial palsy, cleft palate, and dysphagia are often present. Most cases are sporadic but some show a familial pattern.

Charlin syndrome, pain, iritis, corneitis, rhinorrhea, and tenderness along the nose as a result of neuralgia of the nasociliary nerve. Called also [nasociliary neuralgia](#).

Chédiak-Higashi syndrome, a lethal autosomal recessive syndrome associated with oculocutaneous albinism, massive leukocyte inclusions (giant lysosomes), histiocytic infiltration of multiple body organs, development of pancytopenia, hepatosplenomegaly, recurrent or persistent bacterial infections, and a possible predisposition to development of malignant lymphoma. Called also [Béguet César disease](#) and [Chédiak-Higashi anomaly](#).

Chiari syndrome, [Budd-Chiari s.](#)

Chiari-Arnold syndrome, [Arnold-Chiari malformation](#).

Chiari-Frommel syndrome, [galactorrhea-amenorrhea syndrome](#) occurring after pregnancy; called also [Frommel-Chiari s.](#), [Chiari-Frommel disease](#), and [Frommel disease](#).

chiasma syndrome, **chiasmatic syndrome**, a syndrome indicative of lesions affecting the optic chiasma: impairment of vision, limitations of the field of vision, central scotoma, headache, vertigo, and syncope.

Chilaiditi syndrome, interposition of the colon between the liver and diaphragm. Usually the condition is asymptomatic in adults, but symptoms are evident in children and include vomiting, abdominal pain, anorexia, constipation, and aerophagia. Signs include abdominal distention and absence of liver dullness.

CHILD syndrome, a disorder of skin cornification characterized by unilateral erythema, scaling, and [epidermal nevi](#); ipsilateral limb defects; and sometimes skeletal hypoplasia, brain defects, and visceral defects. It first appears in infancy and is believed to be an X-linked dominant trait.

Chinese restaurant syndrome, a transient syndrome associated with arterial dilatation, due to ingestion of [monosodium glutamate](#), which is sometimes used liberally in seasoning Chinese food; it is characterized by throbbing of the head, lightheadedness, tightness of the jaw, neck, and shoulders, and backache.

Chotzen syndrome, an autosomal dominant disorder characterized by acrocephalosyndactyly in which the syndactyly is mild and by hypertelorism, ptosis, and sometimes mental retardation. Called also [acrocephalosyndactyly, type III](#) and [Saethre-Chotzen s.](#)

Christ-Siemens-Touraine syndrome, [anhidrotic ectodermal dysplasia](#).

Christian syndrome, [Hand-Schüller-Christian disease](#).

chronic fatigue syndrome, persistent debilitating fatigue lasting longer than six months, with other known medical conditions having been ruled out by clinical diagnosis, accompanied by at least four of the following: significantly impaired short-term memory or concentration, muscle weakness, pain in multiple joints without swelling or redness, sore throat, tender lymph nodes, headaches, unrefreshing sleep, and malaise that lasts more than 24 hours following exertion. The cause is unknown and may be multifactorial; immune dysfunction has been suggested, and viral infection may be associated with it, although no causal relationship has been demonstrated. A number of names have been used for this syndrome, including [Iceland disease](#), [myalgic](#) or [benign myalgic encephalomyelitis](#), [chronic Epstein-Barr virus infection](#), [chronic mononucleosis](#), and [epidemic neuromyasthenia](#).

chronic infantile neurologic cutaneous and articular syndrome, [neonatal-onset multisystem inflammatory disease](#).

chronic pelvic pain syndrome, name given to nonspecific pain in pelvic organs or tissues, which has been continuous for at least six months and is not caused by malignancy, infection, or other obvious pathology.

Churg-Strauss syndrome, a type of small vessel vasculitis that is also grouped with the [systemic necrotizing vasculitides](#) and has prominent lung involvement with severe asthma, eosinophilia, and granulomatous reactions. If present, cutaneous lesions consist of tender subcutaneous nodules, large ecchymotic plaques, and cutaneous infarcts. There are several different types; one is a type of ANCA-associated vasculitis. Called also [allergic granulomatosis](#), [allergic granulomatous angiitis](#), and [Churg-Strauss vasculitis](#).

chylomicronemia syndrome, [familial hyperchylomicronemia](#).

CINCA syndrome, [neonatal-onset multisystem inflammatory disease](#).

Citelli syndrome, mental dullness, loss of power of concentration, and drowsiness or insomnia, seen in persons with adenoids or sinus infection.

Clarke-Hadfield syndrome, congenital pancreatic disease with infantilism; with enlarged liver, bulky fatty stools, and extensive atrophy of the pancreas in an undersized and underweight child.

Claude syndrome, paralysis of the third (oculomotor) nerve on one side and asynergia on the other side, together with dysarthria; called also [inferior s. of red nucleus](#) and [rubrospinal cerebellar peduncle s.](#)

Claude Bernard-Horner syndrome, [Horner s.](#)

click syndrome, **click-murmur syndrome**, [mitral valve prolapse s.](#)

closed head syndrome, the complex of symptoms characteristic of cerebral injury without cranial penetration. See also [concussion](#) and [postconcussional s.](#)

Clouston syndrome, [hidrotic ectodermal dysplasia](#).

cloverleaf skull syndrome, [kleeblattschädel s.](#)

Cockayne syndrome, a rare, pleiotropic, autosomal recessive disorder characterized by a variety of cutaneous, neurologic, and somatic abnormalities that vary in severity, including growth failure; delayed psychomotor development, progressive hearing loss, and other neurologic manifestations; pigmentary retinal degeneration and other progressive ocular abnormalities; cutaneous photosensitivity, and calcification of basal ganglia. It is caused by mutations affecting the [transcription-coupled repair](#) subpathway of [nucleotide excision repair](#).

Coffin-Lowry syndrome, a condition with onset in the postnatal period characterized by incapability of speech, severe mental deficiency, and muscle, ligament, and skeletal abnormalities; it is transmitted with X-linked intermediate inheritance.

Coffin-Siris syndrome, hypoplasia or absence of the nails of the fifth fingers and toes associated with growth and mental deficiencies, coarse facies, mild microcephaly, hypotonia, lax joints, mild hirsutism, and occasionally cardiac, vertebral, or gastrointestinal anomalies.

Cogan syndrome, nonsyphilitic interstitial keratitis with tinnitus and deafness; it usually occurs in children, often associated with polyarteritis nodosa. [Cogan oculomotor apraxia](#).

cold agglutinin syndrome, the presence of circulating [cold agglutinins](#), usually IgM, which are directed against three types of polysaccharide red cell antigens: [I antigens](#), expressed primarily on adult red cells, [i antigens](#), expressed primarily on cells of fetuses and infants, and [Pr antigens](#), which, unlike I and i antigens, are protease sensitive. The primary clinical manifestations are intravascular hemolysis in exposed extremities and mild hemolytic anemia due to complement fixation, both occurring only upon exposure to cold. There are two major types: chronic cold agglutinin disease, a condition seen in the elderly with gradual onset and a chronic course; and postinfectious cold agglutinin syndrome, which usually follows [Mycoplasma pneumoniae](#) infection or infectious mononucleosis and lasts a few months. The syndrome can also develop secondary to malignancy.

Collet syndrome, **Collet-Sicard syndrome**, [Vernet syndrome](#) with ipsilateral paralysis of the tongue, due to complete lesion of the ninth, tenth, eleventh, and twelfth cranial nerves. Called also [Sicard s.](#)

combination syndrome, a condition sometimes seen in patients with a mandibular [distal extension partial denture](#) opposing a complete maxillary denture; if there is not enough primary occlusal force, excessive load may be placed on the anterior part of the maxillary denture, causing loss of the anterior maxillary alveolus.

compartment syndrome, **compartmental syndrome**, a condition in which increased tissue pressure in a confined anatomical space causes decreased blood flow leading to ischemia and dysfunction of contained myoneural elements, marked by pain, muscle weakness, sensory loss, and palpable tenseness in the involved compartment. Ischemia can lead to necrosis resulting in permanent impairment of function.

complete androgen insensitivity syndrome, an extreme type of androgen insensitivity syndrome with [male pseudohermaphroditism](#), in which an individual is phenotypically female but has XY chromosomes; this results from mutations in the gene for the androgen receptor. There may be a rudimentary uterus and tubes, but the gonads are typically testes, which may be abdominal or inguinal in position. Called also [complete androgen resistance](#), [feminizing testes syndrome](#), [Morris syndrome](#), [testicular feminization](#), and [testicular feminization syndrome](#).

complex regional pain syndrome, a chronic pain syndrome of uncertain pathogenesis, usually affecting an extremity, and characterized by intense burning pain, changes in skin color and texture, increased skin temperature and sensitivity, sweating, and edema. Type 1 (called also [reflex sympathetic dystrophy](#)) often follows tissue injury, but without demonstrable nerve injury, and may be accompanied by [posttraumatic osteoporosis](#); when limited to the upper extremity it is called [shoulder-hand syndrome](#). Type 2 (called also [causalgia](#)) is associated with injury to the peripheral nerves.

compression syndrome, [crush s.](#)

concussion syndrome, [postconcussional s.](#)

congenital central hypoventilation syndrome, a congenital form of primary alveolar hypoventilation, generally resulting from a mutation in the short arm of chromosome 4 (4p12). Affected infants appear cyanotic, with symptoms of respiratory failure, within a few hours of birth, and hypoventilate during sleep but, unless the disease is severe, breathe normally when awake.

congenital high airway obstruction syndrome, a syndrome sometimes seen in utero, consisting of obstruction of the upper airway and dilation of the trachea and lungs; most fetuses with this condition die before birth.

congenital rubella syndrome, developmental anomalies resulting from [transplacental infection](#) of the fetus with [rubella](#), usually in the first trimester of pregnancy; maternal infection may be subclinical. The anomalies may include cardiac lesions, ocular lesions, deafness, microcephaly, mental retardation, and generalized growth retardation, sometimes associated with acute self-limited conditions such as thrombocytopenic purpura, anemia, hepatitis, encephalitis, and radiolucencies of long bones. Infected infants may shed virus to all contacts for an extended period. Called also [rubella s.](#)

congenital tremor syndrome, any of several congenital neurological diseases of piglets characterized by noticeable trembling, often owing to defective myelination of nerves. Mild varieties may clear up within a month but in severe cases the animals cannot function normally and soon die. One variety is autosomal recessive; another is sex-linked; and others are caused by intrauterine viral infections such as with hog cholera virus. Affected pigs are called [dancing](#) or [shaker pigs](#). Called also [congenital trembles](#) and [myoclonia congenita](#).

congenital varicella syndrome, developmental anomalies resulting from [transplacental infection](#) of the fetus with [varicella](#) during the first or second trimester of pregnancy; they may include eye abnormalities, brain damage, and moderate to severe limb abnormalities.

Conn syndrome, [primary aldosteronism](#).

Conradi syndrome, [chondrodysplasia punctata](#).

Conradi-Hünermann syndrome, an autosomal dominant form of chondrodysplasia punctata, characterized by asymmetric shortening of the extremities and scoliosis; intelligence and life expectancy are normal. The syndrome is also associated with maternal use of warfarin sodium during pregnancy.

constriction band syndrome, [amniotic band s.](#)

contiguous gene syndrome, any syndrome known to be caused by the involvement of contiguous genes on a chromosome, e.g., aniridia-Wilms tumor association, which may also have genitourinary tract abnormalities, gonadoblastoma, and mental retardation; they are usually caused by chromosome deletions.

continuous muscle activity syndrome, **continuous muscle fiber activity syndrome**, [Isaacs s.](#)

conus medullaris syndrome, a group of symptoms caused by compression of the conus medullaris, with diminished control of bowel and bladder functions, pain in the lower back, anesthesia in the pelvic region, and sometimes weakness in the lower limbs.

Cornelia de Lange syndrome, [de Lange s.](#)

syndrome of corpus striatum, [Vogt s.](#)

Costeff optic atrophy syndrome, [3-methylglutaconicaciduria type III](#).

Costen syndrome, [temporomandibular disorder](#).

costoclavicular syndrome, a [thoracic outlet syndrome](#) caused by compression or friction on nerves and blood vessels between a drooping clavicle and the first rib.

costosternal syndrome, [costochondritis](#).

Cotard syndrome, paranoia with delusions of negation, a suicidal tendency, and sensory disturbances.

Courvoisier-Terrier syndrome, dilatation of the gallbladder, retention jaundice, and discoloration of the feces, indicating obstruction due to a tumor of the ampulla of Vater.

cracked tooth syndrome, a group of symptoms caused by presence of a cracked tooth, including pain on pressure or application of cold, with pulpitis if untreated.

cramp-fasciculation syndrome, a relatively mild form of peripheral nerve hyperexcitability characterized by fasciculations, cramps, and intermittent myokymia, without neuromyotonia; it is usually idiopathic but in some cases is autoimmune in origin.

craniosynostosis-radial aplasia syndrome, [Baller-Gerold s.](#)

crazy cow syndrome, a type of neurotoxicity seen in cattle in the United States, Brazil, and South Africa after they have eaten any of various plants of the genus [Solanum](#); characteristics include cerebellar damage with staggering and incoordination.

CREST syndrome, a form of [systemic scleroderma](#) usually less severe than other forms, consisting of calcinosis cutis, Raynaud phenomenon, esophageal dysfunction, sclerodactyly, and telangiectasia. When esophageal dysfunction is not prominent, it is known as [CRST s.](#)

cricopharyngeal achalasia syndrome, [Asherson s.](#)

cri du chat syndrome, a hereditary congenital syndrome characterized by hypertelorism, microcephaly, severe mental deficiency, and a plaintive catlike cry, due to deletion of part of the short arm of chromosome 5. Called also [cat's cry s.](#)

Crigler-Najjar syndrome, an autosomal recessive form of [nonhemolytic jaundice](#) due to the absence of the hepatic enzyme [glucuronosyltransferase](#). It is characterized by the presence in the blood of excessive amounts of unconjugated [bilirubin](#), with [kernicterus](#) and severe disorders of the central nervous system. Called also [congenital hyperbilirubinemia](#) and [congenital nonhemolytic jaundice](#).

syndrome of crocodile tears, spontaneous lacrimation occurring parallel with the normal salivation of eating. It follows facial paralysis and seems to be due to inaccurate regrowth of the regenerating nerve fibers into the wrong nerve sheaths, with some of those destined for the salivary glands going to the lacrimal glands.

Cronkhite-Canada syndrome, a rare syndrome of sporadic, widespread intestinal polyps and [malabsorption](#) accompanied by ectodermal defects such as [alopecia](#) and [onychodystrophy](#); called also [Canada-Cronkhite s.](#)

Cross syndrome, **Cross-McKusick-Breen syndrome**, an autosomal recessive syndrome marked by cutaneous hypopigmentation, microphthalmos, small opaque corneas, gingival hypertrophy, and cerebral defect manifested by spasticity, mental and physical retardation, and athetoid movements. Called also [oculocerebral-hypopigmentation s.](#)

Crouzon syndrome, an autosomal dominant disorder caused by mutations in the FGFR2 gene (locus: 10q26), which encodes a fibroblast growth factor receptor, characterized by acrocephaly, exophthalmos, hypertelorism, strabismus, parrot-beaked nose, and hypoplastic maxilla with relative mandibular prognathism. Called also [craniofacial dysostosis](#).

Crow-Fukase syndrome, [POEMS s.](#)

crowned dens syndrome, crystal-induced arthritis around the dens axis, often accompanied by fever and neck pain.

CRST syndrome, see [CREST s.](#)

crush syndrome, the edema, oliguria, and other symptoms of [acute renal failure](#) that follow the crushing of a part, especially a large muscle mass; see [acute tubular necrosis](#), under necrosis.

Cruveilhier-Baumgarten syndrome, [cirrhosis](#) with [portal hypertension](#), associated with congenital patency of the umbilical or paraumbilical veins. It is characterized by [hematemesis](#), [ascites](#), [splenomegaly](#), [hypersplenism](#), [esophageal varices](#), [caput medusae](#), large tortuous veins in the abdominal wall, and a [venous hum](#), often accompanied by a [thrill](#), usually heard over the region of the [xiphoid process](#). Called also [Cruveilhier-Baumgarten cirrhosis](#).

cryopyrin-associated periodic syndrome, any in a group of autoinflammatory diseases caused by mutations in the gene that codes for cryopyrin, resulting in fever, arthralgia, urticaria, and inflammation. Called also [cryopyrinopathy](#).

cryptophthalmos syndrome, an autosomal recessive abnormality, characterized by absence of the palpebral apertures, disorganization of one or both ocular globes, malformed ears, cleft palate, laryngeal stenosis, syndactyly, meningoencephalocele, imperforate anus, cardiac defects, and maldeveloped kidneys. Called also [Fraser s.](#)

cubital tunnel syndrome, a type of entrapment neuropathy with a complex of symptoms resulting from injury or compression of the ulnar nerve at the elbow, including pain and numbness along the ulnar aspect of the hand and forearm, and weakness of the hand.

culture-specific syndrome, a form of disturbed behavior highly specific to certain cultural systems and that does not conform to Western nosologic entities; examples are amok, koro, piblokto, and windigo.

Currarino syndrome, a complex of congenital anomalies in the anococcygeal region; it consists of partial sacral agenesis; presacral anterior meningocele, teratoma, or cyst; and rectal malformations such as stenosis, ectopia, or imperforation. Gynecologic and renal malformations are also common. In at least some cases it is caused by mutation of the HLBX9 gene (locus: 7q36), a homeobox gene. Called also [Currarino triad](#).

Currarino-Silverman syndrome, premature obliteration of the sternal sutures with synostosis as well as a protruding manubrium, causing pectus carinatum; other abnormalities may also be present such as hyperostosis of ribs or hypotrophy of the anterior diaphragm. Called also [Silverman s.](#)

Curschmann-Batten-Steinert syndrome, [myotonic dystrophy](#).

Curtius syndrome, hypertrophy of one side of the entire body or a portion of one side of the body, as of the face; called also [hemihypertrophy](#).

Cushing syndrome, a complex of symptoms caused by [hyperadrenocorticism](#) due either to a neoplasm of the adrenal cortex or adenohypophysis, or to excessive intake of glucocorticoids. Symptoms may include adiposity of the face, neck, and trunk; kyphosis from osteoporosis of the spine; hypertension; diabetes mellitus; amenorrhea and hypertrichosis in females; impotence in males; dusky complexion with purple striae; polycythemia; and muscular wasting and weakness. When secondary to excessive pituitary secretion of [corticotropin](#), it is known as [Cushing disease](#). See also [ectopic ACTH s.](#) Called also [Cushing](#) or [pituitary basophilism](#).



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Moon facies, plethora, and supraclavicular fat associated with Cushing syndrome.

[equine Cushing disease](#).

Cushing syndrome, iatrogenic, Cushing syndrome caused by prolonged excessive use of glucocorticoid medications; called also [Cushing s. medicamentosus](#).

Cushing syndrome medicamentosus, iatrogenic Cushing s.

cyclic vomiting syndrome, cyclic vomiting.

Cyriax syndrome, a syndrome due to slipped rib cartilages pressing on the nerves at the interchondral joint, resulting in pain in the region of the cartilage, radiation of pain to the shoulder and arm, or pain similar to that of angina pectoris.

Da Costa syndrome, neurocirculatory asthenia.

Danbolt-Closs syndrome, opsoclonus-myoclonus s.

Dandy-Walker syndrome, see under [malformation](#).

Debré-Sémélaigne syndrome, autosomal recessive athrotic cretinism associated with myotonia and muscular pseudohypertrophy. Called also [Kocher-Debré-Sémélaigne s.](#)

de Clérambault syndrome, [erotomania](#).

defibrination syndrome, [diffuse intravascular coagulation](#).

Dejean syndrome, [orbital floor s.](#)

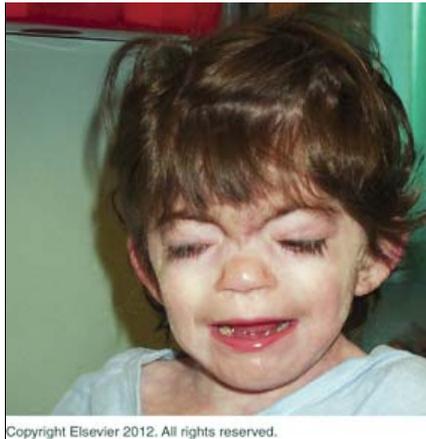
Dejerine syndrome, symptoms of radiculitis; namely, distribution of the pain, motor, and sensory defects in the region of the radicular or segmental disturbance of the nerve roots rather than along the course of the peripheral nerve. [bulbar s.](#) a polyneuropathy resembling tabes dorsalis, secondary to infection by [Corynebacterium diphtheriae](#) and the resultant lesions of peripheral nerves and of the posterior column of the spinal cord; deep sensibility is depressed but tactile sense is normal. Called also [diphtheritic polyneuropathy](#).

Dejerine-Klumpke syndrome, [Klumpke paralysis](#).

Dejerine-Roussy syndrome, [thalamic s.](#)

Dejerine-Thomas syndrome, [olivopontocerebellar atrophy](#).

de Lange syndrome, a congenital syndrome in which severe mental retardation is associated with many abnormalities, including short stature (Amsterdam dwarf), brachycephaly, low-set ears, webbed neck, Cupid's-bow mouth, depressed bridge of the nose with the end tilted up and forward-directed nostrils, arched heavy eyebrows meeting at the midline, unruly coarse hair growing low on the forehead and neck, and flat spadelike hands with short tapering fingers. Called also [Brachmann-de Lange](#) or [Cornelia de Lange s.](#)



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de Lange syndrome. Child showing typical facial features including finely arched heavy eyebrows, long eyelashes, small upturned nose, and long smooth philtrum.

del Castillo syndrome, [Sertoli-cell-only s.](#)

dementia syndrome of depression, reversible dementia occurring in association with depression in the elderly, the cognitive deficits resolving with treatment of the depression.

de Morsier syndrome, [septo-optic dysplasia](#).

dengue shock syndrome, see [hemorrhagic dengue](#), under dengue.

Dennie-Marfan syndrome, spastic paralysis and mental retardation in association with congenital syphilis.

Denys-Drash syndrome, a rare syndrome that includes male [pseudohermaphroditism](#), nephropathy leading to renal failure, and, in most cases, [Wilms tumor](#). It is caused by a genetic abnormality in the p13 region of chromosome 11.

depressive syndrome, [depression](#) (def. 3).

De Sanctis-Cacchione syndrome, an autosomal recessive syndrome consisting of [xeroderma pigmentosum](#) associated with mental retardation, retarded growth, gonadal hypoplasia, and sometimes neurologic complications and photosensitivity.

descending perineum syndrome, sagging of the perineum and levator plate, which causes constipation and other problems in the perineal region; it is seen most often after years of straining, vaginal deliveries, or surgical procedures in the area.

de Toni-Fanconi syndrome, see [Fanconi s.](#) (def. 2).

dialysis dysequilibrium syndrome, a group of symptoms seen during or after overly rapid [hemodialysis](#) or [peritoneal dialysis](#), resulting from an osmotic shift of water into the brain; usually there is headache and less often nausea, muscle cramps, nervous irritability, drowsiness, and convulsions.

Diamond-Blackfan syndrome, [congenital hypoplastic anemia](#) (def. 1).

diarrheogenic syndrome, [Verner-Morrison s.](#)

DIDMOAD syndrome, [Wolfram s.](#)

diencephalic syndrome, failure to thrive, emaciation, and sometimes nevus unius lateralis.

DiGeorge syndrome, a congenital disorder in which defective development of the third and fourth pharyngeal pouches results in hypoplasia or aplasia of the thymus and parathyroid glands, often associated with congenital heart defects, anomalies of the great vessels, esophageal atresia, and abnormalities of facial structures. Depending on the degree of parathyroid and thymic hypoplasia, there are hypocalcemic tetany or seizures due to lack of parathyroid hormone and deficiency of cell-mediated immunity resulting in increased susceptibility to low-grade or opportunistic pathogens. The syndrome is usually a phenotype of [22q11 deletion syndrome](#) (q.v.) and is associated with loss of the [T-box gene](#) Tbx1, and even within the phenotype shows variability. A small number of cases have other causes, notably a defect in chromosome 10p13. Called also [thymic aplasia](#) or [hypoplasia](#) and [pharyngeal pouch s.](#)

Di Guglielmo syndrome, [erythroleukemia](#).

disconnection syndrome, any neurologic disorder caused by an interruption in impulse transmission along cerebral fiber pathways; one result may be an inability to carry out a desired movement in response to a given sensory input, as in the apraxias.

disseminated intravascular coagulation syndrome, see under [coagulation](#).

distal intestinal obstruction syndrome, a complication of cystic fibrosis consisting of obstruction of the intestine by thickened feces; it resembles meconium ileus but lasts throughout life.

Dobrin syndrome, [TINU s.](#)

Donohue syndrome, a rare, lethal, autosomal recessive condition caused by mutation in the INSR gene (locus: 19p13.2), which encodes the insulin receptor, characterized by slow physical and mental development, [elfin facies](#), and endocrine abnormalities such as hyperinsulinemia and precocious puberty. Death usually occurs in utero or in the first year of life. Called also [leprechaunism](#).

DOOR syndrome, a rare syndrome of congenital deafness, onycho-osteodystrophy, and mental retardation, existing in autosomal dominant and recessive forms.

dorsal midbrain syndrome, [Parinaud s.](#)

double cortex syndrome, [band heterotopia](#).

double crush syndrome, compression or other damage of the same nerve at two different points, such as carpal tunnel syndrome with cervical radiculopathy. Some authorities believe that damage at one point makes a nerve group more vulnerable to injury somewhere else.

Down syndrome, a chromosome disorder characterized by a small, anteroposteriorly flattened skull, short, flat-bridge nose, epicanthal fold, short phalanges, widened spaces between the first and second digits of hands and feet, and moderate to severe mental retardation, with Alzheimer disease developing in the fourth or fifth decade. The chromosomal aberration is trisomy of chromosome 21 associated with late maternal age. Called also [trisomy 21](#) and [nondisjunction](#); formerly called [mongolism](#).



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Boy with typical facial features and small hands of Down syndrome.

downer cow syndrome, parturient paresis in a cow that is intractable to treatment and usually fatal.

Drash syndrome, [Denys-Drash s.](#)

Dresbach syndrome, [hereditary elliptocytosis](#).

Dressler syndrome, [post-myocardial infarction s.](#)

dropped head syndrome, weakness of posterior neck muscles, especially late in the day, so that the head drops forward; seen most often in polymyositis and certain motor neuron diseases.

dry eye syndrome, [keratoconjunctivitis sicca](#).

Duane syndrome, a hereditary congenital syndrome in which the affected eye shows limitation or absence of abduction, restriction of adduction, retraction of the globe on adduction, narrowing of the palpebral fissure on adduction and widening on abduction, and deficient convergence. It is transmitted as an autosomal dominant trait. Called also [retraction](#), [Stilling](#), or [Stilling-Türk-Duane s.](#)

Dubin-Johnson syndrome, **Dubin-Sprinz syndrome**, a familial chronic form of [nonhemolytic jaundice](#) thought to be due to a defect in the excretion of conjugated [bilirubin](#) and certain other organic anions (such as [sulfobromophthalein](#)) by the liver. It is characterized by the presence of a brown, coarsely granular pigment in the hepatic cells, which is pathognomonic of the condition. Called also [Sprinz-Dubin s.](#) and [Sprinz-Nelson s.](#)

Dubreuil-Chambardel syndrome, dental caries of the incisors, in most instances only the upper ones, usually appearing during adolescence; within a few years the teeth are irreparably damaged. Some authorities do not consider this syndrome a legitimate entity.

Duchenne syndrome, [progressive bulbar palsy](#).

Duchenne-Erb syndrome, [Erb-Duchenne paralysis](#).

dumping syndrome, a complex reaction seen with [rapid gastric emptying](#) (see under emptying). Characteristics include nausea, weakness, sweating, palpitation, varying degrees of syncope, often a sensation of warmth, and sometimes diarrhea. It occurs most often in patients who have had partial [gastrectomy](#) with [gastrojejunostomy](#). Called also [jejunal s.](#) and [postgastrectomy s.](#)

Duncan syndrome, [X-linked lymphoproliferative s.](#)

Dyke-Davidoff-Masson syndrome, a syndrome possibly due to injury to or severe disease affecting one side of the brain during the neonatal period, characterized by mental retardation, asymmetry of the face, and varying degrees of hemiplegia, neurological impairment, and atrophy of the side of the body contralateral to the lesion.

dysarthria-clumsy hand syndrome, [clumsy-hand dysarthria](#).

dyscontrol syndrome, a pattern of episodic, abnormal, and often violent and uncontrollable social behavior with little or no provocation; it may result from diseases of the limbic system or the temporal lobe or may accompany abuse of alcohol or some other psychoactive substance. Called also [episodic dyscontrol](#).

dyskinetic cilia syndrome, [primary ciliary dyskinesia](#).

dysmaturity syndrome, [postmaturity s.](#)

dysmnesic syndrome, [amnesic s.](#)

dysplastic nevus syndrome, the occurrence of [dysplastic nevi](#) in persons having or at risk for having familial [malignant melanoma](#). Called also [atypical mole s.](#), [atypical nevus s.](#), [familial atypical mole-malignant melanoma s.](#), and [FAMMM s.](#)

Eagle-Barrett syndrome, [prune-belly s.](#)

Eaton-Lambert syndrome, [Lambert-Eaton myasthenic s.](#)

ectopic ACTH syndrome, a condition caused by production of [corticotropin](#) by cells outside the pituitary, such as those of carcinoma of the lung; depending on its duration, it may be subtle, resembling true [Cushing disease](#), but hypokalemic alkalosis and weakness are often prominent.

ectopic corticotropin-releasing hormone syndrome, a disorder clinically indistinguishable from ectopic ACTH syndrome but caused by ectopic secretion of corticotropin-releasing hormone by a variety of tumors, generally bronchial carcinoid tumors.

ectrodactyly-ectodermal dysplasia-clefting syndrome, [EEC s.](#)

Edwards syndrome, [trisomy 18 s.](#)

EEC syndrome, a congenital syndrome inherited as an autosomal dominant trait involving both ectodermal and mesodermal tissues, which consists of ectodermal dysplasia associated with hypopigmentation of the skin and hair, scanty hair and eyebrows, absence of lashes, nail dystrophy, hypo- and microdontia, ectrodactyly, and cleft lip and palate. Called also [ectrodactyly-ectodermal dysplasia-clefting s.](#)

effort syndrome, [neurocirculatory asthenia](#).

egg drop syndrome, a viral disease of ducks and geese, caused by an adenovirus; apparently healthy birds begin laying eggs with thin or soft shells or without shells.

egg white syndrome, biotin deficiency; see [biotin](#).

Ehlers-Danlos syndrome, a group of inherited disorders of the [connective tissue](#); they were formerly classified into ten types, but more recently only six types are distinguished, varying widely in severity. The major manifestations include hyperextensible skin and joints, easy

bruability, friability of tissues with bleeding and poor wound healing, calcified subcutaneous [spheroids](#), and [pseudotumors](#). The [hypermobility](#) type (formerly type III) is autosomal dominant and the most common type; [mitral valve prolapse](#) accompanies the skin and joint anomalies. The classical type (formerly types I and II) has both autosomal dominant and autosomal recessive subtypes; it includes [mitral valve prolapse](#) as well as fibrous growths on pressure areas such as the knees and elbows. The vascular type (formerly type IV) is autosomal dominant and is characterized by fragile blood vessels and organs that may rupture, as well as distinctive facial features such as protruding eyes and thin nose and lips. The [kyphoscoliosis](#) type (formerly type VI) is a rare, autosomal recessive type characterized by [kyphoscoliosis](#) and eye fragility accompanying the bone and joint anomalies. The [arthrochalis](#) type (formerly types VIIA and VIIB) is a rare, autosomal dominant type in which joints are particularly loose and prone to dislocation; patients also suffer from arthritis and bone loss. The [dermatosparaxis](#) type (formerly type VIIC) is an autosomal recessive type characterized by particularly fragile and sagging skin. Most types are related to defects in [procollagen](#), and the [kyphoscoliosis](#) type is caused by a deficiency in the enzyme [lysyl hydroxylase](#). Called also [cutis hyperelastica](#).



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Hyperextensible skin of Ehlers-Danlos syndrome.

Eisenmenger syndrome, ventricular septal defect with pulmonary hypertension and cyanosis due to right-to-left (reversed) shunt of blood. Sometimes defined as pulmonary hypertension (pulmonary vascular disease) and cyanosis with the shunt being at the atrial, ventricular, or great vessel area.

Ekbom syndrome, [restless legs s.](#)

Ekman-Lobstein syndrome, [osteogenesis imperfecta](#), type I.

elfin facies syndrome, [Williams s.](#)

Ellis-van Creveld syndrome, an autosomal recessive disorder caused by mutation in either of two genes, EVC or EVC2 (locus for both: 4p12), the functions of whose products are unknown. It is characterized by short limbs, polydactyly, defective development of skin, hair, and teeth, and cardiac defects, most commonly affecting the atrial septum. Called also [chondroectodermal dysplasia](#).

embryonic testicular regression syndrome, [vanishing testes s.](#)

EMG syndrome, [Beckwith-Wiedemann s.](#)

empty sella syndrome, a syndrome diagnosed radiologically in which the diaphragma sellae is vestigial and the enlarged sella turcica forms an extension of the subarachnoid space and is filled with cerebrospinal fluid. The pituitary fossa appears empty, although the pituitary gland is present in a flattened form; pituitary hormone secretion may be normal, deficient, or excessive. Sometimes there is downward herniation of the optic chiasm, which leads to defects in the visual field.

encephalotrigeminal vascular syndrome, [Sturge-Weber s.](#)

eosinophilia-myalgia syndrome, a sometimes fatal combined syndrome of eosinophilia and severe generalized myalgia in patients ingesting L-tryptophan, occurring in the absence of infection, neoplasm, or other known causes of eosinophilia; other characteristics may include subjective weakness, fever, arthralgia, shortness of breath, rash, peripheral edema, and pneumonia.

epidermal nevus syndrome, any of several syndromes that combine [epidermal nevi](#) with involvement of other body systems, such as [CHILD syndrome](#), [Proteus syndrome](#), and [sebaceous nevus syndrome](#).

epiphyseal syndrome, precocious development of external genitalia and sexual function, precocious abnormal growth of long bones, appearance of signs of internal hydrocephalus, in the absence of all other motor and sensory symptoms. It has been attributed to pineal body dysfunction and to mechanical effects on the brain caused by tumors of the pineal body. Called also [Pellizzi s.](#), [pineal s.](#), and [macrogenitosomia praecox](#).

Epstein syndrome, [nephrotic s.](#)

equine Cushing syndrome, see under [disease](#).

erythrocyte auto sensitization syndrome, [painful bruising s.](#)

Escobar syndrome, [multiple pterygium s.](#) (def. 1).

euthyroid sick syndrome, subnormal levels of triiodothyronine in patients who have systemic illnesses but do not manifest symptoms of hypothyroidism. Called also [low T3 s.](#) and sick euthyroid s.

Evans syndrome, autoimmune hemolytic anemia accompanied by immune thrombocytopenia.

excited skin syndrome, nonspecific cutaneous hyperirritability of the back, sometimes occurring when multiple positive reactions are elicited in [patch tests](#) screening a battery of substances. Called also [angry back](#).

exfoliation syndrome, degenerative alterations of anterior ocular tissues, first presenting as disturbances of pigmentary distribution in the iris, with partial atrophy of the iris pigmentary epithelium and deposition of liberated pigment granules on the lens capsule, papillary margin, ciliary body, and zonule. In later stages, gray, flakelike material increasingly covers the same areas, and glaucoma is a frequent sequela. The incidence of the syndrome increases markedly with age. Called also [pseudoexfoliation](#).

exomphalos-macroglossia-gigantism syndrome, [Beckwith-Wiedemann s.](#)

exploding head syndrome, a rare, benign disturbance of the transition between waking and sleeping in which the person is aroused by the sensation of a loud explosion in the head, sometimes accompanied by flashing lights.

extrapyramidal syndrome, any of a group of clinical disorders considered to be due to malfunction in the [extrapyramidal system](#) and characterized by abnormal involuntary movements; included are parkinsonism, athetosis, and chorea.

Faber syndrome, [hypochromic anemia](#).

facet syndrome, **facet joint syndrome**, pain in the facet joints of the vertebral column, usually owing to degeneration of intervertebral cartilages.

faciodigitogenital syndrome, [Aarskog s.](#)

familial atypical mole syndrome, **familial atypical mole?melanoma syndrome**, **familial atypical mole?malignant melanoma syndrome**,

FAMMM syndrome, [dysplastic nevus s.](#)

familial cold autoinflammatory syndrome, an autosomal dominant [autoinflammatory disease](#) (q.v.) in the cryopyrin-associated periodic

syndromes subgroup, characterized by fever, polyarthralgia, myalgia, urticarial rash, leukocytosis, headache, and conjunctivitis.

Fanconi syndrome, a rare recessive disorder with a poor prognosis, characterized by pancytopenia, bone marrow hypoplasia, and patchy brown skin discoloration due to deposition of [melanin](#), as well as multiple congenital anomalies of the musculoskeletal and genitourinary systems. Called also [Fanconi anemia](#), [pancytopenia](#), or [panmyelopathy](#); [congenital hypoplastic anemia](#); [congenital pancytopenia](#); and [pancytopenia-dysmelia s](#), a general term for a group of diseases marked by dysfunction of the proximal renal tubules (see [proximal renal tubular acidosis](#), under acidosis) with generalized [hyperaminoaciduria](#), [renal glycosuria](#), [hyperphosphaturia](#), and bicarbonate and water loss. The most common cause is [cystinosis](#) (q.v.), but it is also associated with other genetic diseases and occurs in idiopathic and acquired forms. When unassociated with [cystinosis](#), the disorder is also called [de Toni-Fanconi syndrome](#).

Farber syndrome, **Farber-Uzman syndrome**, see under [disease](#).

fat cow syndrome, a syndrome seen in overly fat cows just after they have given birth; loss of appetite postpartum leads to mobilization of body fat stores with deposition of fat in the liver and ketosis, sometimes ending in coma and death. Called also [fatty liver disease](#) and [pregnancy toxemia in cows](#).

fat emboli syndrome, **fat embolism syndrome**, dyspnea, altered mental state, and petechiae caused by the entrance of fat into the vascular system, most often following trauma, especially that involving the fracture of long bones. It may also follow nontraumatic events, including lipid infusion and corticosteroid administration.

Favre-Racouchot syndrome, a type of [actinic elastosis](#) usually seen in elderly men, in which giant comedones, pilosebaceous cysts, and large folds of furrowed and yellowish skin are seen in the periorbital region. Called also [nodular elastosis](#) or [elastoidosis](#).

feline urological syndrome, dysfunction of the feline lower urinary tract. In male cats there is usually partial or complete obstruction from uroliths or other plugs, and in females there is more often cystitis or urethritis. There may be various causes, including decreased physical activity and excessive dietary magnesium.

Felty syndrome, a syndrome of splenomegaly with chronic rheumatoid arthritis and leukopenia; there are usually pigmented spots on the skin of the lower extremities, and sometimes there is other evidence of hypersplenism such as anemia or thrombocytopenia.

feminizing testes syndrome, [complete androgen insensitivity s](#).

fertile eunuch syndrome, a syndrome of [hypogonadotropic hypogonadism](#), with variable development of [secondary sex characters](#), associated with normal spermatogenesis, normal levels of follicle-stimulating hormone, and variably low levels of luteinizing hormone.

fetal alcohol syndrome, a syndrome of altered prenatal growth and morphogenesis seen in infants born to mothers who were chronically alcoholic during pregnancy; it includes facial anomalies such as maxillary hypoplasia, prominent forehead and mandible, short palpebral fissures, microphthalmia, and epicanthal folds, as well as growth retardation, microcephaly, and mental retardation.



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Typical facial features of an infant with fetal alcohol syndrome.

fetal face syndrome, [Robinow s](#).

fetal hydantoin syndrome, a symptom complex characterized by poor growth and development with craniofacial and skeletal abnormalities, produced by prenatal exposure to hydantoin analogues, including phenytoin.

Feuerstein-Mims syndrome, [sebaceous nevus s](#).

Fèvre-Languépin syndrome, popliteal webbing associated with cleft lip and palate, fistula of the lower lip, syndactyly, onychodysplasia, and pes equinovarus. Called also [popliteal pterygium s](#).

FG syndrome, an X-linked recessive syndrome of mental retardation, megalencephaly, imperforate anus and other gastrointestinal defects, delayed motor development, congenital hypotonia, characteristic facies and personality, short stature, skeletal anomalies, and congenital cardiac defects.

Fiessinger-Leroy-Reiter syndrome, [Reiter s](#).

first arch syndrome, anomalies, including macrostomia, hemignathia, and deformities of the external ear, resulting from an inhibitory process occurring toward the seventh week of embryonic life and affecting the facial bones derived from the first pharyngeal (branchial) arch.

first bite syndrome, pain in the parotid region upon initiation of mastication, caused by denervation supersensitivity of salivary gland myoepithelial cells following loss of sympathetic innervation to the parotid gland.

Fisher syndrome, a variant of [Guillain-Barré syndrome](#) characterized by areflexia, ataxia, and ophthalmoplegia. Called also [Miller Fisher s.one-and-a-half s](#).

Fitz-Hugh-Curtis syndrome, acute [perihepatitis](#), occurring as a complication of gonorrhea or chlamydial infection; it is usually seen in women, secondary to extension of infection from the fallopian tube to the hepatic capsule and overlying peritoneum. It is marked by fever, hepatic tenderness, abdominal pain, and signs of right upper quadrant peritoneal inflammation.

floppy infant syndrome, abnormal posture in an infant suspended in the prone position, the limbs and head hanging limply. It may be due to any of numerous conditions, including disorders of the brain, spinal cord, peripheral nerves, neuromuscular junction, muscles, or ligaments, and only sometimes is characterized by true limb weakness; the most common causes are perinatal injury to the brain or spinal cord, spinal muscular atrophy, and genetic disorders.

floppy valve syndrome, [mitral valve prolapse s](#).

Flynn-Aird syndrome, a rare autosomal dominant syndrome with abnormalities of the nervous system and ectodermal structures, including cataracts, retinitis pigmentosa, myopia, dental caries, skin atrophy and ulceration, peripheral neuropathy, ataxia, deafness, and cystic bone changes.

Foix syndrome, [cavernous sinus s](#).

Foix-Alajouanine syndrome, a necrotizing myelopathy characterized by necrosis of the gray matter of the spinal cord, thickening of the walls of the spinal vessels, and abnormal spinal fluid; symptoms include subacute spastic paraplegia of the lower extremities that progresses to flaccid paralysis, often ascending, loss of sphincter control, and progressive sensory loss. Death occurs in one to two years. Called also [subacute necrotic myelitis](#).

Foix-Chavany-Marie syndrome, [anterior opercular s](#).

folded lung syndrome, [round atelectasis](#).

follicular degeneration syndrome, a type of [cicatrical alopecia](#), usually beginning in the vertex of the scalp and extending outwards, caused by inflammation of hair follicles. It is sometimes called [hot comb alopecia](#) because it was originally seen in African-American women and thought to be related to hair relaxing procedures.

Forbes-Albright syndrome, [galactorrhea-amenorrhea syndrome](#) not associated with pregnancy; usually a prolactin-secreting pituitary tumor is present.

Forsius-Eriksson syndrome, X-linked [tapetoretinal degeneration](#), formerly thought to be a form of ocular albinism, caused by a mutation in the CACNA1F gene (locus: Xp11.23), which encodes a voltage-gated calcium channel subunit. Symptomatic males exhibit foveal hypoplasia, axial myopia, and protnomaly; female carriers have slightly defective color discrimination and latent nystagmus, but no mosaic pigment pattern in the fundus. Called also [Åland eye disease](#).

Förster syndrome, [Förster atonic-astatic syndrome](#), [atonic-astatic diplegia](#).

Foster Kennedy syndrome, [Kennedy s.](#)

Foville syndrome, a syndrome similar to the [Millard-Gubler syndrome](#), except that, in addition to paralysis of the outward movement of the eye, there is paralysis of conjugate movement.

fragile X syndrome, an X-linked syndrome associated with a [fragile site](#) on the X chromosome at q27.3, associated with mental retardation, enlarged testes, high forehead, and enlarged jaw and ears in most males and mild mental retardation in many heterozygous females. It is a [triplet repeat disorder](#), associated with expansion of CGG triplet repeat sequences in the promoter region of a gene expressed in human brain cells, the FMR1 (fragile X mental retardation 1) gene, with [triplet repeats](#) expanding from the normal 50 to as many as several thousand in the full syndrome. Between 50 and 200 copies, the sequence is unstable and prone to expansion; individuals appear normal, but are said to carry a (heritable) [premutation](#), and when over 200 copies are present, abnormalities of methylation occur in the promoter region and prevent expression of the gene, and clinical symptoms are apparent. The expansion and methylation abnormalities also appear to interfere with chromosome replication, producing the characteristic [fragile site](#).

Fraleigh syndrome, nephralgia with dilation of the renal calices around the upper pole of the kidney due to compression of the adjacent infundibulum, usually caused by pressure from vessels serving that part of the kidney.

Franceschetti syndrome, the complete form of [mandibulofacial dysostosis](#).

Franceschetti-Jadassohn syndrome, a rare autosomal dominant disorder characterized by gray to brown [reticular hyperpigmentation](#) beginning after infancy without preceding inflammatory changes; characteristics include [palmoplantar keratoderma](#), vasomotor changes with [hypohidrosis](#), and yellowing of the dental enamel.

François syndrome, [oculomandibulofacial s.](#)

Fraser syndrome, [cryptophthalmos s.](#)

Freeman-Sheldon syndrome, an autosomal dominant syndrome caused by mutation in the MYH3 gene (locus: 17p13.1), which encodes a myosin heavy chain. It consists of characteristic flattened, masklike facies; microstomia, the lips protruding as in whistling; deep-set eyes with hypertelorism; camptodactyly with ulnar deviation of the fingers; and talipes equinovarus. Called also [distal arthrogryposis type 2A](#), [craniocarpotarsal dystrophy](#), [whistling face syndrome](#), and [whistling face-windmill vane hand syndrome](#).

Frey syndrome, [auriculotemporal s.](#)

Friderichsen-Waterhouse syndrome, [Waterhouse-Friderichsen s.](#)

Fröhlich syndrome, [adiposogenital dystrophy](#).

Froin syndrome, a condition of the lumbar spinal fluid consisting of a transparent clear yellow color (xanthochromia), with the finding of large amounts of protein, rapid coagulation, and the absence of an increased number of cells. It is seen in certain organic nervous diseases in which the lumbar fluid is cut off from communication with the fluid in the ventricles. Called also [loculation s.](#)

Frommel-Chiari syndrome, [Chiari-Frommel s.](#)

Fuchs syndrome, unilateral heterochromia, fine keratic precipitates, and secondary cataract.

Fukuhara syndrome, [MERRF s.](#)

Fukuyama syndrome, [Fukuyama type congenital muscular dystrophy](#); see under dystrophy.

functional prepubertal castrate syndrome, [vanishing testes s.](#)

G syndrome, [Opitz s.](#)

Gaillard syndrome, dextrocardia from retraction of lungs and pleura to the right.

galactorrhea-amenorrhea syndrome, [amenorrhea](#) accompanied by [galactorrhea](#), sometimes associated with increased levels of [prolactin](#); several different types are known. See [Ahumada-del Castillo s.](#), [Chiari-Frommel s.](#), and [Forbes-Albright s.](#)

Ganser syndrome, the giving of inappropriate, ridiculous, or approximate answers to questions, sometimes associated with amnesia, disorientation, perceptual disturbances, and conversion symptoms; it is most commonly seen in malingering prisoners feigning psychosis.

Garcin syndrome, unilateral paralysis of all or most of the cranial nerves due to a tumor at the base of the skull or in the nasopharynx; called also [half base s.](#)

Gardner syndrome, a phenotypic variant of [familial adenomatous polyposis](#) characterized by extracolonic lesions, including supernumerary teeth, fibrous dysplasia of the skull, retinal pigmentation, osteomas, fibromas, and epithelial cysts.

Gardner-Diamond syndrome, [painful bruising s.](#)

gas bloat syndrome, excessive gas in the gastrointestinal tract because of incompetence of the [lower esophageal sphincter](#), such as after an operation to correct [gastroesophageal reflux disease](#).

Gasser syndrome, [hemolytic uremic s.](#)

gay bowel syndrome, term coined in the 1970s for an assortment of sexually transmitted bowel and rectal diseases affecting homosexual males and others who engage in frequent anal intercourse; it is caused by a wide variety of infectious agents.

Gélineau syndrome, [narcolepsy](#).

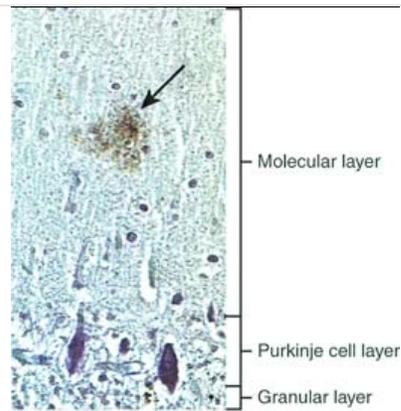
gender dysphoria syndrome, a group of psychological problems associated with discrepancy between the physical sex assignment and the psychological gender identity.

general adaptation syndrome, a syndrome defined by Hans Selye to include all nonspecific systemic reactions of the body to prolonged exposure to systemic stress; he described three stages in the reacting: the alarm reaction, resistance, and exhaustion.

genital ulcer syndrome, any of the diseases or conditions causing ulcerations of the genitalia, most commonly syphilis or herpes simplex, but also chancroid, lymphogranuloma venereum, granuloma inguinale, or trauma.

Gerstmann syndrome, a combination of finger agnosia, right-left disorientation, agraphia, acalculia, and often constructional apraxia; it was formerly attributed to a lesion in the angular gyrus of the dominant hemisphere, but now that etiology is in doubt.

Gerstmann-Sträussler syndrome, **Gerstmann-Sträussler-Scheinker syndrome**, a group of rare [prion diseases](#), of autosomal dominant inheritance but linked to different mutations of the [prion protein](#) gene, having the common characteristics of cognitive and motor disturbances and the presence of multicentric amyloid plaques in the brain. In the ataxic form, there are progressive cerebellar ataxia and dementia; in the telencephalic form, there are dysarthria, dementia, rigidity, tremor, and hyperreflexia; in GSS with neurofibrillary tangles, there are progressive short-term memory loss and clumsiness. Death occurs in 1 to 5 years.



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Gerstmann-Sträussler syndrome; immunostaining of a section through the cerebellum with antibodies to prion protein demonstrates an amyloid plaque (arrow) in the molecular layer.

Gianotti-Crosti syndrome, a viral disease of young children, generally benign and self-limited, characterized by skin-colored or reddish, flat-topped, firm papules forming a symmetrical eruption in an acral distribution, usually on the face, buttocks, or limbs, including the palms and soles. Symptoms are mild; the child may have malaise and a low grade fever. Several different viruses may cause it, but most commonly the [hepatitis B virus](#). Called also [infantile acrodermatitis](#) and [papular acrodermatitis of childhood](#).

giant platelet syndrome, [Bernard-Soulier s.](#)

Gilbert syndrome, an inborn error of [bilirubin](#) metabolism, probably autosomal dominant, characterized by a benign elevation of unconjugated bilirubin without liver damage or hematologic abnormalities. Called also [familial](#) or [Gilbert cholemia](#), [hyperbilirubinemia I](#), [constitutional hyperbilirubinemia](#), and [familial nonhemolytic jaundice](#).

Gilles de la Tourette syndrome, a syndrome comprising both multiple motor and one or more vocal tics, occurring over a period of at least one year, at least intermittently but sometimes as frequently as many times daily. Obsessions, compulsions, hyperactivity, distractibility, and impulsivity are often associated. Onset is in childhood and tics often lessen in severity and frequency and may even remit during adolescence and adulthood. Called also [Guinon disease](#), [maladie des tics](#), and [Tourette s.](#)

Gillespie syndrome, a rare autosomal recessive syndrome consisting of aniridia, cerebellar ataxia, and mental retardation.

Gitelman syndrome, a syndrome of hypertrophy of [juxtaglomerular cells](#) similar to [Bartter syndrome](#) but with [hypocalciuria](#) and [hypomagnesemia](#); usually seen in adolescents or adults.

Gjessing syndrome, recurrent episodes of stupor or excitement occurring in [catatonic schizophrenia](#) in association with variations in nitrogen metabolism and thyroid hormone level; it is related to nitrogen retention caused by inadequate metabolism of dietary protein.

glioma-polyposis syndrome, [Turcot s.](#)

glucagonoma syndrome, the spectrum of symptoms caused by a [glucagonoma](#), associated with high blood levels of [glucagon](#), mild [diabetes mellitus](#), weight loss, anemia, glossitis, [stomatitis](#), [angular cheilitis](#), [blepharitis](#), and [necrolytic migratory erythema](#).

Goldberg syndrome, [galactosialidosis](#).

Goldenhar syndrome, [oculoauriculovertebral dysplasia](#).

Goltz syndrome, **Goltz-Gorlin syndrome**, [focal dermal hypoplasia](#).

Good syndrome, [immunodeficiency with thymoma](#).

Goodman syndrome, an autosomal recessive disorder resembling [Carpenter syndrome](#) but characterized also by congenital heart defects, clinodactyly, camptodactyly, and ulnar deviation; it is now generally believed to be a variant of [Carpenter syndrome](#). Called also [acrocephalopolysyndactyly, type IV](#).

Goodpasture syndrome, a syndrome of [glomerulonephritis](#), pulmonary hemorrhage, and circulating [anti-GBM antibodies](#); it usually occurs in young men and is characterized by rapidly progressing [renal failure](#) as well as hemoptysis, pulmonary infiltrates, and dyspnea. Cf. [anti-GBM antibody nephritis](#) and [pulmonary renal s.](#)

Gopalan syndrome, a symptom complex resulting from malnutrition, probably from deficiency of riboflavin or pantothenic acid; it consists of a burning sensation in the extremities, a feeling of "pins and needles" in the distal parts, and hyperhidrosis. Called also [burning feet](#) and [burning feet s.](#)

Gordon syndrome, [pseudohypoadosteronism type 2](#). an autosomal dominant disorder characterized by camptodactyly, talipes, and cleft palate; called also [distal arthrogyriposis type 3](#).

Gorlin syndrome, **Gorlin-Goltz syndrome**, [nevoid basal cell carcinoma s.](#)

Gougerot-Blum syndrome, [pigmented purpuric lichenoid dermatitis](#).

Gougerot-Carteaud syndrome, [confluent and reticulate papillomatosis](#).

Gougerot-Nulock-Houwer syndrome, [Sjögren s.](#)

Gowers syndrome, [vasovagal syncope](#).

Gradenigo syndrome, paralysis of the abducens nerve and unilateral headache in chronic suppurative otitis media, caused by direct spread of the infection to involve the abducens and trigeminal nerves.

Graham Little syndrome, a syndrome characterized by cicatricial patches of alopecia of the scalp with prominent follicular plugging and follicular keratoses on the trunk and limbs, sometimes associated with noncicatricial alopecia of the axillae, pubic region, trunk, and limbs.

gray syndrome, **gray baby syndrome**, a potentially fatal condition seen in neonates, particularly premature infants, due to a reaction to [chloramphenicol](#), characterized by an ashen gray cyanosis, listlessness, weakness, and hypotension.

gray collie syndrome, [cyclic neutropenia](#).

gray platelet syndrome, a rare deficiency of the [alpha granules](#) of [platelets](#), resulting in a [bleeding disorder](#) that may include [ecchymoses](#), [petechiae](#), and [epistaxis](#) from infancy on.

green nail syndrome, greenish-black discoloration of the nail plate secondary to *Pseudomonas aeruginosa* infection of the paronychial tissues, caused by the diffusion of pyocyanin produced by the bacteria into the nail bed.

Greig syndrome, [ocular hypertelorism](#).

Griscelli syndrome, an albinoidism of autosomal recessive inheritance, marked by hypomelanosis, frequent pyogenic infection, hepatosplenomegaly, neutro- and thrombopenia, and possible immunodeficiency. Called also [hypopigmentation-immunodeficiency disease](#).

Grisel syndrome, subluxation of the atlantoaxial joint after an upper respiratory tract infection or an adenoidectomy, usually seen in children.

Grönblad-Strandberg syndrome, angioid streaks in the retina together with pseudoxanthoma elasticum of the skin.

Gruber syndrome, [Meckel s.](#)

Guillain-Barré syndrome, rapidly progressive ascending motor neuron paralysis of unknown etiology, frequently seen after an enteric or respiratory infection. An autoimmune mechanism following viral infection has been postulated. It begins with paresthesias of the feet, followed

by flaccid paralysis of the entire lower limbs, ascending to the trunk, upper limbs, and face; other characteristics include slight fever, bulbar palsy, absent or lessened tendon reflexes, and increased protein in the cerebrospinal fluid without a corresponding increase in cells. Variant forms include acute autonomic neuropathy, Miller-Fisher syndrome, acute motor axonal neuropathy, and acute motor-sensory axonal neuropathy. Called also [Guillain-Barré polyneuritis](#) or [polyneuropathy](#); Landry paralysis or syndrome; and acute idiopathic, [acute infectious](#), or [acute postinfectious polyneuritis](#).

Gulf War syndrome, a group of symptoms of unknown cause, seen in military personnel of the United States and its allies in the Persian Gulf conflict of the early 1990s, consisting of widespread pain including fibromyalgia and headaches, gastrointestinal distress, and memory disorders.

Gunn syndrome, unilateral ptosis of the eyelid, with the association of movements of the affected upper eyelid with those of the jaw; called also [Gunn phenomenon](#), [Marcus Gunn s.](#) or [phenomenon](#), and [jaw-winking s.](#) or [phenomenon](#).

gustatory sweating syndrome, [auriculotemporal s.](#)

Haddad syndrome, a rare association of congenital central hypoventilation syndrome and Hirschsprung disease.

Hadfield-Clarke syndrome, [Clarke-Hadfield s.](#)

Hakim syndrome, [normal-pressure hydrocephalus](#).

half base syndrome, [Garcin s.](#)

Hallermann-Streiff syndrome, **Hallermann-Streiff-François syndrome**, [oculomandibulofacial s.](#)

Hallopeau-Siemens syndrome, [recessive epidermolysis bullosa dystrophica](#).

halzoun syndrome, [halzoun](#).

Hamman syndrome, [pneumomediastinum](#).

Hamman-Rich syndrome, [acute interstitial pneumonia](#).

hand-arm vibration syndrome, a common [occupational disease](#) consisting of [acrocyanosis](#) in a person experiencing prolonged repetitive hand and arm vibrations. Called also [dead finger](#) and [vibration white finger](#).

hand-foot syndrome, a syndrome of [erythema](#) and [dysesthesias](#) of touch in the palms and soles, followed by desquamation; seen as a reaction to certain chemotherapeutic agents. Called also [acral](#), [palmar-plantar](#), or [palmoplantar erythrodysesthesia](#) and [palmar-plantar erythrodysesthesia syndrome](#).

hand-foot-uterus syndrome, a congenital syndrome consisting of small feet with unusually short great toes, abnormal thumbs, and, in females, duplication of the genital tract.

hand-shoulder syndrome, [shoulder-hand s.](#)

Hanhart syndrome, any of several syndromes of variable inheritance, characterized chiefly by severe micrognathia, high nose root, small eyelid fissures, low-set ears, and variable absence of digits or limbs, usually below the elbow or knee.

Hanot-Chauffard syndrome, hypertrophic cirrhosis with pigmentation and diabetes mellitus.

hantavirus pulmonary syndrome, a sometimes fatal febrile illness caused by viruses of the genus [Hantavirus](#), spread to humans by various species of mice and rats; characteristics include variable respiratory symptoms followed by acute respiratory distress that may progress to respiratory failure.

happy puppet syndrome, [Angelman s.](#)

Harada syndrome, [Vogt-Koyanagi-Harada s.](#)

HARD syndrome, [Walker-Warburg s.](#)

Hare syndrome, [Pancoast s.](#) (def. 1).

Harris syndrome, hyperinsulinism due to organic endogenous factors, such as insulinoma, manifested by hypoglycemia, weakness, perspiration, jitteriness, tachycardia, mental confusion, and disturbances of vision.

Hartnup syndrome, see under [disease](#).

haw syndrome, protrusion of one or both of the nictitating membranes of a dog or cat. Called also [haw](#).

Haw River syndrome, an autosomal dominant neurodegenerative disorder characterized by ataxia, chorea, seizures, and dementia, with onset usually between ages 15 to 30 years, and death within 15 to 25 years. It is a [triplet repeat disorder](#) caused by expansion of the CAG triplet repeat in the DPRLA gene encoding the atrophin-1 protein, as occurs in [dentatorubral-pallidoluysian atrophy](#) (q.v.), although there are some differences in clinical expression and pathology, including lack of myoclonic seizures, and extensive demyelination of the subcortical white matter, basal ganglia calcifications, and neuroaxonal dystrophy.

Hay-Wells syndrome, an autosomal dominant syndrome of ectodermal dysplasia, cleft lip and palate, and ankyloblepharon filiforme adnatum; it is also characterized by hypodontia, palmar and plantar keratoderma, partial anhidrosis, sparse wiry hair, and sometimes otologic defects.

Called also [AEC s.](#) and [ankyloblepharon-ectodermal dysplasia-clefting s.](#)

Hayem-Widal syndrome, former name for [hemolytic anemia](#).

heart-hand syndrome, [Holt-Oram s.](#)

Hecht syndrome, **Hecht-Beals syndrome**, **Hecht-Beals-Wilson syndrome**, [trismus-pseudocamptodactyly s.](#)

Heerfordt syndrome, an occasional manifestation of sarcoidosis consisting of enlargement of the parotid and lacrimal glands, anterior uveitis, Bell palsy, and fever. Called also [uveoparotid fever](#).

Heidenhain syndrome, a rapidly progressive degenerative disease manifested by cortical blindness, presenile dementia, dysarthria, ataxia, athetoid movements, and generalized rigidity.

HELLP syndrome, hemolysis, elevated liver enzymes, and low platelet count occurring in association with [preeclampsia](#).

Helweg-Larsen syndrome, an autosomal dominant syndrome consisting of anhidrosis present from birth and labyrinthitis later in life.

hemangioma-thrombocytopenia syndrome, [Kasabach-Merritt s.](#)

hemolytic uremic syndrome, a form of [thrombotic microangiopathy](#) usually seen in children, characterized by [renal failure](#), [hemolytic anemia](#), and severe [thrombocytopenia](#) and [purpura](#). Some authorities consider it identical to [thrombotic thrombocytopenic purpura](#). Called also [Gasser s.](#)

hemophagocytic syndrome, see under [lymphohistiocytosis](#).

hemopleuropneumonic syndrome, a syndrome of dyspnea, hemoptysis, tachycardia, fever, pneumonia, and hydrothorax occurring after a puncture wound of the chest.

Hench-Rosenberg syndrome, [palindromic rheumatism](#).

Henoch-Schönlein syndrome, see under [purpura](#).

hepatopulmonary syndrome, arterial [hypoxemia](#) caused by pulmonary [vasodilation](#) in conjunction with chronic liver disease, usually occurring as a result of [portal hypertension](#) in [cirrhosis](#).

hepatorenal syndrome, functional [renal failure](#), [oliguria](#), and low urinary [sodium](#) concentration, without pathological renal changes, associated with [cirrhosis](#) and [ascites](#) or with [obstructive jaundice](#).

hereditary mixed polyposis syndrome, an autosomal dominant syndrome characterized by the occurrence of atypical [juvenile polyps](#) of mixed histologic type, colonic [adenomas](#), and colorectal [carcinomas](#).

hereditary periodic fever syndromes, [autoinflammatory diseases](#).

Hermansky-Pudlak syndrome, a rare, genetically heterogeneous, multisystem disorder characterized by oculocutaneous albinism, hemorrhagic diathesis secondary to a platelet defect, pulmonary fibrosis, colitis, and accumulation of ceroid in the reticuloendothelial system and oral mucosa and its excretion in urine; cardiomyopathy and renal failure may also occur. Eight types have been identified, and its inheritance is autosomal recessive.

Herrmann syndrome, an autosomal dominant syndrome characterized initially by photomyogenic seizures and progressive deafness, with

later development of diabetes mellitus, nephropathy, and mental deterioration progressing to dementia.

heterotaxy syndrome, a variable set of complex congenital anomalies of the gastrointestinal and cardiovascular systems that results from [heterotaxia](#) of the abdominal and thoracic viscera.

HHH syndrome, [hyperornithinemia-hyperammonemia-homocitrullinuria s.](#)

high-pressure neurologic syndrome, a group of neurologic and neuropsychiatric signs and symptoms seen in deep sea divers below 100 meters, especially in those breathing heliox mixtures; symptoms vary, depending on the depth and gas mixture, and include tremor, incoordination, myoclonus, opsochonus, headache, vertigo, nausea, euphoria, and cognitive disturbances.

Hines-Bannick syndrome, intermittent attacks of low temperature and disabling sweating.

Hinman syndrome, a psychogenic disorder seen in children, imitating a [neurogenic bladder](#), consisting of [detrusor-sphincter dyssynergia](#) without evidence of any neural lesions. Called also [nonneurogenic neurogenic bladder](#).

HIV wasting syndrome, [wasting s.](#)

Hoffmann-Werdnig syndrome, [Werdnig-Hoffmann disease](#).

holiday heart syndrome, paroxysms of arrhythmias, most commonly atrial fibrillation, in patients without overt cardiomyopathy after a weekend bout of alcoholic consumption, especially during the year-end holiday season.

Holmes-Adie syndrome, [Adie s.](#)

Holt-Oram syndrome, autosomal dominant heart disease of varying severity, usually an atrial or ventricular septal defect, in association with upper limb malformation consisting of narrow shoulders, hypoplasia of the radius, and phocomelia, most commonly absence of the thumb and radius. Called also [heart-hand s.](#)



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Holt-Oram syndrome. Radiograph shows the absence of a radius shadow (arrow) and missing thumb.

honker syndrome, a disease of feedlot cattle, of unknown etiology, characterized by edema of the lower trachea with dyspnea and a honking sound during inspiration.

Horner syndrome, Horner-Bernard syndrome, sinking in of the eyeball, ptosis of the upper eyelid, slight elevation of the lower lid, constriction of the pupil, narrowing of the palpebral fissure, and anhidrosis and flushing of the affected side of the face; caused by a brainstem lesion on the ipsilateral side that interrupts sympathetic nerve fibers. See also [Horner ptosis](#). Called also [Bernard](#) or [Bernard-Horner s.](#) and [oculosympathetic palsy](#).

Horton syndrome, [giant cell arteritis](#).

hot foot syndrome, [Pseudomonas hot foot s.](#)

Howel-Evans syndrome, a rare type of [diffuse palmoplantar keratoderma](#) usually seen in children and associated with development of esophageal cancer later in life.

Hughes-Stovin syndrome, thrombosis of the pulmonary arteries and peripheral veins, characterized by headache, fever, cough, papilledema, and hemoptysis.

hungry bone syndrome, a condition seen after [parathyroidectomy](#) in patients who had had [hyperparathyroidism](#); rapid deposition of calcium in bones leads to [hypocalcemia](#).

Hunt syndrome, [Ramsay Hunt s.](#)

Hunter syndrome, a [mucopolysaccharidosis](#) caused by deficiency of iduronate-2-sulfatase, characterized by excretion of dermatan sulfate and heparan sulfate in the urine; it differs clinically from [Hurler syndrome](#) by (1) X-linked inheritance; (2) slower progression, less severity, and longer survival (thus resembling the Hurler-Scheie syndrome); and (3) absence of corneal clouding. Two clinical forms exist: the severe form has Hurler-Scheie-like symptoms with death before 15, usually from heart disease; the mild form has onset in the first decade, reduced somatic involvement, and near-normal intelligence and lifespan. Called also [mucopolysaccharidosis II](#).

Hurler syndrome, the prototype of the [mucopolysaccharidoses](#), and the gravest of the three allelic disorders of mucopolysaccharidosis I. It is caused by deficiency of [L-iduronidase](#), and onset is after the first year with progressive physical and mental deterioration; there is corneal clouding, with death usually by age 10 caused by respiratory infection and heart failure. Further symptoms include gargoyle-like facies with hypertelorism, depressed nasal bridge, large tongue, and widely spaced teeth; dwarfism; severe somatic and skeletal changes, including short neck and trunk, scaphocephaly, and kyphosis with gibbus; short broad hands with short fingers; progressive opacities of the cornea; deafness; cardiovascular defects; hepatosplenomegaly; and joint contractures. Called also [mucopolysaccharidosis IH](#).



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Hurler syndrome. Child showing typical coarsening of facial features including prominent forehead, flattened nasal bridge, short broad nose, and widening of the lips.

Hurler-Scheie syndrome, one of the three allelic disorders of mucopolysaccharidosis I, with clinical features intermediate between the Hurler and the Scheie syndromes, caused by deficiency of I-iduronidase, and specifically characterized by receding chin (micrognathism). Symptoms include mental retardation, dwarfism, dysostosis multiplex, corneal clouding, deafness, hernia, stiff joints (claw hand), and valvular heart disease. Patients survive until their late teens or twenties. Called also [mucopolysaccharidosis IH/S](#).

Hutchinson syndrome, see under [triad](#).

Hutchinson-Gilford syndrome, [progeria](#).

Hutchison syndrome, neuroblastoma with metastases to the skull.

17-hydroxylase deficiency syndrome, 17 α -hydroxylase deficiency; see under H.

hyperabduction syndrome, a [thoracic outlet syndrome](#) due to compression of the brachial plexus trunk roots and axillary vessels by the pectoralis minor muscle and the coracoid process when the arms are stretched above the head, as during sleep. Called also [Wright s.](#)

hyperactive child syndrome, former name for [attention-deficit/hyperactivity disorder](#).

hypercalcemia syndrome, [milk-alkali s.](#)

hypereosinophilic syndrome, any of several diseases characterized by a massive increase in the number of [eosinophils](#) in the blood and bone marrow, with eosinophilic infiltration of other organs. Symptoms vary, depending on the organ involved, and may include pruritic skin ulcers or [erythroderma](#), [endomyocarditis](#), lymph node or spleen enlargement, and ophthalmologic or gastrointestinal complications.

[Eosinophilic leukemia](#) is a potentially fatal member of the group.

hyperglycemic hyperosmolar syndrome, hyperglycemic hyperosmolar nonketotic syndrome, see under [state](#).

hyper-IgE syndrome, [hyperimmunoglobulinemia E s.](#)

hyperimmunoglobulinemia D syndrome, hyperimmunoglobulinemia D with periodic fever syndrome, an [autoinflammatory disease](#) (q.v.) inherited as an autosomal recessive trait, caused by mutations in the gene for [mevalonate kinase](#), with onset usually before one year of age. It is characterized by attacks of high fever preceded by chills, occurring about every 4 to 8 weeks and lasting 4 to 6 days, often with headache, arthritis and arthralgia, erythematous lesions, and hepatosplenomegaly; serum IgD levels are continuously high. Called also [Dutch type periodic fever](#).

hyperimmunoglobulinemia E syndrome, a primary [immunodeficiency](#) disorder caused by mutations of the STAT3 gene (see [STAT](#)). It is usually of autosomal dominant inheritance but sometimes occurs sporadically and is characterized by recurrent staphylococcal abscesses of skin, lungs, joints, and other sites; pruritic dermatitis; distinctive coarse facies; retained primary dentition; skeletal abnormalities; eosinophilia; and very high serum immunoglobulin E (IgE) levels. Called also [Buckley s.](#) and [Job s.](#) an autosomal recessive syndrome that is clinically similar to the autosomal dominant form but lacks the skeletal abnormalities.

hyperinfection syndrome, a highly fatal form of strongyloidiasis, most often occurring in immunocompromised persons and characterized by massive infection and widespread hematogenous dissemination of parasites to organs not normally affected. Manifestations are highly variable; pulmonary signs and symptoms resemble those of acute respiratory distress syndrome.

hyperkinetic syndrome, former name for [attention-deficit/hyperactivity disorder](#).

hyperkinetic heart syndrome, increased cardiac output of unknown cause associated with slightly elevated systolic and pulse pressures, normal mean arterial pressure, and low systemic vascular resistance.

hyperlucent lung syndrome, a syndrome simulating localized emphysema, but due to congenital absence or hypoplasia of pulmonary arteries; there may be lobar or segmental agenesis, as well as accessory lungs, lobes, or segments.

hypermobility syndrome, [benign joint hypermobility s.](#)

hyperornithinemia-hyperammonia-homocitrullinuria syndrome, an autosomal recessive syndrome characterized by elevated plasma levels of [ornithine](#), postprandial [hyperammonemia](#) and [homocitrullinuria](#), and aversion to protein ingestion. It is believed to result from a defect in the transport of [ornithine](#) into mitochondria, which disturbs the cycle of [ureagenesis](#). Called also [HHH s.](#)

hyperosmolar hyperglycemic syndrome, [hyperglycemic hyperosmolar state](#).

hyperperfusion syndrome, a complex of symptoms that sometimes follows carotid endarterectomy procedures, consisting of headache, seizures, focal deficits, brain edema, and sometimes cerebral hemorrhage; the cause is believed to be loss of arterial regulatory ability on the side where an occlusion has been removed.

hyperreactive malarial spleen syndrome, [tropical splenomegaly s.](#)

hypersomnia-bulimia syndrome, [Kleine-Levin s.](#)

hypertelorism-hypospadias syndrome, [Opitz s.](#)

hyperventilation syndrome, a complex of symptoms that accompany [hypocapnia](#) caused by [hyperventilation](#), including palpitations, shortness of breath, lightheadedness, profuse perspiration, and tingling sensations in the fingertips, face, or toes; prolonged overbreathing may result in vasomotor collapse and loss of consciousness. [Hyperventilation](#) unrecognized by the patient is a common cause of the subjective somatic symptoms associated with chronic anxiety or panic attacks (see [panic disorder](#), under disorder).

hyperviscosity syndrome, any of various syndromes associated with increased [viscosity](#) of the blood. One type is due to serum [hyperviscosity](#) and is characterized by spontaneous bleeding with neurologic and ocular disorders. Another type is characterized by [polycythemia](#) with retarded blood flow, organ congestion, reduced capillary perfusion, and increased cardiac effort. A third group includes conditions in which the deformability of [erythrocytes](#) is impaired, such as [sickle cell anemia](#).

hypocomplementemic urticarial vasculitis syndrome, a potentially severe vasculitis characterized by recurrent urticaria with hypocomplementemia and the presence of autoantibodies to complement component C1q. Extracutaneous involvement may occur and can be life-threatening.

hypoglossia-hypodactyly syndrome, a rare syndrome consisting of partial to complete absence of the tongue and of the digits or one or more limbs. Called also [aglossia-adactylia s.](#)

hypoplastic left heart syndrome, any of a group of congenital anomalies consisting of hypoplasia or atresia of the left ventricle and of the

aortic or mitral valve or both and hypoplasia of the ascending aorta; it is characterized by respiratory distress and extreme cyanosis, with cardiac failure and death in early infancy.

hypothernar hammer syndrome, traumatic aneurysm of the ulnar artery at the hypothenar in persons who repeatedly use the hypothenar to push or pound.

hypotonic syndromes, a group of syndromes involving inadequate water excretion in comparison to the amount ingested, so that body fluids become hypotonic and hyponatremic; some are due to excessive water intake as in [water intoxication](#), while others are caused by derangements of the excretory process such as the [vasopressin](#) excess in the [syndrome of inappropriate diuretic hormone](#) or complications of the [nephrotic syndrome](#), congestive heart failure, or kidney failure.

hypotrichosis-lymphedema-telangiectasia syndrome, a rare, autosomal recessive condition consisting of progressive hair loss, lymphedema of the lower limbs, and telangiectasias on various parts of the body.

IBIDS syndrome, [ichthyosis, brittle hair, infertility, developmental delay, short stature] [Tay s.](#)

idiopathic postprandial syndrome, the repeated occurrence of the clinical manifestations of hypoglycemia after meals; a controversial disease entity.

iliotibial band syndrome, a type of overuse injury in athletes caused by repetitive rubbing of the iliotibial tract (or band) against the lateral femoral epicondyle as the hip is repetitively extended and flexed; the primary symptom is severe pain in the thigh and knee.

Imlerslund syndrome, **Imlerslund-Graesbeck syndrome**, a rare familial form of [megaloblastic anemia](#), usually transmitted as an autosomal recessive trait, characterized by selective intestinal malabsorption of vitamin B₁₂ uninfluenced by intrinsic factor, and associated with proteinuria and structural genitourinary tract anomalies.

immobile cilia syndrome, [primary ciliary dyskinesia](#).

impingement syndrome, a type of overuse injury with progressive pathologic changes resulting from mechanical [impingement](#) by the [acromion](#), [coracoacromial ligament](#), [coracoid process](#), or [acromioclavicular joint](#) against the [rotator cuff](#); changes may include reversible edema and hemorrhage, fibrosis, tendinitis, pain, bone [spur](#) formation, and tendon rupture.

syndrome of inappropriate antidiuresis, a disorder of sodium and water balance characterized by [hyponatremia](#) with [hypovolemia](#) and elevated urine [osmolality](#) in the absence of kidney disease; it may be caused by inappropriate secretion of vasopressin (s. of inappropriate antidiuretic hormone) or mutations affecting the ability of the renal collecting tubules to absorb water (nephrogenic s. of inappropriate antidiuresis).

syndrome of inappropriate antidiuretic hormone, hyponatremia with decreased extracellular fluid osmolality, inability to produce dilute urine, normovolemia, and elevated urinary sodium excretion, in the absence of kidney disease or other cause of normovolemic hypo-osmolality. Causes include [vasopressin](#)-secreting tumor cells, neoplasms (especially oat cell carcinoma of the lung or pancreatic carcinoma), pulmonary disorders, central nervous system disease, and adverse drug reactions.

inferior syndrome of red nucleus, [Claude s.](#)

inhibitory syndrome, the manifestations produced by a somatostatinoma, including diabetes mellitus, cholecystolithiasis, steatorrhea, indigestion, hypochlorhydria, and occasionally anemia.

inspissated bile syndrome, biliary obstruction caused by plugging of the outflow tract.

insulin autoimmune syndrome, a rare condition in which hypoglycemia is produced by extremely high concentrations of autoantibodies to insulin, seen primarily in persons of Asian descent and in most cases associated with treatment with sulfhydryl-containing medications.

intersection syndrome, a type of overuse injury with tenosynovitis about 4 cm proximal to the wrist, at the point where the tendons of the abductor pollicis longus and extensor pollicis brevis muscles cross over those of the extensor carpi radialis longus and brevis muscles; it is caused by frequent, repetitive wrist movements such as in athletes. Symptoms are similar to those of de Quervain disease.

intrauterine parabiocytic syndrome, [placental transfusion](#); see under transfusion.

irritable bowel syndrome, **irritable colon syndrome**, a common, chronic, noninflammatory condition characterized by abdominal pain and altered bowel habits (diarrhea or constipation or both), but no detectable pathologic change; there may be spasms of the intestinal muscles. A variant form is characterized by painless diarrhea. It is usually due to a combination of psychologic and physiologic factors. Called also [irritable](#) or [spastic colon](#).

Irukandji syndrome, a clinical syndrome observed in Queensland, Australia, due to stinging by the jellyfish [Carukia barnesi](#); symptoms include initial neuromuscular paralysis that can be fatal, and in survivors systemic symptoms with pulmonary edema and skin ulcers at the site of infection.

Isaacs syndrome, **Isaacs-Mertens syndrome**, a rare autoimmune form of peripheral nerve hyperexcitability that affects the potassium channels of motor nerve axons, resulting in abnormal nerve firing and consequent spontaneous muscle activity, characterized by progressive muscle stiffness, delayed muscle relaxation after contraction, cramping, myokymia, and hyperhidrosis. It may accompany a variety of other autoimmune disorders; it sometimes occurs as a paraneoplastic condition or secondary to infection. Called also [acquired neuromyotonia](#).

Ivemark syndrome, congenital splenic agenesis, cardiac defects, and partial situs inversus viscerum; called also [asplenia s.](#) and [Polhemus-Schafer-Ivemark s.](#)

IVIC syndrome, a rare autosomal dominant syndrome of internal ophthalmoplegia, hearing impairment, and radial ray defects varying from a long slender thumb to deformity of an entire upper limb, first observed in Venezuela and later in Italy. Called also [oculo-oto-radial s.](#)

Jaccoud syndrome, see under [arthropathy](#).

Jackson syndrome, paralysis of structures innervated by the tenth, eleventh, and twelfth cranial nerves, including the soft palate, larynx, half of the tongue, and the sternomastoid and trapezius muscles. Called also [Mackenzie s.](#) and [vagoaccessory-hypoglossal s.](#)

Jacod syndrome, unilateral blindness and ophthalmoplegia with facial hemiplegia or trigeminal neuralgia as a result of damage to the second, third, fourth, fifth, and sixth cranial nerves, often from a tumor or other lesion just behind the sphenoid bone. Called also [Jacod triad](#), [Negri-Jacod s.](#), and [petrosphenoid s.](#)

Jadassohn-Lewandowsky syndrome, [pachyonychia congenita](#).

Jahnke syndrome, a variant of [Sturge-Weber syndrome](#) in which glaucoma is absent.

Janz syndrome, [juvenile myoclonic epilepsy](#).

Jarcho-Levin syndrome, an autosomal recessive disorder consisting of multiple vertebral defects, short thorax, rib abnormalities, camptodactyly, and syndactyly; urogenital abnormalities are sometimes present. Death, from respiratory insufficiency, usually occurs in infancy. Called also [spondylothoracic dysplasia](#).

jaw-winking syndrome, [Gunn s.](#)

Jefferson syndrome, [cavernous sinus s.](#)

jejunal syndrome, [dumping s.](#)

Jervell and Lange-Nielsen syndrome, a rare, autosomal recessive form of the [long QT syndrome](#), characterized by [neural hearing loss](#) and [syncope](#), sometimes with [ventricular fibrillation](#) and sudden death.

Jeune syndrome, [asphyxiating thoracic dystrophy](#).

Job syndrome, [hyperimmunoglobulinemia E s.](#)

Johanson-Blizzard syndrome, a rare autosomal recessive syndrome characterized by a small, pointed nose with underdeveloped nostrils; malformed or absent teeth; sparse, coarse hair; and sometimes growth retardation or [failure to thrive](#).

Joubert syndrome, an autosomal recessive syndrome consisting of partial or complete agenesis of the cerebellar vermis, with hypotonia, episodic hyperpnea, mental retardation, and abnormal eye movements; most patients die in infancy.

jugular foramen syndrome, [Vernet s.](#)

jumping Frenchmen of Maine syndrome, a form of [jumping disease](#) observed in a group of lumbermen of French-Canadian descent

working in a remote area of Maine; affected individuals had exaggerated startle responses, automatic obedience, and often echolalia. It is believed to have represented a form of operant conditioning rather than a true disease.

juvenile polyposis syndrome, [juvenile polyposis](#).

Kabuki make-up syndrome, a congenital, possibly inherited, syndrome of mental retardation, dwarfism, scoliosis, peculiar facies resembling the makeup of Japanese actors of Kabuki, and frequently cardiovascular abnormalities.

Kallmann syndrome, a type of [hypogonadotropic hypogonadism](#) caused by failure of fetal [gonadotropin-releasing hormone](#) neurons to migrate to the thalamus, usually associated with [anosmia](#) or [hyposmia](#). It is usually passed by autosomal recessive inheritance, and some cases are X-linked.

Kanner syndrome, [autistic disorder](#).

Kartagener syndrome, a hereditary disorder involving a combination of dextrocardia (situs inversus), bronchiectasis, and sinusitis, transmitted as an autosomal recessive trait.

Kasabach-Merritt syndrome, a blood disorder usually occurring in the first few months of life in which severe thrombocytopenia and other evidence of intravascular coagulation are accompanied by rapidly expanding hemangiomas of the trunk, extremities, and abdominal viscera, sometimes associated with bleeding and anemia. Bleeding is thought to be due to trapping and destruction of platelets within the tumor and depletion of circulating clotting factors. Called also [hemangioma-thrombocytopenia s.](#)

Kast syndrome, [Maffucci s.](#)

Kaufman-McKusick syndrome, a rare autosomal recessive disorder of hydrometrocolpos accompanied by postaxial polydactyly, congenital cardiac defects, and sometimes subsequent bilateral hydronephrosis. Manifestations in males include hypospadias and prominent scrotal raphe. Called also [McKusick-Kaufman s.](#)

Kearns-Sayre syndrome, progressive ophthalmoplegia, pigmentary degeneration of the retina, myopathy, ataxia, and cardiac conduction defect; onset is before age 20. Almost all patients have large mitochondrial DNA deletions, and ragged red fibers are seen on muscle biopsy. Called also [ophthalmoplegia plus](#).

Kennedy syndrome, retrobulbar optic neuritis, central scotoma, optic atrophy on the side of the lesion and papilledema on the opposite side, occurring in tumors of the frontal lobe of the brain which press downward. Called also [Foster Kennedy s.](#)

Kenny-Caffey syndrome, a hereditary syndrome characterized by hypoparathyroidism with intermittent hypocalcemia, short stature, and medullary stenosis of tubular bones. It is usually autosomal dominant, but autosomal recessive cases have also been observed.

keratitis-ichthyosis-deafness syndrome, a rare syndrome, often inherited in autosomal recessive fashion, consisting of [erythroderma](#), [lamellar ichthyosis](#), [sensorineural hearing loss](#), and sometimes [keratitis](#), inflammatory corneal vascularization, postnatal growth deficiency, [alopecia](#), nail dystrophy, tooth malformations, and decreased sweating. Called also [KID s.](#) and [Senter s.](#)

Key-Gaskell syndrome, [feline dysautonomia](#).

KID syndrome, [keratitis-ichthyosis-deafness s.](#)

Kiloh-Nevin syndrome, ocular myopathy in patients with ptosis and progressive external ophthalmoplegia. [anterior interosseous s.](#)

Kimmelstiel-Wilson syndrome, the nodular type of [intercapillary glomerulosclerosis](#).

Kindler syndrome, a rare type of [genodermatosis](#) of variable inheritance, characterized by [acrokeratosis](#) with blistering, [photosensitivity](#), and [poikiloderma](#). Called also [bullous](#) or [hereditary acrokeratotic poikiloderma](#).

King syndrome, a form of [malignant hyperthermia](#) (q.v.) in which patients also exhibit characteristic physical abnormalities including short stature, characteristic facies, kyphoscoliosis, pectus carinatum, cryptorchidism, delayed motor development, progressive myopathy, and cardiovascular structural defects.

kinky hair syndrome, [Menkes disease](#)

Kinsbourne syndrome, [myoclonic encephalopathy of childhood](#); see under encephalopathy.

kleeblatzschädel syndrome, a congenital disorder, characterized by synostosis of multiple or all cranial sutures, hydrocephalus, and in some cases facial dysostosis and long bone anomalies.

Klein-Waardenburg syndrome, [Waardenburg s. type 3](#).

Kleine-Levin syndrome, episodic periods of excessive sleep and overeating lasting for several weeks, with amnesia for the attacks; it usually occurs in adolescent boys.

Klinefelter syndrome, a syndrome in males characterized by small testes, hyalinization of [seminiferous tubules](#), [azoospermia](#), variable degrees of [masculinization](#), and increased urinary excretion of [gonadotropin](#). Patients tend to be tall, with long legs, and about half have [gynecomastia](#). It is associated typically with an XXY chromosome complement, although variants include XXYY, XXXY, XXXXY, and several mosaic patterns. Called also [seminiferous tubule dysgenesis](#) and [XXY s.](#)

Klippel-Feil syndrome, a condition characterized by shortness of the neck resulting from reduction in the number of cervical vertebrae or the fusion of multiple hemivertebrae into one osseous mass; the hairline is low and motion of the neck is limited.



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Klippel-Feil syndrome. Radiograph shows severe osseous involvement with fusion and hypoplasia of spinal segments C3–C7.

Klippel-Trénaunay syndrome, **Klippel-Trénaunay-Weber syndrome**, a rare condition usually affecting one extremity, characterized by hypertrophy of the bone and related soft tissues, large cutaneous hemangiomas, persistent nevus flammeus (see [port-wine stain](#), under stain), and skin varices.

Klumpke-Dejerine syndrome, [Klumpke paralysis](#).

Klüver-Bucy syndrome, bizarre behavior disturbances seen in monkeys following experimental bilateral temporal lobectomy which destroys important limbic structures; reported in humans after large injuries, usually from trauma, affecting the undersurface of the anterior temporal lobes. It is characterized by a tendency to examine objects orally, depression of drive and emotional reactions, hypermetamorphosis, and lack of sexual inhibitions.

Kocher-Debré-Sémélaigne syndrome, [Debré-Sémélaigne s.](#)

Koerber-Salus-Elschnig syndrome, [sylvian s.](#)

König syndrome, constipation alternating with diarrhea and attended with abdominal pain, [tympanites](#), and gurgling sounds in the right iliac fossa.

Korsakoff syndrome, a syndrome of anterograde and retrograde amnesia with confabulation associated with alcoholic or nonalcoholic polyneuritis described as "cerebropathia psychica toxemica" by Korsakoff; currently used synonymously with the term amnesic syndrome or, more narrowly, to refer to the amnesic component of the [Wernicke-Korsakoff syndrome](#), i.e., an [amnesic syndrome](#) resulting from thiamine deficiency. Spelled also Korsakov s. Called also [Korsakoff psychosis](#).

Kostmann syndrome, an autosomal recessive form of severe congenital neutropenia (gene locus: 1q21.3), characterized by early onset of recurrent pyogenic infections of the skin and lung, absence of neutrophils in the blood, absolute monocytosis and eosinophilia, and early death. Called also [infantile genetic agranulocytosis](#) and [Kostmann neutropenia](#).

Krause syndrome, a retinal and cerebral dysplasia found in premature infants several months after birth, characterized by malformations of the choroid, retina, and optic nerve, and possible blindness, cataract, coloboma, glaucoma, and microphthalmos. Cerebral symptoms include aplasia, hyperplasia, and hypertrophy of the brain, hydrocephaly, microcephaly, and mental retardation. Called also [encephalo-ophthalmic dysplasia](#).

Kugelberg-Welander syndrome, type III [spinal muscular atrophy](#) (see under atrophy), an autosomal recessive condition caused by lesions of the anterior horns of the spinal cord. Onset is in the first or second decade, principally between two and seventeen years, with atrophy and weakness of the proximal muscles of the lower limbs and pelvic girdle, followed by involvement of the distal muscles and muscular twitching. Called also [juvenile](#) or [proximal spinal muscular atrophy](#) and [Wohlfart-Kugelberg-Welander s.](#)

Kunkel syndrome, [chronic active hepatitis](#).

lacrimo-auriculo-dento-digital syndrome, **LADD syndrome**, a syndrome of autosomal dominant inheritance, characterized by abnormalities of the [nasolacrimal ducts](#), ears, limbs, and teeth, often with [mixed hearing loss](#). Abnormalities of the [salivary glands](#) and [urogenital system](#) may also occur. Spelled also lacrimoauriculodentodigital s. Called also [Levy-Hollister s.](#)

lacunar syndrome, see under [stroke](#).

Ladd syndrome, congenital obstruction of the duodenum due to peritoneal bands ([Ladd bands](#)) and [cecal volvulus](#).

LAMB syndrome, a syndrome of familial myxomas with cutaneous, cardiac, and endocrine involvement, manifested as lentiginos, atrial myxoma, and blue nevi. Cf. [NAME s.](#)

Lambert-Eaton syndrome, **Lambert-Eaton myasthenic syndrome**, an autoimmune, myasthenialike syndrome caused by autoantibodies to the voltage-gated calcium channel (alpha 1 antibodies) that interfere with the release of acetylcholine at the motor nerve terminal. Weakness usually affects the limbs, but ocular and bulbar muscles are spared; there is reduced muscle action potential on stimulation of its nerve but with repetitive stimulation it becomes augmented. It is often associated with oat-cell carcinoma of the lung. Called also [Eaton-Lambert s.](#) and [myasthenic s.](#)

Lance-Adams syndrome, the chronic form of [posthypoxic myoclonus](#).

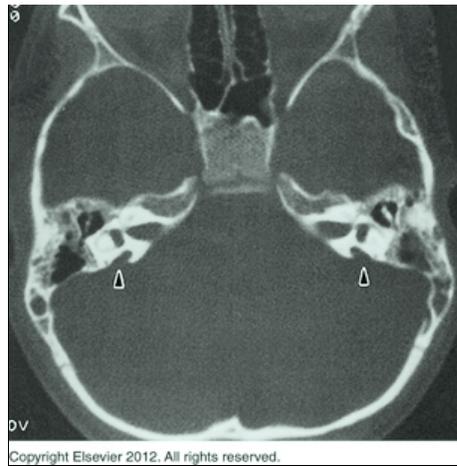
Landau-Kleffner syndrome, an epileptic syndrome of childhood characterized by partial or generalized seizures, psychomotor abnormalities, and aphasia progressing to mutism. The electroencephalogram from bilateral temporal regions is abnormal, with spikes like those of benign rolandic epilepsy. Called also [acquired epileptic aphasia](#).

Landry syndrome, [Guillain-Barré s.](#)

Langer-Giedion syndrome, an inherited disorder characterized by mental retardation, microcephaly, multiple exostosis, characteristic facies with bulbous nose, sparse hair, cone-shaped epiphyses, loose redundant skin, joint laxity, and other anomalies.

Lannois-Gradenigo syndrome, [Gradenigo s.](#)

large vestibular aqueduct syndrome, symptoms resulting from enlargement of the vestibular aqueduct of the inner ear, with sensorineural hearing loss that may be progressive and become severe, and frequently vertigo.



Large vestibular aqueduct syndrome showing bilateral enlargement of the vestibular aqueducts (arrows) on an axial CT scan.

Laron syndrome, an autosomal recessive syndrome of skeletal growth retardation due to impaired inability to synthesize [insulin-like growth factor I](#), usually because of growth hormone receptor defects. Called also [Laron dwarfism](#).

Larsen syndrome, a rare syndrome of variable inheritance, characterized by cleft palate, flattened facies, multiple congenital bone dislocations, and foot deformities.

lateral medullary syndrome, [Wallenberg s.](#)

Launois syndrome, [pituitary gigantism](#).

Launois-Bensaude syndrome, [multiple symmetric lipomatosis](#).

Laurence-Moon syndrome, an autosomal recessive disorder characterized by mental retardation, pigmentary retinopathy, hypogonadism, and spastic paraplegia; cf. [Bardet-Biedl s.](#) and [Biemond s., II](#).

Lawrence-Seip syndrome, [total lipodystrophy](#).

lazy leukocyte syndrome, a syndrome of unknown etiology, usually seen in children, marked by recurrent low-grade infections, associated with a defect in neutrophil chemotaxis and deficient random mobility of neutrophils.

Legg-Calvé-Perthes syndrome, osteochondrosis of the capital epiphysis.

Lemierre syndrome, thrombophlebitis of the internal jugular vein with secondary spread of infection, resulting from an acute oropharyngeal infection. Called also [postanginal sepsis](#).

Lemieux-Neemeh syndrome, an autosomal dominant syndrome consisting of Charcot-Marie-Tooth disease with progressive deafness.

Lennox syndrome, Lennox-Gastaut syndrome, an atypical form of [absence epilepsy](#) characterized by diffuse slow spike waves, often with atonic, tonic, or clonic seizures and mental retardation; there may also be other neurological abnormalities or multiple seizure types. Unlike typical [absence epilepsy](#), it may persist into adulthood. Called also [petit mal variant](#).

lentiginosis profusa syndrome, LEOPARD syndrome.

Lenz syndrome, a hereditary syndrome, transmitted as an X-linked trait, consisting of microphthalmia or anophthalmos, unilateral or bilateral, and digital anomalies; narrow shoulders, double thumbs, and other skeletal abnormalities; dental, urogenital, and cardiovascular defects may also occur.

LEOPARD syndrome, an autosomal dominant syndrome consisting of multiple lentigines, asymptomatic electrocardiographic abnormalities, and often ocular hypertelorism, pulmonary stenosis, abnormal genitalia, growth retardation, and sensorineural deafness. Patients have characteristic coarse facies. Called also [lentiginosis profusa](#), [lentiginosis profusa s.](#), and [multiple lentiginos s.](#)

Leredde syndrome, severe dyspnea on exertion, combined with advanced emphysema and recurrent attacks of acute febrile bronchitis; seen in children with congenital syphilis.

Léri-Weill syndrome, see under [dyschondrosteosis](#).

Leriche syndrome, a syndrome caused by obstruction of the terminal aorta, usually occurring in males and characterized by fatigue in the hips, thighs, or calves on exercising, absence of pulsation in the femoral arteries, and impotence, and often pallor and coldness of the lower extremities.

Lermoyez syndrome, tinnitus and hearing loss preceding an attack of vertigo and then subsiding after the vertigo has become established. Cf. [Meniere disease](#).

Lesch-Nyhan syndrome, a rare X-linked disorder of purine metabolism due to deficient hypoxanthine phosphoribosyltransferase, characterized by physical and mental retardation, compulsive self-mutilation of the fingers and lips by biting, choreoathetosis, spastic cerebral palsy, impaired renal function; and by excessive purine synthesis and consequent hyperuricemia and uricaciduria.

lethal multiple pterygium syndrome, a lethal autosomal recessive disorder caused by mutations in any of several genes that encode acetylcholine receptor components, characterized by multiple pterygia, lung hypoplasia, flexion contractures of the limbs, characteristic facies, and other abnormalities.

levator syndrome, levator ani syndrome, a functional pain syndrome occurring chiefly in women under 45 years of age and consisting of chronic or recurrent episodes of vague, dull aching or pressure high in the rectum that last at least 20 minutes; the pain is often worse when sitting. It is attributed to spasm of the levator muscles. The term is often used interchangeably with [proctalgia fugax](#) (q.v.). Called also [pelvic tension myalgia](#), [levator spasm](#), and [piriformis or puborectalis s.](#)

Levy-Hollister syndrome, lacrimo-auriculo-dento-digital s.

Lévy-Roussy syndrome, Roussy-Lévy s.

Lewis-Sumner syndrome, an immune-mediated multifocal neuropathy characterized by conduction blocks of sensory and motor nerves in asymmetric locations in the body, usually one upper limb, with sensory deficits, weakness, and wasting.

Leyden-Möbius syndrome, limb-girdle muscular dystrophy; see under dystrophy.

Li-Fraumeni syndrome, an autosomal dominant cancer syndrome with early onset of tumors, multiple tumors in individuals, and multiple affected family members; the most common tumors include soft tissue sarcomas and osteosarcomas, breast cancer, brain tumors, leukemia, and adrenocortical carcinoma. Most cases are caused by mutation in the [p53 tumor suppressor gene](#).

Lichtheim syndrome, subacute combined degeneration of spinal cord; see under degeneration.

licking syndrome, a form of pica in cattle in which they lick their own or each other's hair and skin, or other surfaces; it is often due to dietary deficiency of copper or sodium.

Liddle syndrome, a rare autosomal dominant syndrome resulting from [epithelial sodium channel](#) mutations that lead to abnormally increased channel function, characterized by hypertension with excessive renal reabsorption of sodium, depletion of potassium, and low activity of [renin](#) and [aldosterone](#). Cf. [pseudoprimary aldosteronism](#).

Lightwood syndrome, a transient, nonhereditary type of [distal renal tubular acidosis](#) seen in infant boys.

Lignac syndrome, Lignac-Fanconi syndrome, Fanconi s. (def. 2), cystinosis.

linear nevus sebaceous syndrome, linear sebaceous nevus syndrome, sebaceous nevus s.

lissencephaly syndrome, Miller-Dieker s.

Lobstein syndrome, osteogenesis imperfecta, type I.

locked-in syndrome, quadriplegia and mutism with intact consciousness and the preservation of voluntary vertical eye movements and blinking; usually due to a vascular lesion of the pars ventralis pontis. Called also [coma vigil](#), [de-efferented state](#), and [pseudocoma](#). Cf. [akineti mutism](#).

loculation syndrome, Froin s.

Löffler syndrome, transient infiltrations and [eosinophilia](#) of the lungs, accompanied by cough, fever, and dyspnea; it may be idiopathic or due to infestation by parasites (particularly [Ascaris lumbricoides](#)), infection, or drug therapy. See also [Ascaris pneumonitis](#), under pneumonitis. Called also [Löffler eosinophilia](#) or [pneumonia](#) and [simple pulmonary eosinophilia](#).

Löfgren syndrome, erythema nodosum in conjunction with bilateral adenopathy of hilar lymph nodes, seen as a manifestation of [sarcoidosis](#).

loin pain-hematuria syndrome, a syndrome of intense loin pain, either unilateral or bilateral, lasting from a few days to weeks, followed by [hematuria](#), usually seen in young women; the etiology is unknown, but some cases have been linked to treatment with [estrogen](#) compounds.

long QT syndrome, prolongation of the [Q-T interval](#) combined with [torsades de pointes](#), one of the most common types of [ion channelopathy](#). Acquired forms are usually due to a metabolic or cardiac abnormality or to drug administration. Congenital forms result from a variety of mutations in genes coding for channel subunits and are usually noted early in life. The condition may lead to serious arrhythmia and [sudden cardiac death](#) ([sudden arrhythmia death syndrome](#)).



Quinidine-induced long QT syndrome. Patient on quinidine (monitor lead) developed marked prolongation of repolarization with low amplitude T-U waves. Cardiac arrest with torsades de pointes ventricular tachycardia later developed.

loose anagen hair syndrome, a syndrome of unknown etiology, usually seen in children, in which scalp hair can be plucked easily and painlessly during the [anagen](#) part of the [hair cycle](#), owing to defective anchorage of the hair shaft to the follicle; there is also slowing of hair growth.

Looser-Milkman syndrome, Milkman s.

Lorain-Lévi syndrome, pituitary dwarfism.

Louis-Bar syndrome, ataxia-telangiectasia.

low cardiac output syndrome, the clinical manifestations of [low-output heart failure](#) (see under failure). Called also [low output s.](#)

Low syndrome, Lowe-Terrey-MacLachlan syndrome, oculocerebrorenal s.
lower radicular syndrome, Klumpke paralysis.
Lown-Ganong-Levine syndrome, a preexcitation syndrome of electrocardiographic abnormality characterized by a short P–R interval with a normal QRS complex, accompanied by atrial tachycardia.
low output syndrome, low cardiac output s.
low T3 syndrome, euthyroid sick s.
Lucey-Driscoll syndrome, a syndrome of retention jaundice due to defective bilirubin conjugation, occurring in infants; apparently the result of an unidentified factor, presumably a steroid in maternal blood, transmitted to the infant.
lupus-like syndrome, see [systemic lupus erythematosus](#), under lupus.
Lutembacher syndrome, atrial septal defect associated with mitral stenosis. Called also [Lutembacher complex](#).
Lyell syndrome, toxic epidermal necrolysis.
lymphadenopathy syndrome, a condition seen in [immunocompromised](#) persons, characterized by unexplained [lymphadenopathy](#) of extralingual sites that lasts for three or more months and on biopsy reveals nonspecific lymphoid hyperplasia; in some patients it may be a prodrome of [acquired immunodeficiency syndrome](#).
lymphoproliferative syndromes, see under [disorder](#).
lymphoreticular syndromes, see under [disorder](#).
Lynch syndrome, hereditary nonpolyposis colorectal cancer.
McCune-Albright syndrome, Albright s.
Mackenzie syndrome, Jackson s.
McKusick-Kaufman syndrome, Kaufman-McKusick s.
McLeod syndrome, a syndrome seen in some individuals having the [McLeod phenotype](#) of blood, characterized by mild hemolytic anemia with acanthocytes, elevated serum creatinine phosphokinase, and sometimes muscle wasting and neurological defects. A few cases have manifested as X-linked types of [chronic granulomatous disease](#).
Macleod syndrome, Swyer-James s.
macrophage activation syndrome, hemophagocytic lymphohistiocytosis.
Maffucci syndrome, enchondromatosis associated with multiple cutaneous or visceral hemangiomas. Called also [Kast s.](#)
magenblase syndrome, excessive gas in the stomach, owing to habitual taking in of air with the food.
MAGIC syndrome, an inflammatory syndrome of mouth and genital ulcers with inflamed cartilage; a term encompassing overlapping features of Behçet syndrome and relapsing polychondritis.
malabsorption syndrome, any of a group of disorders characterized by subnormal absorption of dietary constituents, and thus excessive loss of nonabsorbed substances in the feces; the malabsorption may be due to an intraluminal (digestive) defect (e.g., pancreatic insufficiency), a mucosal abnormality ([celiac disease](#) or [disaccharidase deficiency](#)), or a lymphatic obstruction ([intestinal lymphangiectasia](#)). Unless there is a specific enzyme or transport defect, [steatorrhea](#) is usually present. Deficiency syndromes may result from excessive loss of vitamins, electrolytes, iron, calcium, or other substances.
malarial hyperreactive spleen syndrome, tropical splenomegaly s.
Mallory-Weiss syndrome, hematemesis or [melena](#) that follows typically upon many hours or days of severe vomiting and retching, traceable to one or several slitlike lacerations of the gastric mucosa, longitudinally placed at or slightly below the [esophagogastric junction](#).
manic syndrome, mania.
man-in-the-barrel syndrome, brachial diplegia of proximal areas of the upper limbs, usually owing to an infarction in the blood supply to the area, giving the appearance of having the arms and trunk constrained by a barrel; it often begins with weakness and numbness and can progress to become life threatening.
Marchesani syndrome, Weill-Marchesani s.
Marchiafava-Micheli syndrome, paroxysmal nocturnal hemoglobinuria.
Marcus Gunn syndrome, Gunn s.
Marfan syndrome, one of the manifestations of abnormal [fibrillin](#) metabolism, a congenital disorder of connective tissue characterized by abnormal length of extremities, especially fingers and toes, subluxation of the lens, cardiovascular abnormalities (commonly dilatation of the ascending aorta), and other deformities. It is an autosomal dominant disorder with variable degrees of expression, caused by mutations in the FBN1 gene, which encodes [fibrillin-1](#).



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 Marfan syndrome in a young man. (A), Long, thin fingers, with clubbing due to associated cardiopulmonary problems; (B), pectus carinatum, kyphosis, and joint contractures.

Marie-Bamberger syndrome, hypertrophic pulmonary osteoarthropathy.
Marinesco-Sjögren syndrome, a hereditary syndrome transmitted as an autosomal recessive trait, consisting of cerebellar ataxia, mental and somatic growth retardation, congenital cataracts, inability to chew, thin brittle fingernails, and sparse, incompletely keratinized hair.
Maroteaux-Lamy syndrome, a mucopolysaccharidosis caused by deficiency of N-acetylgalactosamine-4-sulfatase (arylsulfatase B), and characterized biochemically by the predominance of dermatan sulfate in the urine and the presence of coarse metachromatic granules in the leukocytes, and clinically by Hurler-like signs with normal intelligence. There are three clinical forms: the severe or classic form shows Hurler-like symptoms; the intermediate form has the same phenotype as mucopolipidosis III (pseudo-Hurler polydystrophy); the mild form is difficult to distinguish from the Scheie syndrome. Called also [mucopolysaccharidosis VI](#) and [arylsulfatase B \(ARSB\) deficiency](#).
marrara syndrome, halzoun.
Marshall syndrome, a rare, autosomal dominant syndrome characterized by ectodermal dysplasia, hypertelorism, myopia progressing to retinal detachment and blindness, and degenerative changes in the joints; sensorineural deafness may also occur. It resembles hereditary

progressive arthro-ophthalmopathy, but has been found to be caused by a different genetic defect.

Martin-Bell syndrome, [fragile X s.](#)

Martorell syndrome, [Takayasu arteritis](#).

mastocytosis syndrome, an episodic syndrome occurring in certain patients with systemic mastocytosis, usually those with skin lesions, bone lesions, and hepatosplenomegaly, presumably associated with histamine release from degranulation of mast cells, and characterized mainly by intense pruritus, flushing, headache, tachycardia, hypotension, and syncope.

maternal deprivation syndrome, failure to thrive with severe growth retardation (deprivation dwarfism), unresponsiveness to the environment, depression, retarded mental and emotional development, and behavioral problems resulting from loss of, absence of, or neglect by the mother or other primary caregiver.

Mauriac syndrome, dwarfism, hepatomegaly, obesity, and retarded sexual maturation, in association with diabetes mellitus.

May-Thurner syndrome, deep vein thrombosis of the left lower limb resulting from compression of the left common iliac vein between the right common iliac artery and the fifth lumbar vertebra, characterized by severe pain and swelling of the limb and venous claudication.

May-White syndrome, a rare autosomal dominant syndrome of myoclonus, cerebellar ataxia, and deafness.

Mayer-Rokitansky-Küster-Hauser syndrome, lack of müllerian development, congenital absence of the vagina, and a rudimentary uterus (typically bicornuate remnants), with normal uterine tubes, ovaries, and secondary female sex characteristics and normal growth. Called also [Rokitansky-Küster-Hauser s.](#)

Mazabraud syndrome, a syndrome of fibrous dysplasia associated with soft tissue myxomas.

Meckel syndrome, **Meckel-Gruber syndrome**, a hereditary syndrome, transmitted as an autosomal recessive trait, usually characterized by sloping forehead, posterior meningoencephalocele, polydactyly, and polycystic kidneys, with death occurring in the perinatal period. Called also [Gruber s.](#) and [dysencephalia splanchnocystica](#).

meconium aspiration syndrome, the respiratory complications resulting from the passage and aspiration of [meconium](#) prior to or during delivery. Postterm infants and those who had [hypoxia](#) or [acidosis](#) in utero are at higher risk.

meconium plug syndrome, [intestinal obstruction](#) in an infant due to unusually thick or hard [meconium](#) in which neither enzymatic nor ganglion cell deficiency can be demonstrated, most often seen in premature infants.

median arcuate ligament syndrome, [celiac artery compression syndrome](#).

median cleft facial syndrome, a hereditary form of defective midline development of the head and face, including ocular hypertelorism, occult cleft nose and maxilla, and sometimes mental retardation or other defects. Called also [frontonasal dysplasia](#).

megacystis-megaureter syndrome, chronic ureteral dilatation ([megaureter](#)) associated with hypotonia and dilatation of the bladder ([megacystis](#)) and gaping of ureteral orifices, permitting [vesicoureteral reflux](#) of urine, and resulting in chronic [pyelonephritis](#). Called also [megaureter-megacystis s.](#)

megacystis-microcolon-intestinal hypoperistalsis syndrome, a congenital syndrome with a female preponderance, usually autosomal recessive and fatal in the first year of life, consisting of an enlarged bladder ([megacystis](#)) and small colon with decreased or absent peristalsis ([microcolon](#) and intestinal [hypoperistalsis](#)); patients also have the abdominal muscle defect seen in the [prune-belly syndrome](#). Called also [Berdon s.](#) and [MMIH s.](#)

megaureter-megacystis syndrome, [megacystis-megaureter s.](#)

Meige syndrome, [focal dystonia](#) of facial and oromandibular muscles with [blepharospasm](#), grimacing mouth movements, and protrusion of the tongue, usually occurring in older women. Called also [Brueghel s.](#)

Meigs syndrome, **Meigs-Salmon syndrome**, ascites and hydrothorax associated with ovarian fibroma or other pelvic tumor.

MELAS syndrome, mitochondrial encephalopathy, lactic acidosis, and stroke-like episodes; a familial syndrome, of maternal (mitochondrial) inheritance.

Melkersson syndrome, **Melkersson-Rosenthal syndrome**, an autosomal dominant condition usually beginning in childhood or adolescence, characterized chiefly by chronic noninflammatory facial swelling, usually confined to the lips in the form of [granulomatous cheilitis](#), with recurrent [facial palsy](#) and sometimes [fissured tongue](#). Associated ophthalmic symptoms may include [lagophthalmos](#), [blepharochalasis](#), swollen eyelids, burning sensation of the eyes, corneal opacities, [retrobulbar neuritis](#), and [exophthalmos](#).

Melnick-Fraser syndrome, [branchio-oto-renal s.](#)

Melnick-Needles syndrome, a rare X-linked disorder (gene locus: Xq28), caused by mutations in the gene encoding filamin A. It is characterized by severe congenital bone abnormalities manifested by distinctive facies (exophthalmos, full cheeks, micrognathia, and malalignment of the teeth), flaring of the metaphyses of long bones, S-like curvature of the bones of the lower limbs, irregular constrictions in the ribs, and sclerosis of the base of the skull. It is lethal in males, resulting in death in utero or during the first few months of life, but not in females. Called also [Melnick-Needles osteodysplasty](#).

Mendelson syndrome, [pulmonary acid aspiration s.](#)

Mendes da Costa syndrome, [erythrokeratoderma variabilis](#).

MERRF syndrome, a familial type of [mitochondrial encephalopathy](#) of maternal (mitochondrial) inheritance, characterized by [myoclonic epilepsy](#) and mitochondrial myopathy with [ragged red fibers](#), and also by abnormal brainstem evoked responses, sensorineural hearing loss, ataxia, renal dysfunction, diabetes, cardiomyopathy, and dementia. It is usually caused by mutation within the mitochondrial gene encoding the tRNA specific for lysine. Called also [Fukuhara s.](#)

metabolic syndrome, **metabolic syndrome X**, a combination including at least three of the following: abdominal obesity, [hypertriglyceridemia](#), low level of [high-density lipoproteins](#), [hypertension](#), and high [fasting plasma glucose](#) level. It is associated with an increased risk for development of [diabetes mellitus](#) and cardiovascular disease. Called also [s. X](#).

metameric syndrome, [segmentary s.](#)

methionine malabsorption syndrome, an autosomal recessive disorder of [methionine](#) absorption in which the urine has a characteristic odor resembling that of the interior of an oasthouse (room where tobacco, hops, and malt are dried). It is due to alpha-[hydroxybutyric acid](#) formed by bacterial action on the unabsorbed [methionine](#). Characteristics include white hair, mental retardation, convulsions, and attacks of hyperpnea. Called also [oasthouse urine disease](#) and [Smith-Strang disease](#).

Meyer-Schwickerath and Weyers syndrome, [oculodentodigital dysplasia](#).

Michelin tire syndrome, **Michelin tire baby syndrome**, a rare, autosomal dominant syndrome characterized by deep creases in the skin, particularly around the limbs, resembling those between the treads on a tire; it is usually most pronounced in infants.

MIDAS syndrome, [MLS s.](#)

middle lobe syndrome, lobar atelectasis in the right middle lobe of the lung, with chronic pneumonitis; called also [Brock s.](#)

mid-systolic click-late systolic murmur syndrome, [mitral valve prolapse s.](#)

Mikulicz syndrome, a chronic bilateral hypertrophy of the lacrimal, parotid, and salivary glands, associated with decreased or absent lacrimation and xerostomia, and often accompanied by chronic lymphocytic infiltration. It may be associated with other diseases, such as [Sjögren syndrome](#), [sarcoidosis](#), [lupus erythematosus](#), [leukemia](#), [lymphoma](#), and [tuberculosis](#). See also [benign lymphoepithelial lesion](#), under lesion.

mild androgen insensitivity syndrome, the least severe form of androgen insensitivity syndrome; affected males have normal male genitalia but may be infertile. Some develop gynecomastia and are at risk for breast cancer. Called also [minimal androgen insensitivity s.](#)

milk-alkali syndrome, a syndrome characterized by hypercalcemia without hypercalciuria or hypophosphatemia, with only mild alkalosis, normal serum phosphatase, severe renal insufficiency with hyperazotemia, and calcinosis, attributed to ingestion of milk and absorbable alkali for long periods of time. Called also [Burnett](#) or [hypercalcemia s.](#)

Milkman syndrome, a generalized bone disease marked by multiple transparent stripes of absorption in the long and flat bones; called also

[Looser-Milkman s.](#)

Millard-Gubler syndrome, crossed paralysis, affecting the limbs on one side of the body and the face on the opposite side, together with paralysis of outward movement of the eye; it is due to infarction of the pons, involving the sixth and seventh cranial nerves and the fibers of the corticospinal tract. Called also [Gubler hemiplegia](#) or [paralysis](#) and [Millard-Gubler paralysis](#). Cf. [Foville s.](#)

Miller syndrome, a syndrome of extensive facial and limb defects, characterized by malar hypoplasia, downslanting palpebral fissures, micrognathia, cleft lip and palate, cup-shaped ears, lower lid ectropion, postaxial limb deficiencies, and syndactyly. Less frequently present are heart defects and hearing loss. The syndrome is probably an autosomal recessive trait. Called also [postaxial acrofacial dysostosis](#).

Miller-Dieker syndrome, an autosomal recessive syndrome characterized by [lissencephaly](#), [microcephaly](#), mental retardation, dysmorphic facial appearance, and sometimes [polydactyly](#), [cryptorchidism](#), heart lesions, kidney defects, and defects of the gastrointestinal system. It is caused by hemizygous deletion of chromosome locus 17p13.3. Called also [lissencephaly s.](#)

Miller Fisher syndrome, [Fisher s.](#) (def. 1).[one-and-a-half s.](#)

Milwaukee shoulder syndrome, a type of crystal-induced arthropathy seen most often in older women, characterized by calcium phosphate crystals around the shoulder joint, often bilaterally, with pain that may be worse at night. Called also [apatite-associated destructive arthritis](#).

minimal androgen insensitivity syndrome, [mild androgen insensitivity s.](#)

minimal change nephrotic syndrome, [minimal change disease](#).

Minkowski-Chauffard syndrome, [hereditary spherocytosis](#).

Minot-von Willebrand syndrome, [von Willebrand disease](#).

Mirizzi syndrome, a rare syndrome of extrahepatic obstruction of the [common bile duct](#) owing to impaction of a [gallstone](#) in the [cystic duct](#); symptoms include recurring pain, fever, and jaundice.

mitral valve prolapse syndrome, prolapse of the mitral valve, often with regurgitation, associated with myxomatous proliferation of the leaflets of the mitral valve, a common, usually benign, often asymptomatic condition characterized by midsystolic clicks and late systolic murmurs on auscultation. Palpitations and chest discomfort may occur, and in some cases progressive mitral regurgitation necessitates valve replacement. Called also [Barlow s.](#), [click-murmur s.](#), [floppy valve s.](#), [MVP s.](#), and [systolic click-murmur s.](#)

MLS syndrome, [microphthalmia, linear skin defects] a rare X-linked syndrome of linear defects on the face and neck with [microphthalmia](#) and sometimes brain defects such as agenesis of the corpus callosum and mental retardation. Most patients are female because it is lethal for male fetuses. Called also [MIDAS s.](#)

MMIH syndrome, [megacystis-microcolon-intestinal hypoperistalsis s.](#)

Möbius syndrome, agenesis or aplasia of the motor nuclei of the cranial nerves characterized by congenital bilateral facial palsy in various combinations, with unilateral or bilateral paralysis of the abductors of the eye, sometimes associated with involvement of the cranial nerves, particularly the oculomotor, trigeminal, and hypoglossal, and anomalies of the extremities. Called also [nuclear agenesis](#) or [aplasia](#), [congenital facial diplegia](#), [congenital abducens-facial paralysis](#), and [congenital oculo-facial paralysis](#).

Mohr syndrome, an autosomal recessive disorder characterized by brachydactyly, clinodactyly, polydactyly, syndactyly, and bilateral hallucal polysyndactyly; by cranial, facial, lingual, palatal, and mandibular anomalies; and by episodic neuromuscular disturbances. Called also [orodigitofacial dysostosis](#), [oral-facial-digital s., type II](#), and [orofacioidigital s., type II](#). See also [oral-facial-digital s., type I](#).

Monakow syndrome, hemiplegia on the side opposite the lesion in occlusion of the anterior choroidal artery, sometimes with hemianesthesia and hemianopia.

monosomy 9p- syndrome, a rare chromosomal disorder in which a piece of the short arm of the ninth chromosome is broken and often lost. Symptoms include mental retardation, a triangular head with forward angulation of the frontal bone, and various other physical deformities.

Moore syndrome, [abdominal epilepsy](#).

Morel syndrome, [hyperostosis frontalis interna](#).

Morgagni-Adams-Stokes syndrome, [Adams-Stokes s.](#)

morning glory syndrome, a coloboma in which there is a funnel-shaped optic nerve head with a dot of whitish, fluffy material in the center, an elevated ring of pigment around the disk, and vessels radiating from the ring like spokes. Vision is severely affected.

Morquio syndrome, either of two biochemically distinct, but clinically nearly indistinguishable, forms of [mucopolysaccharidosis](#) characterized by excretion of keratan sulfate in the urine. Clinical features, affecting primarily the skeletal and secondarily the nervous system, include genu valgum, pectus carinatum, progressive platyspondyly, short neck and trunk, normal but broad-mouthed facies with spacing between the teeth, progressive deafness, and very mild corneal clouding. Intelligence is normal. The two enzymatic types are type A, caused by N-acetylgalactosamine-6-sulfatase deficiency; and type B, caused by β -galactosidase deficiency. Called also [mucopolysaccharidosis IV](#).

Morris syndrome, [complete androgen insensitivity s.](#)

Morton syndrome, a congenital insufficiency of the first metatarsal segment of the foot, characterized by metatarsalgia due to shortening or relaxation of the part.

Morvan syndrome, [syringomyelia](#). a manifestation of syringomyelia in which the subcutaneous tissues of the hands become thickened, edematous, soft, swollen, cyanotic, and cold (see [Marinesco succulent hand](#), under hand), associated with analgesic ulceration of the fingertips and paresthesia and atrophy of the hands and forearms.

Mosse syndrome, polycythemia vera with cirrhosis of the liver.

Mounier-Kuhn syndrome, [tracheobronchomegaly](#).

Mount syndrome, **Mount-Reback syndrome**, a rare autosomal dominant disorder characterized by paroxysmal attacks of choreoathetosis and dystonic movements with Kayser-Fleischer rings on the corneas. It appears in childhood or young adulthood and does not involve a change in consciousness. Called also [paroxysmal](#) or [familial paroxysmal choreoathetosis](#). See also [paroxysmal kinesigenic choreoathetosis](#).

Mowat-Wilson syndrome, an autosomal dominant complex developmental disorder caused by a microdeletion or mutation in the long arm of chromosome 2 (2q22). It is characterized by a typical facies; mental retardation; delayed motor development; epilepsy; and multiple congenital anomalies, including agenesis of the corpus callosum and genital, cardiac, and eye defects. Hirschsprung disease is often present.

Moynahan syndrome, multiple symmetric lentiginos, congenital mitral valve stenosis, dwarfism, genital hypoplasia, and mental retardation. Called also [progressive cardiomyopathic lentiginosis](#). a familial congenital syndrome consisting of delayed hair growth on the scalp, epilepsy, mental retardation, and unusual electroencephalogram.

Muckle-Wells syndrome, an autosomal dominant [autoinflammatory disease](#) in the cryopyrin-associated periodic syndromes subgroup, characterized by amyloidosis involving the kidneys and causing nephritis, recurrent urticaria, deafness, and pain in the extremities.

mucocutaneous lymph node syndrome, [Kawasaki disease](#).

mucosal neuroma syndrome, [multiple endocrine neoplasia, type 2B](#).

Muir-Torre syndrome, [Torre s.](#)

multiple endocrine deficiency syndrome, **multiple glandular deficiency syndrome**, primary failure of any combination of endocrine glands, including adrenals, thyroid, gonads, parathyroids, and endocrine pancreas, often accompanied by nonendocrine autoimmune abnormalities.

multiple hamartoma syndrome, [Cowden disease](#).

multiple lentiginos syndrome, [LEOPARD s.](#)

multiple pterygium syndrome, an autosomal recessive syndrome caused by mutations in the CHRNG gene (locus: 2q33-q34), which encodes an acetylcholine receptor subunit. It is characterized by [pterygia](#) of the neck, axillae, and popliteal, antecubital, and intercrural areas, accompanied by hypertelorism, cleft palate, micrognathia, ptosis of eyelids, and short stature. Skeletal abnormalities include camptodactyly, syndactyly, equinovarus, and rocker-bottom feet, as well as vertebral fusion and rib anomalies. Cryptorchidism is present in males and labia majora are absent in females. Called also [Escobar s.](#) and [pterygium s.](#) Cf. [lethal multiple pterygium s.](#) an autosomal dominant syndrome that in

its most severe forms resembles the recessive disorder; milder cases may present with contractures of the extremities and ptosis. Called also [distal arthrogryposis type 8](#).

Munchausen syndrome, a condition characterized by habitual presentation for hospital treatment of an apparent acute physical illness, the patient giving a plausible and dramatic history, all of which is false; it is a subtype of [factitious disorder](#) (q.v.).

Munchausen syndrome by proxy, see [factitious disorder by proxy](#), under disorder.

MVP syndrome, [mitral valve prolapse s.](#)

myasthenic syndrome, [Lambert-Eaton myasthenic s.](#)

myelodysplastic syndrome, any of a group of related bone marrow disorders of varying duration preceding the development of overt [acute myelogenous leukemia](#); they are characterized by abnormal [hematopoietic stem cells](#), [anemia](#), [neutropenia](#), and [thrombocytopenia](#).

[Splenomegaly](#), [hepatomegaly](#), and [lymphadenopathy](#) may not occur until the onset of leukemia, which is sometimes sudden and fulminating. Called also [preleukemia](#).

myelofibrosis-osteosclerosis syndrome, [agnogenic myeloid metaplasia](#).

myeloproliferative syndromes, see under [disorder](#).

myoneuropathic syndrome, myoglobinuria, hemodynamic instability, lactic acidosis, and hyperkalemia that occur during reperfusion of the lower limb, caused by release of the products of muscle breakdown.

Naegeli syndrome, former name for [Franceschetti-Jadassohn s.](#)

Naffziger syndrome, [scalenus s.](#)

Nager syndrome, **Nager-de Reynier syndrome**, [Nager acrofacial dysostosis](#).

nail-patella syndrome, a hereditary, autosomal dominant syndrome consisting of dystrophy of the nails, absence or hypoplasia of the patella, hypoplasia of the lateral side of the elbow joint, bilateral iliac horns, and often thickening of the [lamina densa](#) of the renal glomeruli. Called also [hereditary osteo-onychodysplasia](#).

NAME syndrome, a syndrome of familial myxomas with cutaneous, cardiac, and endocrine involvement, manifested as nevi, atrial myxoma, and neurofibroma ephelides. Cf. [LAMB s.](#)

narcotic bowel syndrome, a group of intestinal symptoms seen in persons who use narcotic analgesics over a long period, including intestinal pseudoobstruction, abdominal pain, vomiting, and weight loss.

neck-tongue syndrome, pain in the neck, sometimes followed by numbness of the neck and tongue, on sudden turning of the head; it is thought to be due to compression of C2 nerve roots in the area of the atlantoaxial articulations because the C2 roots contain proprioceptive fibers from the tongue.

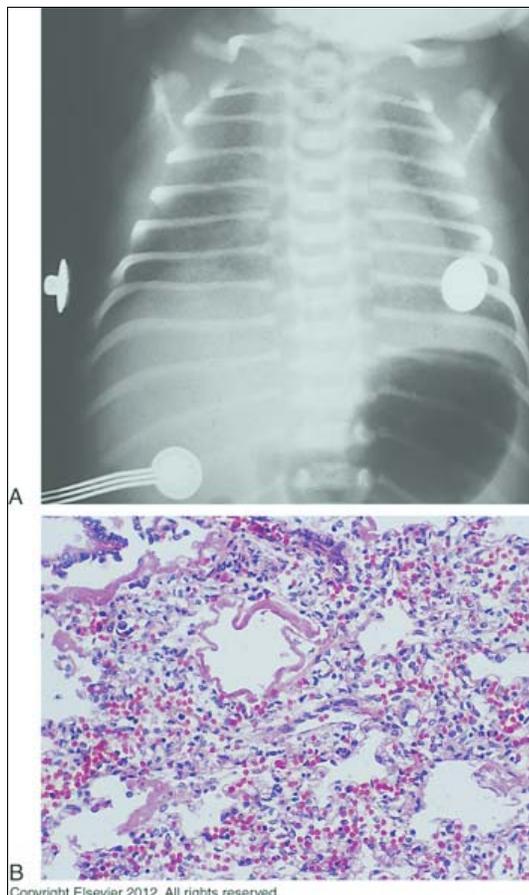
Negri-Jacod syndrome, [Jacod s.](#)

Nelson syndrome, the development of an ACTH-producing pituitary tumor after bilateral adrenalectomy for Cushing syndrome; it is characterized by aggressive growth of the tumor and hyperpigmentation of the skin.

neonatal abstinence syndrome, signs of [substance withdrawal](#), including tremor, sweating, yawning, poor feeding, sleep disturbance, and high-pitched cry, occurring in newborns exposed in utero to drugs taken by the mother.

neonatal maladjustment syndrome, a disease of newborn foals, caused by perinatal hypoxia and characterized by behavioral disturbances such as inability to nurse, aimless wandering, apparent blindness, and uttering of a barklike sound; it may progress to convulsions, coma, and death. Affected foals are called [barkers](#), [dummies](#), or [wanderers](#).

neonatal respiratory distress syndrome, [dyspnea](#) with [cyanosis](#) in the newborn, a condition caused by deficiency of [surfactant](#). Prodromal signs include dilatation of the alae nasi, expiratory grunt, and retraction of the suprasternal notch or costal margins. It is usually seen in infants who are premature, have diabetic mothers, or were delivered by cesarean section; sometimes there is no known cause. Some infants die of respiratory failure in the first few days of life and at autopsy have eosinophilic hyaline material lining the alveoli, alveolar ducts, and bronchioles. Called also [respiratory distress s. of newborn](#) and [idiopathic respiratory distress of newborn](#).



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Neonatal respiratory distress syndrome. (A), Ground-glass appearance and air bronchograms. (B), Alveoli are collapsed and the alveolar ducts and respiratory bronchioles are dilated and lined by hyaline membrane.

nephrogenic syndrome of inappropriate antidiuresis, a hyponatremic syndrome, clinically resembling the syndrome of inappropriate diuretic hormone, caused by inability of the renal collecting tubules to absorb water in response to antidiuretic hormone. Type I, which accounts for 90 percent of cases, is an X-linked syndrome caused by loss of vasopressin receptor function as a result of mutations in the gene encoding the receptors (locus: Xq28). Type II is caused by a mutation in the gene encoding aquaporin-2 (locus: 12q13); inheritance may be autosomal dominant or recessive.

nephrotic syndrome, general name for any of a large group of diseases involving defective [renal glomeruli](#), characterized by massive [proteinuria](#) and [lipiduria](#) with varying degrees of [edema](#), [hypoalbuminemia](#), and [hyperlipidemia](#). See also [glomerulonephropathy](#).

nerve compression syndrome, [entrapment neuropathy](#).

Netherton syndrome, a rare syndrome, believed to be autosomal recessive, consisting of [lamellar ichthyosis](#) or [ichthyosis linearis circumflexa](#), [trichorrhexis invaginata](#), [atopy](#), and sometimes mental retardation and [aminoaciduria](#).

Nettleship-Falls syndrome, see under [albinism](#).

neurocutaneous syndrome, [phakomatosis](#).

neuroleptic malignant syndrome, a rare, sometimes fatal reaction to antipsychotic (neuroleptic) agents, characterized by hyperthermia, rigidity, and coma.

nevoid basal cell carcinoma syndrome, nevoid basalioma syndrome, an autosomal dominant syndrome characterized by the development in early life of numerous [basal cell carcinomas](#), associated with abnormalities of the skin (especially an unusual erythematous pitting edema of the hands and feet), bones, nervous system, eyes, and reproductive tract. Called also [basal cell nevus s.](#), [Gorlin s.](#), and [Gorlin-Goltz s.](#)



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Nevoid basal cell carcinoma syndrome.

nevus sebaceus syndrome, [sebaceous nevus s.](#)

Nezelof syndrome, any of a heterogeneous group of immunodeficiency disorders characterized by profoundly deficient cellular immunity and varying degrees of humoral immunodeficiency. Immunoglobulin levels may be normal or increased, but antibody response to immunization may be absent. Patients are highly susceptible to life-threatening infections with low-grade or opportunistic pathogens, such as [Candida albicans](#), [Pneumocystis jiroveci](#), and [cytomegalovirus](#). Both autosomal recessive and X-linked inheritance have been described. Called also [cellular immunodeficiency with immunoglobulins](#).

Nicolau syndrome, a severe cutaneous reaction after intramuscular administration of medications, often in the gluteal region, because of ischemia from accidental arterial obliteration or vasculitis. Manifestations include severe local pain, swelling, livedoid changes, and increased skin temperature; there may also be fever, tachycardia, dyspnea, albuminuria, and eventually gangrene. Called also [livedoid dermatitis](#) and [embolia cutis medicamentosa](#).

Noack syndrome, [Pfeiffer s.](#)

noninsulinoma pancreatogenous hypoglycemia syndrome, a rare syndrome of endogenous hyperinsulinemia and hypoglycemia in the postprandial period, resembling symptoms of an insulinoma; some patients develop this following a Roux-en-Y bypass.

Nonne-Milroy-Meige syndrome, a name formerly given to hereditary lymphedema, now considered to be two disorders, [Milroy disease](#) and [lymphedema praecox](#).

nonsense syndrome, [Ganser s.](#)

nonstaphylococcal scalded skin syndrome, [toxic epidermal necrolysis](#).

Noonan syndrome, the phenotype of [Turner syndrome](#) (webbed neck, ptosis, hypogonadism, congenital heart disease, and short stature) without gonadal dysgenesis; formerly called [male Turner syndrome](#) until the female counterpart was identified. Called also [Ullrich-Turner s.](#)

Nothnagel syndrome, unilateral oculomotor paralysis combined with cerebellar ataxia, in lesions of the cerebral peduncles.

numb chin syndrome, paresthesias or loss of sensation in the lower face and chin, owing to compression of the mental nerve, usually by a tumor.

nutcracker syndrome, see under [phenomenon](#).

Nyssen-van Bogaert syndrome, the adult form of [metachromatic leukodystrophy](#).

obesity-hypoventilation syndrome, a complex of obesity, somnolence, hypoventilation, and erythrocytosis; called also [pickwickian s.](#)

obstructive sleep apnea syndrome, see under [apnea](#).

obstructive sleep apnea-hypopnea syndrome, [hypopneas](#) considered as one syndrome.

occipital horn syndrome, a mild allelic variant of Menkes disease, characterized by hyperelastic and bruisable skin, hernias, bladder diverticula and dysfunction, hyperextensible joints, varicosities, multiple skeletal abnormalities, and relatively normal intelligence.

Characteristic exostoses, resulting from calcification, occur at the sites of muscular attachment to the occipital skull. Called also [X-linked cutis laxa](#).

Ochoa syndrome, [urofacial s.](#)

oculocerebral-hypopigmentation syndrome, [Cross s.](#)

oculocerebrorenal syndrome, an X-linked disorder characterized by [vitamin D–refractory rickets](#), [hydrophthalmia](#), congenital glaucoma and cataracts, mental retardation, and tubule reabsorption dysfunction as evidenced by hypophosphatemia, acidosis, and aminoaciduria. Called also [Lowe disease](#) and [Lowe-Terrey-MacLachlan s.](#)

oculodentodigital syndrome, oculodento-osseous syndrome, see under [dysplasia](#).

oculoglandular syndrome, [Parinaud oculoglandular s.](#)

oculomandibulodyscephaly-hypotrichosis syndrome, oculomandibulofacial s.

oculomandibulofacial syndrome, a syndrome principally characterized by dyscephaly (usually brachycephaly), parrot nose, mandibular

hypoplasia, proportionate nanism, hypotrichosis, bilateral congenital cataracts, and microphthalmia. Called also [mandibulo-oculofacial dyscephaly](#) and [Hallermann-Streiff](#), [Hallermann-Streiff-François](#), or [François s.](#)

oculo-oto-radial syndrome, [IVIC s.](#)

oculopharyngeal syndrome, see under [dystrophy](#).

ODD syndrome, [oculodentodigital dysplasia](#).

OFD syndrome, [oral-facial-digital s.](#)

Ogilvie syndrome, distention of the colon resembling that caused by obstruction, but without evidence of mechanical obstruction; it is usually due to a defect in the sympathetic nerve supply. Called also [false colonic obstruction](#).

Oldfield syndrome, familial polyposis of the colon associated with extensive sebaceous cysts.

Olmsted syndrome, a rare syndrome of unknown etiology in which infants develop [keratoderma](#) on the palms and soles as they begin to use their hands and feet; this increases in severity to cause contractures and sometimes fissuring of the skin. Many also have plaques of keratoderma around body orifices.

Omenn syndrome, an autosomal recessive but genetically heterogeneous disorder combining immunodeficiency with inflammation; it is characterized by failure to thrive, pneumonitis, and chronic diarrhea beginning early in infancy as well as a variety of autoimmune and allergic inflammatory responses.

one-and-a-half syndrome, a disorder of ocular movement caused by a brainstem lesion of one abducens nucleus and the nearby medial longitudinal fasciculus; the ipsilateral eye cannot move beyond the midline horizontally and the contralateral eye abducts on any attempt at conjugate gaze. Called also [Fisher s.](#) and [Miller Fisher s.](#)

Opitz syndrome, **Opitz-Frias syndrome**, an autosomal dominant syndrome consisting of hypertelorism and hernias, and in males hypospadias, cryptorchidism, and bifid scrotum. Cardiac anomalies, laryngotracheal malformations, imperforate anus, renal defects, lung hypoplasia, and downslanted palpebral fissures may also be present. Called also [G s.](#) and [hypertelorism-hypospadias s.](#)

opsoclonus-myoclonus syndrome, a syndrome of movements of the eyes (opsoclonus) and trunk (myoclonus), occurring in conjunction with a number of conditions, including viral infections, trauma, drug toxicity, tumors, and hyperosmolar nonketotic coma. It also occurs as a paraneoplastic syndrome; in some cases of small cell lung carcinoma or breast or fallopian tube cancer, it is associated with an autoantibody ([anti-Ri](#)).

oral-facial-digital syndrome, type I, a male-lethal X-linked dominant disorder characterized by camptodactyly, polydactyly, and syndactyly; by cranial, facial, lingual, and dental anomalies; and by mental retardation, familial trembling, alopecia, and seborrhea of the face and milia. Called also [orodigitofacial dysostosis](#) and [orofaciadigital s., type I](#). See also [Mohr s.](#)

oral-facial-digital syndrome, type II, [Mohr s.](#)

oral-facial-digital syndrome, type III, an autosomal recessive disorder characterized by postaxial hexadactyly of the hands and feet, by ocular, lingual, and dental anomalies, and by profound mental retardation. Called also [orodigitofacial dysostosis](#) and [orofaciadigital s., type III](#).

orbital apex syndrome, ophthalmoplegia with impairment of vision that may lead to blindness, swelling of the eyelids, ptosis, hyper- or hypoesthesia of the upper eyelid, one half of the forehead, and cornea, and vasomotor disturbances; it is caused by traumatic, inflammatory, or neoplastic processes involving the sphenoidal fissure and optic canal or the structures they contain.

orbital floor syndrome, exophthalmos, diplopia, and anesthesia in the areas innervated by the trigeminal nerve, occurring with a lesion in the floor of the orbit. Called also [Dejean s.](#)

organic anxiety syndrome, a term used in a former system of classification, denoting an organic mental syndrome characterized by prominent, recurrent panic attacks or generalized anxiety caused by a specific organic factor and not associated with delirium. Such disorders are now mainly classified as [substance-induced anxiety disorders](#) and anxiety disorders due to a general medical condition.

organic brain syndrome, [organic mental s.](#)

organic delusional syndrome, a term used in a former system of classification, denoting an organic mental syndrome characterized by the presence of delusions caused by a specific organic factor and not associated with delirium. Such disorders are now mainly classified as [substance-induced psychotic disorders](#) and psychotic disorders due to a general medical condition.

organic dust toxic syndrome, pneumonitis, usually [hypersensitivity pneumonitis](#), resulting from an allergic reaction to inhaled organic dust, as in [bagassosis](#) and various other conditions.

organic mental syndrome, former term for a constellation of psychological or behavioral signs and symptoms associated with brain dysfunction of unknown or unspecified etiology and grouped according to symptoms (cf. [organic mental disorder](#)). Designating certain conditions as having an organic basis, possibly implying that others do not, is currently discouraged.

organic mood syndrome, a term used in a former system of classification, denoting an organic mental syndrome characterized by the presence of manic or depressive mood disturbance caused by a specific organic factor and not associated with delirium. Such disorders are now mainly classified as [substance-induced mood disorders](#) and mood disorders due to a general medical condition.

organic personality syndrome, a term used in a former system of classification, denoting an organic mental syndrome characterized by a marked change in behavior or personality, caused by a specific organic factor and not associated with delirium or dementia. The most common causes are space-occupying lesions of the brain, head trauma, and cerebrovascular disease.

orofaciadigital syndrome, type I, [oral-facial-digital s., type I](#).

orofaciadigital syndrome, type II, [Mohr s.](#)

orofaciadigital syndrome, type III, [oral-facial-digital s., type III](#).

Ortner syndrome, laryngeal paralysis associated with heart disease, due to compression of the recurrent laryngeal nerve between the aorta and a dilated pulmonary artery.

osmotic demyelination syndrome, a form of central pontine myelinolysis that occurs when a patient with chronic hypernatremia has the condition corrected too quickly, so that cells of the central nervous system experience the cerebrospinal fluid as being hypertonic.

Ostrum-Furst syndrome, congenital synostosis of the neck, platybasia, and Sprengel deformity.

Othello syndrome, delusional belief in the infidelity of the sexual partner, often of sudden onset and usually affecting middle-aged men; it is characterized by repeated accusations, intense searches for evidence, and prolonged interrogation of the partner.

outlet syndrome, [thoracic outlet s.](#)

ovarian hyperstimulation syndrome, an iatrogenic condition seen in women undergoing ovulation induction, characterized by mild to severe ovarian enlargement with exudation of fluid and protein, leading to ascites, pleural or pericardial effusion, azotemia, oliguria, and thromboembolism.

ovarian remnant syndrome, pelvic pain, sometimes cyclic, typically occurring several weeks or months after [oophorectomy](#), usually associated with a pelvic mass, most frequently a corpus luteum cyst, which occasionally leads to unilateral ureteral obstruction. It is due to survival of an ovarian fragment after the operation.

ovarian vein syndrome, obstruction of the ureter due to compression by an enlarged or varicose ovarian vein; typically the vein becomes enlarged during pregnancy, the symptoms being those of obstruction or infection of the upper urinary tract. The right side is usually affected.

overlap syndrome, any of a group of connective tissue disorders that either combine scleroderma with polymyositis or systemic lupus erythematosus or combine systemic lupus erythematosus with rheumatoid arthritis or polymyositis. Cf. [mixed connective tissue disease](#).

overtraining syndrome, a group of symptoms seen in athletes who train beyond the body's ability to adapt; there is inability to sustain intense exercise, generalized fatigue, and sometimes mood or sleep disturbances that may last for weeks after the training has stopped or decreased. In some persons a contributing factor is hormonal imbalance caused by the physical stress.

overuse syndrome, see under [injury](#).

overwear syndrome, extreme photophobia, pain, and lacrimation associated with contact lenses, particularly non-gas permeable hard

lenses, usually caused by wearing them excessively. Prolonged lens-induced corneal hypoxia results in corneal epithelial edema and eventually erosion; it can be a chronic condition or an acute episode that usually occurs several hours after lenses are removed.

pacemaker syndrome, vertigo, syncope, and hypotension, often accompanied by dyspnea, cough, nausea, peripheral edema, and palpitations, induced or exacerbated by abnormalities of the [cardiac pacemaker](#) so that it stimulates the ventricle. The symptoms occur because ventricular pacing does not maintain normal atrioventricular synchrony; retrograde ventriculoatrial conduction causes low cardiac output and activates cardiac reflexes that result in increased peripheral vasodilation and hypotension.

pacemaker twiddler syndrome, [twiddler's syndrome](#) in a patient with an artificial [cardiac pacemaker](#).

Paget-Schroetter syndrome, **Paget-von Schroetter syndrome**, a [thoracic outlet syndrome](#) in which a thrombus forms in the axillary vein after strenuous exercise; symptoms include pain, edema, and skin discoloration in the shoulder and upper arm. Called also [effort thrombosis](#).

painful arc syndrome, shoulder pain occurring at a particular portion of the arc described when the arm is abducted from the side to the fully raised position, as in inflammation of the tendons of the supraspinatus muscle.

painful bruising syndrome, a [purpuric](#) reaction usually seen in young to middle-aged women, characterized by spontaneous, chronic, recurring painful [ecchymoses](#), occurring singly or multiply, without significant prior trauma; it may be precipitated by emotional stress. Because some patients exhibit [autoerythrocyte sensitization](#) (q.v.), some authorities believe the etiology to be [autosensitization](#) to a component of the erythrocyte membrane; others believe it is a purely psychosomatic or factitious condition. Called also [autoerythrocyte sensitization s.](#), [erythrocyte autosensitization s.](#), and [Gardner-Diamond s.](#)

painful legs and moving toes syndrome, a syndrome of neuropathic pain in the lower limbs with involuntary movements of the toes or feet; some cases accompany peripheral or central nervous system disorders, and others are idiopathic.

paleostriatal syndrome, [juvenile paralysis agitans \(of Hunt\)](#).

Pallister-Killian syndrome, a rare syndrome of [mosaicism](#) in which only the [fibroblasts](#) are [aneuploid](#), having 47 chromosomes. This results in mental retardation, seizures, streaks of discoloration on the skin, various facial anomalies, and less often defects of limbs, heart, or other structures. Called also [tetrasomy 12p](#).

palmar fasciitis and polyarthritides syndrome, a syndrome of palmar fasciitis, which may be crippling, and polyarthritides in various parts of the body, seen most often as a paraneoplastic condition.

palmar-plantar erythrodysesthesia syndrome, [hand-foot s.](#)

Pancoast syndrome, neuritic pain in the arm, atrophy of the muscles of the arm and hand, and [Horner syndrome](#), observed with a [pulmonary sulcus tumor](#), due to involvement of the brachial plexus and cervical sympathetic nerves. osteolysis in the posterior part of one or more ribs, sometimes also involving the corresponding vertebra.

pancreatic cholera syndrome, [Verner-Morrison s.](#)

pancreatichepatic syndrome, [Zieve s.](#)

pancytopenia-dysmelia syndrome, [Fanconi s.](#) (def. 1).

PAPA syndrome, a rare, autosomal dominant type of [autoinflammatory disease](#) (q.v.), characterized by pyogenic arthritis, pyoderma gangrenosum, and acne.

Papillon-Lefèvre syndrome, an autosomal recessive disorder occurring between the first and fifth years of life, characterized by psoriasiform palmo-plantar keratoderma, which may also involve the elbows, knees, tibias, external malleoli, and other areas; ectopic calcifications of the skull; and periodontitis and premature shedding of both the deciduous and permanent teeth.

papular-purpuric gloves-and-socks syndrome, a syndrome resulting from parvovirus B19 infection, characterized by pruritus, edema, and erythema of the hands and feet, often with shallow ulcerations in and around the mouth, accompanied by fever and leukopenia.

paraganglioma syndrome, an autosomal dominant disorder characterized by tumors of the [paraganglia](#), most frequently of the [carotid body](#), classified into four forms (PGL1–PGL4) on the basis of the site of the mutation; three result from mutations in genes coding for subunits of [succinate dehydrogenase](#), while the site of mutation in the fourth (PGL2) is unknown.

paraneoplastic syndrome, a symptom-complex arising in a cancer-bearing patient that cannot be explained by local or distant spread of the tumor.

paratrigeminal syndrome, [Raeder paratrigeminal s.](#)

Parinaud syndrome, paralysis of conjugate upward movement of the eyes without paralysis of convergence, associated with lesions of the midbrain, such as a lesion in the superior colliculi or a tumor of the pineal gland.

Parinaud oculoglandular syndrome, conjunctivitis, most often unilateral and of the follicular type, followed by tenderness and enlargement of the preauricular lymph nodes; it is often associated with other infections, such as [cat-scratch fever](#), [lymphogranuloma venereum](#), or [tularemia](#).

Parkes Weber syndrome, [Sturge-Weber s.](#)

parkinsonian syndrome, any disorder manifesting the symptoms of [Parkinson disease](#).

Parry-Romberg syndrome, [facial hemiatrophy](#).

Parsonage-Turner syndrome, [neuralgic amyotrophy](#).

partial androgen insensitivity syndrome, a type of androgen insensitivity syndrome of less severity than the [complete androgen insensitivity syndrome](#), with genetic males having decreased sensitivity of target tissues to androgen. Affected boys are often born with ambiguous genitalia, such as hypospadias and a small vaginal pouch, a hooded phallus, or a bifid scrotum that may or may not contain gonads. Called also [incomplete testicular feminization](#) and [partial androgen resistance](#).

Patau syndrome, [trisomy 13 s.](#)

Paterson syndrome, **Paterson?Brown Kelly syndrome**, **Paterson-Kelly syndrome**, [Plummer-Vinson s.](#)

Pearson syndrome, a rare, often fatal, congenital syndrome characterized by transfusion-dependent [sideroblastic anemia](#) with vacuolation of bone marrow precursors and pancreatic dysfunction; it is caused by a large deletion in the mitochondrial genome.

peeling skin syndrome, [keratolysis exfoliativa](#).

Pellizzi syndrome, [epiphyseal s.](#)

Pendred syndrome, an autosomal recessive syndrome of congenital bilateral nerve deafness with development in middle childhood of goiter without hypothyroidism; the main biochemical feature is [dyschoronogenesis](#) of [thyroxine](#).

PEP syndrome, [POEMS s.](#)

Pepper syndrome, neuroblastoma with metastases to the liver.

pericolic membrane syndrome, symptoms resembling those of chronic [appendicitis](#), owing to the pressure of [pericolic membranes](#).

Perlman syndrome, a rare, lethal syndrome consisting of renal dysplasia, nephroblastoma, fetal gigantism, and hypertrophy of the islets of Langerhans with hyperinsulinism. It may be transmitted by autosomal recessive inheritance.

persistent müllerian duct syndrome, a hereditary syndrome in males, either X-linked or autosomal recessive, consisting of persistence of müllerian structures in addition to male genital ducts, with undescended testes and bilateral uterine tubes, a uterus, and an upper vagina. There may be cryptorchidism on just one side with a contralateral inguinal hernia that contains a testis, uterus, and uterine tube ([hernia uteri inguinalis](#)).

pertussis syndrome, [pertussis](#).

pertussis-like syndrome, a syndrome clinically indistinguishable from [pertussis](#), but without evidence of infection with [Bordetella pertussis](#) or [B. parapertussis](#); the most common infectious agents are [adenoviruses](#). Cf. [parapertussis](#).

petrosphenoid syndrome, [Jacod s.](#)

Peutz-Jeghers syndrome, an autosomal dominant [polyposis](#) syndrome characterized by [hamartomas](#) of the small intestine and excessive [melanin](#) pigmentation of the skin and mucous membranes; gastrointestinal bleeding and [intussusception](#) are common complications.

Pfeiffer syndrome, an autosomal dominant disorder characterized by acrocephalosyndactyly associated with broad short thumbs and great toes. Called also [acrocephalopolysyndactyly, type I](#), [acrocephalosyndactyly, type V](#), and [Noack s.](#)

PHACES syndrome, a congenital syndrome of posterior fossa malformations, hemangioma, arterial anomalies, cardiac defects, eye anomalies, and sternal defects.

pharyngeal pouch syndrome, [DiGeorge s.](#)

PHC syndrome, an autosomal dominant syndrome consisting of premolar aplasia, hyperhidrosis, and premature canities. Called also [Böök s.](#)

pickwickian syndrome, [obesity-hypoventilation s.](#)

PIE syndrome, any syndrome characterized by pulmonary infiltrates with eosinophilia, such as [Löffler syndrome](#) or [chronic eosinophilic pneumonia](#). a syndrome of diffuse pulmonary infiltration and peripheral eosinophilia, seen in dogs and sometimes cats; the cause varies but sometimes it may be an allergic reaction. Affected animals are dyspneic with decreased exercise tolerance. Called also [eosinophilic pneumonia](#).

Pierre Robin syndrome, see under [sequence](#).

pineal syndrome, [epiphyseal s.](#)

piriformis syndrome, [levator ani s.](#)

placental dysfunction syndrome, [postmaturity s.](#)

plica syndrome, pain, tenderness, crepitus, and swelling of the knee joint, sometimes accompanied by weakness or locking of the joint, caused by fibrosis and calcification of the synovial plicae, usually the mediopatellar plica.

Plummer-Vinson syndrome, a syndrome usually seen in middle-aged women with [hypochromic anemia](#), characterized by cracks or fissures at the corners of the mouth, painful tongue with atrophy of filiform and later fungiform papillae, and dysphagia due to [esophageal webs](#) or stenosis. Called also [Paterson](#), [Paterson-Kelly](#), or [Vinson s.](#) and [sideropenic dysphagia](#).

POEMS syndrome, a multisystem syndrome combining polyneuropathy, organomegaly, endocrinopathy, M component, and skin changes. It may be linked to a dysproteinemia such as the presence of unusual monoclonal proteins and light chains. Called also [Crow-Fukase s.](#) and [PEP s.](#)

Poland syndrome, unilateral absence of the sternocostal head of the pectoralis major muscle and ipsilateral syndactyly; called also [Poland anomaly](#).

Polhemus-Schafer-Ivemark syndrome, [Ivemark s.](#)

polyangiitis overlap syndrome, a systemic form of necrotizing vasculitis with clinicopathologic signs overlapping those of polyarteritis nodosa and allergic granulomatous angiitis, but also showing features of hypersensitivity vasculitis. Immunoglobulins are often deposited in involved organs. Called also [overlap vasculitis](#).

polycystic ovary syndrome, a clinical symptom complex associated with [polycystic ovaries](#), characterized by [oligomenorrhea](#) or [amenorrhea](#), [anovulation](#) (hence [infertility](#)), and [hirsutism](#). Both [hyperestrogenism](#) (from peripheral conversion of androgen) and [hyperandrogenism](#) are present. Called also [polycystic ovary disease](#) and [Stein-Leventhal s.](#)

polyendocrine autoimmune syndromes, **polyglandular autoimmune syndromes**, syndromes comprising combinations of endocrine and nonendocrine autoimmune diseases. Type I is caused by mutations in the autoimmune regulator gene (AIRE, locus: 21q22.3) and occurs in infants and children. It is characterized by the presence of two of three major clinical symptoms: candidiasis, hypoparathyroidism, and adrenal insufficiency. Pernicious anemia, vitiligo, gonadal failure, alopecia, insulin-dependent diabetes, or thyroid autoimmune disease may also occur. Type II is [Schmidt s.](#) Called also [autoimmune polyendocrine](#) or [autoimmune polyglandular s.](#)

polyglandular deficiency syndrome, [multiple endocrine deficiency s.](#)

polysplenia syndrome, a congenital syndrome characterized by multiple splenic masses, left-sidedness, abnormal position and development of visceral organs, complex cardiovascular defects, and abnormal, usually bilobate, lungs; it may be related to [Ivemark syndrome](#).

pontine syndrome, [Raymond-Cestan s.](#)

popliteal pterygium syndrome, [popliteal web s.](#) [Fèvre-Languépin s.](#)

popliteal web syndrome, a congenital syndrome consisting chiefly of popliteal webs, cleft palate, lower lip pits, and dysplasia of the toenails; a wide variety of other abnormalities may be associated. Called also [popliteal pterygium s.](#)

porcine epidemic abortion and respiratory syndrome, a disease caused by a virus of the genus [Arterivirus](#), affecting pregnant pigs and characterized by fever, anorexia, and respiratory distress followed by unusually high numbers of stillbirths and other piglets born with respiratory distress, weakness, and neurological conditions such as splayleg. Called also [mystery pig disease](#).

porcine stress syndrome, sudden death of a pig in response to a stressor such as fighting, transport, or malignant hyperthermia, or as part of a drug reaction. Susceptibility in most cases is inherited as an autosomal recessive gene. Called also [herztod](#).

post-cardiac injury syndrome, fever, chest pain, pleuritis, and pericarditis occurring several weeks to months after injury to the heart, including that inflicted by surgery (often called [postpericardiectomy s.](#)) or myocardial infarction (often called [post-myocardial infarction s.](#)).

postcardiotomy syndrome, [postpericardiectomy s.](#)

postcardiotomy psychosis syndrome, anxiety, confusion, and perceptual disturbances occurring three or more days after open heart surgery.

postcholecystectomy syndrome, a type of [sphincter of Oddi dysfunction](#) (see under dysfunction) seen after [cholecystectomy](#).

postcommissurotomy syndrome, [postpericardiectomy s.](#)

postconcussion syndrome, **postconcussional syndrome**, physical and personality changes that sometimes occur after [concussion of the brain](#) (q.v.); they include amnesia, headache, dizziness, tinnitus, irritability, fatigability, sweating, palpitations of the heart, disordered sleep, and difficulty in concentrating. Called also [traumatic encephalopathy](#) and [concussion](#), [posttraumatic](#), or [posttraumatic brain s.](#) Cf. [boxer's dementia](#) and chronic traumatic encephalopathy.

posterior column syndrome, **posterior cord syndrome**, sensory deficits and ataxic phenomena derived from a lesion of the posterior columns of the spinal cord.

posterior inferior cerebellar artery syndrome, [Wallenberg s.](#)

posterior leukoencephalopathy syndrome, **posterior reversible encephalopathy syndrome**, **posterior reversible leukoencephalopathy syndrome**, [reversible posterior leukoencephalopathy s.](#)

postgastrectomy syndrome, [dumping s.](#)

postirradiation syndrome, a symptom complex caused by massive irradiation, with hemorrhage, anemia, and malnutrition. See also [acute radiation s.](#)

post-lumbar puncture syndrome, the [lumbar puncture headache](#) and other symptoms, which may include pain at the back of the neck, nausea, vomiting, sweating, and malaise, felt in the erect position and relieved when the person lies down, lasting from several hours after lumbar puncture for sometimes a few days; it is due to lowering of [intracranial pressure](#) by leakage of cerebrospinal fluid through the needle tract.

postmastectomy pain syndrome, chronic pain in the breast, axilla, or proximal part of the upper limb after a mastectomy, aggravated by movement of the shoulder and sometimes by stretching or other exertion of the arm muscles; it is usually neuropathic, caused by trauma to a nerve in the area, particularly the intercostobrachial nerve or superior lateral cutaneous nerve.

postmaturity syndrome, a syndrome due to placental insufficiency that causes chronic stress and hypoxia, seen in fetuses and newborn infants in postterm pregnancies and characterized by decreased subcutaneous fat, skin desquamation, and long fingernails, often with yellow meconium staining of the nails, skin, and vernix. Called also [dysmaturity](#), [dysmaturity s.](#), and [placental dysfunction s.](#)

post-myocardial infarction syndrome, pericarditis with fever, leukocytosis, pleurisy, and pneumonia occurring after myocardial infarction; called also [Dressler s.](#)

postperfusion syndrome, a type of [cytomegalovirus mononucleosis](#) sometimes seen about 3 to 6 weeks after [extracorporeal circulation](#) or multiple blood transfusions.

postpericardiotomy syndrome, pericardial or pleural reaction occurring more than one week after opening of the pericardium, characterized by fever, chest pain, and signs of pleural and/or pericardial inflammation.

postphlebotic syndrome, the various complications associated with deep vein thrombosis which are caused by greatly increased pressure in the deep and communicating veins, resulting in chronic venous insufficiency, and principally characterized by persistent edema, pain, purpura and increased cutaneous pigmentation, eczematoid dermatitis, pruritus, ulceration, and indurated cellulitis. Called also [postthrombotic s.](#)

postpolio syndrome, **postpoliomyelitis syndrome**, a group of symptoms of unknown etiology seen in patients several years to many years after they have recovered from the major illness of [poliomyelitis](#); it includes new weakness, fatigue, and pain, either generalized or limited to the parts that were affected by the poliomyelitis. Called also [postpoliomyelitis sequela](#).

postpump syndrome, a series of complications occasionally seen in patients after heart surgery that involved [cardiopulmonary bypass](#), with pulmonary edema and other signs of [acute respiratory distress syndrome](#) and damage to kidneys, brain, and other organs that can be fatal.

postthrombotic syndrome, [postphlebotic s.](#)

posttransfusion syndrome, former name for [postperfusion s.](#)

posttraumatic syndrome, **posttraumatic brain syndrome**, [postconcussional s.](#)

postural orthostatic tachycardia syndrome, a group of symptoms (not including hypotension) that sometimes occur when a person assumes an upright position, including tachycardia, tremulousness, lightheadedness, sweating, and hyperventilation; this is seen more often in women than in men, and the etiology is uncertain.

Potter syndrome, [oligohydramnios sequence](#).

Prader-Willi syndrome, a congenital disorder characterized by obesity, short stature, lack of muscle tone, hypogonadism, and central nervous system dysfunction; there is often a characteristic rounded face with almond-shaped eyes and a low forehead. Mental retardation is common. It is caused by loss of expression of genes on paternally derived chromosome 15q11–13, usually as a result of a deletion; the same loss of gene function inherited from the mother causes [Angelman syndrome](#).

preexcitation syndrome, any syndrome characterized by electrocardiographic evidence of preexcitation, such as Wolff-Parkinson-White syndrome or Lown-Ganong-Levine syndrome; sometimes used as a synonym of the former.

premenstrual syndrome, a syndrome of unknown cause, typically occurring in the period between ovulation and the onset of menstruation, marked by some or all of the following symptoms: feelings of depression, hopelessness, anxiety, or anger, emotional lability, bloating, edema, headache, increased fatigue or lethargy, changes in appetite or cravings for selected foods, breast swelling and tenderness, constipation, and decreased ability to concentrate.

premotor syndrome, the association of spastic hemiplegia with increased reflexes, disturbances of skilled movements, forced grasping, and transient vasomotor disturbance; occurring in very large lesions of the premotor cortex.

progeroid syndrome, [Werner s.](#)

prolonged QT interval syndrome, [long QT s.](#)

pronator syndrome, **pronator teres syndrome**, an entrapment neuropathy in which the median nerve or its anterior interosseous branch is compressed by the structures of the cubital fossa or by its passage between the heads of the pronator teres muscle, causing pain in the forearm and weakness or sensory deficits in the radial aspect of the hand.

Proteus syndrome, a rare congenital disorder with highly variable manifestations, including partial gigantism of the hands and feet with hypertrophy of the palms and soles, [epidermal nevi](#), [hemihypertrophy](#), subcutaneous tumors, [macrocephaly](#) and other skull abnormalities, and abdominal or pelvic [lipomatosis](#). The etiology is unknown, although a genetic origin, possibly of autosomal dominant transmission, has been conjectured.

prune-belly syndrome, a syndrome in which the lower part of the rectus abdominis muscle and the lower and medial parts of the oblique muscles are absent, the bladder and ureters are usually greatly dilated, the kidneys are small and dysplastic, with hydronephrosis, and the testes are undescended. The abdomen is protruding and thin-walled, with wrinkled skin, giving the syndrome its name. Called also [abdominal muscle deficiency s.](#) and [Eagle-Barrett s.](#)



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Prune-belly syndrome.

pseudo-Cushing syndrome, the presence of clinical or biochemical features of [Cushing syndrome](#) in patients with certain nonendocrine disorders, such as alcoholism or depression.

Pseudomonas hot foot syndrome, a skin eruption characterized by painful, erythematous nodules on the soles of the feet, seen in persons exposed to [Pseudomonas aeruginosa](#), such as children using a wading pool with abrasive strips of tape on the bottom. Called also [hot foot s.](#)

pterygium syndrome, [multiple pterygium s.](#)

puborectalis syndrome, [levator ani s.](#)

pulmonary acid aspiration syndrome, the disorder produced, as a complication of anesthesia, by inhalation of gastric content with a pH of less than 2.5, including bronchoconstriction and destruction of tracheal mucosa, progressing to a syndrome resembling [acute respiratory distress syndrome](#). Called also [Mendelson s.](#)

pulmonary dysmaturity syndrome, [Wilson-Mikity s.](#)

pulmonary renal syndrome, any of several conditions in which diffuse alveolar hemorrhage occurs in patients with [glomerulonephritis](#), such as with [anti-GBM nephritis](#), [lupus nephritis](#), or [immune complex glomerulonephritis](#).

pulmonary sling syndrome, a constellation of unilateral aeration disturbances caused by a [pulmonary artery sling](#) with tracheal stenosis. See also [sling ring complex](#), under complex.

punch drunk syndrome, [chronic traumatic encephalopathy](#).

Purtilo syndrome, [X-linked lymphoproliferative s.](#)

Putnam-Dana syndrome, [subacute combined degeneration of spinal cord](#) (see under degeneration).

quadrilateral space syndrome, a rare type of [entrapment neuropathy](#) caused by compression of the [axillary nerve](#) as it passes through the [quadrilateral space](#); symptoms are paresthesias and shoulder pain upon abduction of the arm.

rabbit syndrome, a side effect of certain antipsychotic drugs, especially neuroleptics, characterized by involuntary facial movements

resembling those of a rabbit chewing.

Rabson-Mendenhall syndrome, a rare syndrome seen in children, characterized by a mutation or other defect in an [insulin receptor](#) gene, with severe [insulin resistance](#) and [acanthosis nigricans](#) as well as thick hair, abnormalities of teeth and nails, and hyperplasia of the pineal gland.

radial tunnel syndrome, a type of [radial neuropathy](#) in which the posterior interosseous nerve is compressed in the [radial tunnel](#), causing paresthesias and paralysis in the wrist and hand.

radicular syndrome, a syndrome due to lesion of the roots of the spinal nerves, consisting of restricted mobility of the spine and root pain.

Raeder syndrome, **Raeder paratrigeminal syndrome**, paroxysmal neuralgic pain on one side of the face associated with blockage of sympathetic nerve impulses; see also [Horner s.](#) Called also [paratrigeminal s.](#)

Ramsay Hunt syndrome, [herpes zoster](#) involving the facial and vestibulocochlear nerves, often associated with transitory ipsilateral facial paralysis and herpetic vesicles of the external ear or tympanic membrane; there may or may not be tinnitus, vertigo, and hearing disorders. Called also [geniculate](#) or [Hunt neuralgia](#) and [herpes zoster oticus.juvenile paralysis agitans \(of Hunt\).dyssynergia cerebellaris progressiva.](#)

Rapp-Hodgkin syndrome, an autosomal dominant syndrome caused by mutations in the TP63 gene (locus: 3q27), which encodes a regulatory protein important in stem cell maintenance in stratified epithelial tissues, characterized by anhidrotic ectodermal dysplasia, cleft lip and palate, and mid face hypoplasia.

Rasmussen syndrome, see under [encephalitis.](#)

Raymond-Cestan syndrome, a syndrome due to obstruction of twigs of the basilar artery causing lesions of the pontine region; it is characterized by quadriplegia, anesthesia, and nystagmus. Called also [Cestan-Raymond s.](#) and [pontine s.](#)

reactive airways dysfunction syndrome, a rare asthmlike disorder consisting of persistent coughing, wheezing, and dyspnea upon only slight irritation, lasting for months after a person has inhaled a high concentration of irritating fumes.

red man syndrome, sudden flushing of the skin, usually of the head and upper body, often with pruritus, following administration of medication; it is most common with [vancomycin](#) and has been observed with certain other drugs. It is thought to be related to release of [histamine](#) and [mast cells.](#)

redundant supraglottic mucosa syndrome, redundancy of the aryepiglottic folds, the mucosa overlying the arytenoid cartilages, and the interarytenoid region of the larynx, associated with obstructive sleep apnea, with or without stridor.

Reed syndrome, [leiomyomatosis cutis et uteri.](#)

refeeding syndrome, moderate to severe electrolyte and fluid shifts occurring during a period of [refeeding](#) (q.v.); [hypophosphatemia](#) is common, and [heart failure](#) sometimes occurs.

Reichel syndrome, [Henderson-Jones disease.](#)

Reifenstein syndrome, a syndrome of male [hypergonadotropic hypogonadism](#), due to [partial androgen insensitivity syndrome](#), with hypospadias, gynecomastia, primary hypogonadism, and postpubertal testicular atrophy and azoospermia.

Reiter syndrome, the triad of acute aseptic arthritis, nongonococcal urethritis, and conjunctivitis; there may also be mucocutaneous manifestations such as [keratoderma blennorrhagicum](#), [circinate balanitis](#), and stomatitis. It usually affects young men and runs a self-limited but relapsing course. Some authorities now consider this symptom complex to be more appropriately classified as [reactive arthritis](#) and not distinguished or named separately. Called also [Fliessinger-Leroy-Reiter s.](#)



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Reiter syndrome, with bony erosion at the insertion of the Achilles tendon on the posterosuperior margin of the calcaneus (arrows).

renal-retinal syndrome, [Senior-Loken s.](#)

Rendu-Osler-Weber syndrome, [hereditary hemorrhagic telangiectasia.](#)

respiratory distress syndrome, see [acute respiratory distress s.](#) and [neonatal respiratory distress s.](#)

respiratory distress syndrome of newborn, [neonatal respiratory distress s.](#)

restless legs syndrome, unpleasant deep discomfort including [paresthesias](#) inside the calves when sitting or lying down, especially just before sleep, producing an irresistible urge to move the legs; the cause is unknown, but in some cases it may be due to inadequate circulation, a reaction to medication, or a complication of [uremia](#). Called also [restless legs](#) and [Ekbom s.](#)

retained gastric antrum syndrome, a syndrome sometimes seen after partial gastrectomy in the Billroth II operation, if a segment of antrum was retained. Lack of the usual acid-secreting gastric glands means that the antral segment is continually exposed to the alkaline environment of the duodenum, which causes it to secrete excessive acid and be prone to form ulcers.

retraction syndrome, [Duane s.](#)

Rett syndrome, a pervasive developmental disorder affecting the gray matter of the brain, occurring exclusively in females and present from birth; it is progressive and is characterized by autistic behavior, ataxia, dementia, seizures, and loss of purposeful use of the hands, with cerebral atrophy, mild hyperammonemia, and decreased levels of biogenic amines. It is an X-linked dominant disorder caused by a loss-of-function mutation in the MECP2 gene, which encodes a methyl-CpG-binding protein that regulates transcription of other genes. Called also [cerebroatrophic hyperammonemia.](#)

reversible posterior leukoencephalopathy syndrome, a syndrome resulting from [leukoencephalopathy](#) with edema in posterior parts of the occipital and parietal lobes, characterized by headaches, confusion, seizures, and visual disturbances; the brain lesions are most often related to [hypertension](#), and sometimes to use of certain [immunosuppressive](#) drugs or to some other cause. Called also [posterior leukoencephalopathy s.](#), [posterior reversible encephalopathy s.](#), and [posterior reversible leukoencephalopathy s.](#)

Reye syndrome, **Reye-Johnson syndrome**, a rare, acute, sometimes fatal disease of childhood, characterized by recurrent vomiting and elevated serum transaminase levels, with distinctive changes in the liver and other viscera; an encephalopathic phase may follow with acute

brain swelling, disturbances of consciousness, and seizures. It most often occurs as a sequela of chickenpox or a viral upper respiratory infection.

Rh-null syndrome, chronic hemolytic anemia in persons who lack all Rh factors (Rh_{null}); it is marked by spherocytosis, stomatocytosis, and increased osmotic fragility.

Richards-Rundle syndrome, a congenital syndrome consisting of [ketoaciduria](#), mental retardation, low development of [secondary sex characters](#), deafness, lack of muscle coordination, and peripheral muscular wasting that progresses during childhood but eventually becomes static.

Richner-Hanhart syndrome, [type II tyrosinemia](#).

Richter syndrome, [chronic lymphocytic leukemia](#) with diffuse [histiocytic lymphoma](#).

Rieger syndrome, [Rieger anomaly](#) accompanied by hypodontia, anal stenosis, hypertelorism, mental deficiency, and agenesis of the facial bones. See also [anterior chamber cleavage s.](#)

rigid spine syndrome, a rare type of muscular dystrophy characterized by weak neck muscles, fibrous contractures of the vertebral column with scoliosis, and respiratory insufficiency.

Riley-Day syndrome, [familial dysautonomia](#).

Riley-Smith syndrome, macrocephaly without hydrocephalus, multiple hemangiomas, and pseudopapilledema; presumed to be transmitted as an autosomal dominant trait.

Roberts syndrome, a hereditary syndrome, transmitted as an autosomal recessive trait, consisting of imperfect development of the long bones of the limbs associated with cleft palate and lip and other anomalies.

Robinow syndrome, dwarfism associated with increased interorbital distance, malaligned teeth, bulging forehead, depressed nasal bridge, and short limbs. Called also [Robinow dwarfism](#) and [fetal face s.](#)

Rochon-Duvigneaud syndrome, [superior orbital fissure s.](#)

Rokitansky-Küster-Hauser syndrome, [Mayer-Rokitansky-Küster-Hauser s.](#)

rolandic vein syndrome, hemiplegia resulting from interference with the cerebral venous circulation.

Rollet syndrome, [orbital apex s.](#)

Romano-Ward syndrome, the more common, autosomal dominant form of the [long QT syndrome](#), characterized by syncope and sometimes [ventricular fibrillation](#) and sudden death.

Rosenberg-Bergstrom syndrome, an autosomal recessive syndrome characterized by hyperuricemia, renal insufficiency, ataxia, and deafness, probably due to deficiency of ribose-phosphate pyrophosphokinase.

Rosenberg-Chutorian syndrome, a rare X-linked hereditary syndrome characterized by optic atrophy, progressive neural deafness, and polyneuropathy.

Rosenthal syndrome, [factor XI deficiency](#).

Rosenthal-Kloepfer syndrome, a rare hereditary, autosomal dominant syndrome consisting of corneal opacities, features resembling those of [acromegaly](#), and [cutis verticis gyrata](#) of the scalp.

Rosewater syndrome, a mild form of hereditary X-linked [hypergonadotropic hypogonadism](#) in males, characterized by sterility and gynecomastia.

Ross syndrome, a rare syndrome, sometimes considered a variant of Adie syndrome, characterized by tonic pupils, loss of deep tendon reflexes, and hypohidrosis on one side of the body; in some cases there is compensatory hyperhidrosis on the other side.

Roth (Rot) syndrome, **Roth-Bernhardt (Rot-Bernhardt) syndrome**, [meralgia paresthetica](#).

Rothmann-Makai syndrome, a rare syndrome of idiopathic circumscribed [panniculitis](#) with fat cell necrosis, lipophagic granuloma, and cyst formation; it usually subsides spontaneously.

Rothmund-Thomson syndrome, an autosomal recessive syndrome characterized by reticulated, atrophic, hyperpigmented, telangiectatic cutaneous plaques, often with juvenile cataracts, saddle nose, congenital bone defects, disturbances in growth of hair, nails, and teeth, and [hypogonadism](#). At least some cases appear to be caused by mutations affecting a particular DNA [helicase](#). Called also [poikiloderma congenitale](#). Cf. [Thomson disease](#).

Rotor syndrome, a type of chronic [familial nonhemolytic jaundice](#) that differs from [Dubin-Johnson syndrome](#) in the lack of liver pigmentation.

Roussy-Dejerine syndrome, [thalamic s.](#)

Roussy-Lévy syndrome, a slowly progressive autosomal dominant disorder in which sensory ataxia is associated with areflexia, atrophy of muscles of distal extremities, especially the peroneal muscles, static tremor of the hands, pes cavus or clawfoot, and sometimes kyphoscoliosis. Called also [hereditary areflexic dystasia](#), [Lévy-Roussy s.](#), and [Roussy-Lévy hereditary areflexic dystasia](#).

Roux stasis syndrome, a complication sometimes seen in persons who have had a Roux-en-Y procedure, consisting of intractable nausea and abdominal pain because of gastric paresis or failure of food to move through the anastomosis.

Rovsing syndrome, [horseshoe kidney](#) with nausea, abdominal discomfort, and pain on hyperextension.

Rowell syndrome, name given to a rare combination of systemic lupus erythematosus and erythema multiforme.

Rozyccki syndrome, a rare autosomal recessive syndrome characterized by deafness, vitiligo, short stature, and muscle wasting.

RS3PE syndrome, a rare syndrome of acute onset but self-limited course, primarily affecting the elderly, consisting of polyarthritis with seronegativity for rheumatoid factor, accompanied by pitting edema of the hands and sometimes the feet.

rubella syndrome, [congenital rubella s.](#)

Rubinstein syndrome, **Rubinstein-Taybi syndrome**, a congenital condition characterized by mental and motor retardation, broad thumbs and great toes, short stature, characteristic facies including high-arched palate and straight or beaked nose, various eye abnormalities, pulmonary stenosis, keloid formation in surgical scars, large foramen magnum, and abnormalities of the vertebrae and sternum.

rubrospinal cerebellar peduncle syndrome, [Claude s.](#)

Rud syndrome, congenital syndrome consisting of ichthyosis simplex, mental deficiency, epilepsy, and infantilism.

rudimentary testis syndrome, [vanishing testes s.](#)

Rundles-Falls syndrome, [hereditary sideroblastic anemia](#).

runting syndrome, a type of [graft-versus-host reaction](#) characterized by diarrhea, dermatitis, enlargement of the liver and spleen, [hemolytic anemia](#), and [pancytopenia](#).

Russell syndrome, **Russell-Silver syndrome**, [Silver-Russell s.](#)

Rust syndrome, stiff neck, stiff carriage of the head, with the necessity of grasping the head with both hands in lying down or rising up from a horizontal posture, occurring in tuberculosis, cancer, fracture of the spine, rheumatic or arthritic processes, or syphilitic periostitis.

Ruvalcaba syndrome, a syndrome of unknown etiology but present from birth in males; it is characterized by microcephaly, skeletal abnormalities, hypoplastic genitalia, and mental and physical retardation.

Sabin-Feldman syndrome, chorioretinitis and cerebral calcifications, similar to the manifestations of toxoplasmosis, but having all tests for toxoplasmosis negative.

Saethre-Chotzen syndrome, [Chotzen s.](#)

Sakati-Nyhan syndrome, an autosomal dominant disorder characterized by acrocephaly and polysyndactyly with hypoplastic tibias and deformed, displaced fibulas. Called also [acrocephalopolysyndactyly, type III](#).

salt depletion syndrome, **salt-losing syndrome**, vomiting, dehydration, hypotension, and sudden death due to very large sodium losses from the body ([salt wasting](#)). It may be seen in abnormal losses of sodium into the urine (as in congenital adrenal hyperplasia, adrenocortical insufficiency, or one of the forms of [salt-losing nephropathy](#)) or in large extrarenal sodium losses, usually from the gastrointestinal tract. Called also [salt-depletion](#) or [salt-losing crisis](#) and [salt-losing defect](#).

Sandifer syndrome, intermittent [torticollis](#) occurring in children as a symptom of [reflux esophagitis](#) or [hiatal hernia](#).

Sanfilippo syndrome, four heterogeneous, biochemically distinct but clinically indistinguishable forms of mucopolysaccharidosis characterized biochemically by excretion of heparan sulfate in the urine and clinically by severe, rapid mental deterioration and relatively mild somatic symptoms. Onset is from 2 to 6 years of age; the head is large, height normal; Hurler-like features (dysostosis multiplex, hepatomegaly) are mild; hirsutism is generalized; death usually occurs before 20 years of age. The four enzymatic types are type A, the most severe, due to deficiency of heparan N-sulfatase; type B, due to deficiency of a-N-acetylglucosaminidase; type C, due to deficiency of heparan-a-glucosaminidase; and type D, due to deficiency of N-acetylglucosamine-6-sulfatase. Called also [mucopolysaccharidosis III](#).

Santavuori syndrome, Santavuori-Haltia syndrome, Haltia-Santovuori disease.

SAPHO syndrome, a syndrome of unknown etiology, consisting of synovitis, acne, pustulosis, hyperostosis, and osteitis; the osteitis is often painful and usually involves the sternum, ribs, or vertebral column, and less often the mandible or limbs.

scalenus syndrome, scalenus anterior syndrome, scalenus anticus syndrome, a [thoracic outlet syndrome](#) caused by compression of nerves and vessels between a cervical rib and the scalenus anterior muscle; symptoms include pain over the shoulder, often extending down the arm ([brachial plexopathy](#)) or radiating up the back of the neck. Called also [Naffziger s.](#)

scapulocostal syndrome, pain in the superior or posterior aspect of the shoulder girdle, radiating to contiguous regions, as a result of long-standing alteration of the relationship of the scapula and the posterior thoracic wall.

Schäfer syndrome, pachonychia congenita associated with retardation of physical and mental development.

Schanz syndrome, a series of symptoms indicating spinal weakness, consisting of a sense of fatigue, pain on pressure over the spinous processes, pain on lying prone, and indications of spinal curvature.

Schaumann syndrome, sarcoidosis.

Scheie syndrome, a relatively mild allelic variant of [Hurler syndrome](#) and the mildest of the three allelic disorders of [mucopolysaccharidosis I](#), characterized by corneal clouding, claw hand, involvement of the aortic valve, somewhat coarse facies with a broad mouth, genu valgum, and pes cavus. Stature, intelligence, and life span are normal; it is caused by a deficiency of I-iduronidase. Called also [mucopolysaccharidosis IS](#); formerly called [mucopolysaccharidosis V](#).

Schiff-Sherrington syndrome, paraplegia in dogs with rigid extension of the hind limbs, usually associated with acute severe compression of the thoracolumbar spinal cord.

Schimmelpenning syndrome, sebaceous nevus s.

Schinz-Giedion syndrome, a rare syndrome, probably of autosomal recessive inheritance, of [hydronephrosis](#), skeletal abnormalities, flattened midface, [hypertrichosis](#), seizures, and profound growth and developmental retardation.

Schirmer syndrome, a variant of the [Sturge-Weber syndrome](#) in which glaucoma occurs early in the course of the disease.

Schmidt syndrome, [A. Schmidt] paralysis on one side, affecting the vocal cord, the velum palati, the trapezius muscle, and the sternocleidomastoid muscle, due to a brain lesion affecting the vagus nerve and the spinal accessory nerve. Called also [vagoaccessory s.](#) [M. B. Schmidt] hypofunction of more than one endocrine gland, such as the thyroid, adrenals, gonads, parathyroids, and endocrine pancreas (in any combination), along with nonendocrine abnormalities of presumed autoimmune origin, such as vitiligo, alopecia, and pernicious anemia; it occurs primarily in adult females. The term was originally applied to primary failure of the adrenal and thyroid glands. Called also [polyglandular autoimmune syndrome](#), type II.

Schnitzler syndrome, a rare, usually benign syndrome consisting of elevated levels of [immunoglobulin M](#) and chronic, nonpruritic [urticaria](#), fever, and pain in the bones and joints.

Schönlein-Henoch syndrome, Henoch-Schönlein purpura.

Schüller syndrome, Schüller-Christian syndrome, Hand-Schüller-Christian disease.

Schultz syndrome, agranulocytosis.

Schwartz-Jampel syndrome, Schwartz-Jampel-Aberfeld syndrome, an autosomal recessive disorder characterized by muscle abnormalities, dwarfism, [blepharophimosis](#), joint contractures, and flat facies. Called also [chondrodystrophic myotonia](#).

scimitar syndrome, complete or partial venous drainage of the right lung into the inferior vena cava, usually with hypoplasia of the right lung; the anomalous vein has a scimitar shape on a radiograph; see [scimitar sign](#), under sign.

sea-blue histiocyte syndrome, a rare disorder characterized by the presence of abnormal histiocytes that stain blue with Wright and Giemsa stains ([sea-blue histiocytes](#)), accompanied by splenomegaly. Clinically it may range from a benign course with mild purpura secondary to thrombocytopenia, to progressive cirrhosis, hepatic failure, and death. The etiology is unknown, but sometimes it is inherited as an autosomal recessive condition. Called also [sea-blue histiocytosis](#).

Seabright bantam syndrome, pseudohypoparathyroidism.

sebaceous nevus syndrome, a syndrome characterized by a [sebaceous nevus](#) or nevi in a linear formation, present at birth and changing progressively throughout life. Some patients have neurologic symptoms (retardation or seizures) or ophthalmologic abnormalities. After puberty the nevi become thickened and verrucous, often with projections and numerous other skin changes; eventually some lesions become nodular and may develop benign or malignant adnexal tumors or [basal cell carcinoma](#). Called also [Feuerstein-Mims](#), [Schimmelpenning](#), or [Solomon s.](#), [linear nevus sebaceous s.](#), and [linear sebaceous nevus syndrome](#).

Seckel syndrome, a syndrome of unknown etiology, characterized by [intrauterine growth retardation](#) and postnatal dwarfism with a small head, narrow birdlike face with a beaklike nose, large eyes with palpebral fissures higher medially than temporally, receding mandible, and mild mental retardation. Called also [bird-headed dwarfism](#), [Seckel dwarfism](#), and [Virchow-Seckel s.](#)

second impact syndrome, acute, usually fatal, brain swelling and increased cranial pressure, caused by repeated head trauma in a short space of time, so that a second [concussion](#) occurs before recovery from a previous concussion is complete.

Segawa syndrome, dopa-responsive dystonia, particularly that due either to an autosomal dominant defect in [GTP cyclohydrolase I](#) or an autosomal recessive defect in [tyrosine hydroxylase](#).

segmental aneuploidy syndrome, contiguous gene s.

segmentary syndrome, a syndrome produced by a lesion of the gray matter of the spinal cord, and marked by weakness and wasting in the affected segment; called also [metameric s.](#)

Seip-Berardinelli syndrome, total lipodystrophy.

Selye syndrome, general adaptation s.

Senear-Usher syndrome, pemphigus erythematosus.

Senior-Loken syndrome, a rare autosomal recessive syndrome of [tapetoretinal degeneration](#) and [familial juvenile nephronophthisis](#); it is considered by some authorities to be a part of the [juvenile nephronophthisis-medullary cystic disease complex](#). Called also [renal-retinal s.](#)

syndrome of sensory dissociation with brachial amyotrophy, see [syringomyelia](#).

Senter syndrome, keratitis-ichthyosis-deafness s.

serotonin syndrome, a symptom complex caused by [hyperserotonemia](#), usually owing to a person's taking [selective serotonin reuptake inhibitors](#) along with some other drug that increases [serotonin](#) levels. Symptoms are variable but may include mental status changes, autonomic hyperactivity, and irregular muscular movements; severe cases can be fatal.

Sertoli-cell-only syndrome, congenital absence of the germinal epithelium of the testes, so that the [seminiferous tubules](#) contain only [Sertoli cells](#) and the testes are smaller than normal; there is [azoospermia](#) with elevated titers of [follicle-stimulating hormone](#) and sometimes of [luteinizing hormone](#). Called also [del Castillo s.](#) and [germinal or germinal cell aplasia](#).

serum sickness-like syndrome, see [serum sickness](#), under sickness.

Setleis syndrome, a rare, autosomal recessive type of [ectodermal dysplasia](#) characterized by scarlike depressions on the temples,

sometimes missing or supernumerary eyelashes, and redundant skin around the nasal and chin areas.

severe acute respiratory syndrome, an infectious respiratory illness caused by a coronavirus and characterized by fever, dry cough, and breathing difficulties, often accompanied by headache and body aches.

Sézary syndrome, a form of [cutaneous T-cell lymphoma](#) manifested by generalized [exfoliative erythroderma](#), intense pruritus, peripheral lymphadenopathy, and [Sézary cells](#) (abnormal T lymphocytes) in the skin, lymph nodes, and peripheral blood. Called also [Sézary erythroderma](#).

Shah-Waardenburg syndrome, [Waardenburg s. type 4](#).

shaker foal syndrome, a type of botulism in young horses, accompanied by flaccid tetraparesis and inability to swallow, so that there is risk of aspiration pneumonia.

Shapiro syndrome, a rare syndrome of periodic hyperhidrosis and hypothermia owing to agenesis or dysplasia of the corpus callosum, sometimes accompanied by hydrocephalus and mental retardation.

Sheehan syndrome, [postpartum pituitary necrosis](#).

Sheldon-Hall syndrome, an autosomal dominant syndrome caused by mutations in any of several genes that encode for troponin or a myosin heavy chain, with characteristics similar to those of Freeman-Sheldon syndrome but without the severe deformity of the mouth and chin. Called also [distal arthrogryposis 2B](#).

short bowel syndrome, short gut syndrome, any of the malabsorption conditions resulting from massive resection of the small intestine, the degree and kind of malabsorption depending on the site and extent of the resection; it is characterized by diarrhea, steatorrhea, and malnutrition.

short PR syndrome, [Lown-Ganong-Levine syndrome](#).

shoulder-hand syndrome, [reflex sympathetic dystrophy](#) limited to the upper extremity; see under dystrophy.

Shprintzen syndrome, Shprintzen-Goldberg syndrome, [velocardiofacial s.](#)

shrinking lung syndrome, a rare complication of systemic lupus erythematosus, usually because of weakened chest muscles or diaphragm; the diaphragm may be elevated, but radiography of the chest appears normal, although lung volumes are decreased and the patient has dyspnea.

Shulman syndrome, [eosinophilic fasciitis](#).

Shwachman syndrome, Shwachman-Diamond syndrome, a syndrome, believed to be hereditary and autosomal recessive, characterized by primary pancreatic insufficiency and bone marrow failure, with normal sweat chloride values and neutropenia. It may be associated with dwarfism and metaphyseal dysostosis of the hips.

Shy-Drager syndrome, a form of [multiple system atrophy](#) that begins with symptoms of autonomic insufficiency (orthostatic hypotension, impotence in males, constipation, urinary urgency or retention, and anhidrosis); these are followed by signs of generalized neurologic dysfunction, (parkinsonism, cerebellar incoordination, muscle wasting and fasciculations, and coarse tremors of the legs). Called also [chronic, chronic idiopathic](#), or [idiopathic orthostatic hypotension](#).

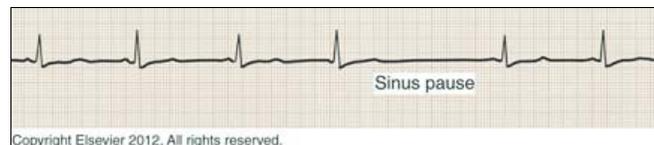
Shy-Magee syndrome, [central core disease](#).

Sicard syndrome, [Collet s.](#)

sicca syndrome, keratoconjunctivitis and xerostomia without connective tissue disease; cf. [Sjögren s.](#)

sick euthyroid syndrome, [euthyroid sick s.](#)

sick sinus syndrome, a syndrome of bradycardia, generally intermittent and sometimes mixed with episodes of atrial tachyarrhythmias (see [bradycardia-tachycardia s.](#)) or periods of sinus arrest, due to malfunction originating in the supraventricular portion of the cardiac conduction system.



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Sick sinus syndrome; the monitor lead shows sinus bradycardia with a long sinus pause.

Silfverskiöld syndrome, a form of ecto-osteochondrodysplasia in which the skeletal changes are chiefly in the extremities and which is inherited as a dominant character.

Silver syndrome, Silver-Russell syndrome, a syndrome consisting of low birth weight despite normal length of gestation, short stature, lateral asymmetry, and slight to moderate increase in excretion of gonadotropins, which may be associated with incurved fifth fingers, café-au-lait spots, syndactyly, triangular face, downturned corners of the mouth, and precocious puberty. Called also [Russell s.](#) or [dwarfism, Russell-Silver s.](#) or [dwarfism](#), and [Silver-Russell dwarfism](#).

Silverman syndrome, [Currarino-Silverman s.](#)

Silvestrini-Corda syndrome, a rare syndrome seen in certain persons with [cirrhosis](#), consisting of eunuchoid body type, loss of body hair, decreased libido, and sterility; affected males have atrophy of the testes and [gynecomastia](#), and affected females have menstrual disorders such as [menorrhagia](#) or [amenorrhea](#). It results from abnormally high estrogenic activity due to failure of the liver to inactivate the circulating [estrogens](#).

Simmonds syndrome, [panhypopituitarism](#).

Sipple syndrome, [multiple endocrine neoplasia, type 2A](#).

Sjögren syndrome, a symptom complex of unknown etiology, usually occurring in middle-aged or older women, marked by the triad of keratoconjunctivitis sicca with or without lacrimal gland enlargement, xerostomia with or without salivary gland enlargement, and the presence of a connective tissue disease, usually rheumatoid arthritis but sometimes systemic lupus erythematosus, scleroderma, or polymyositis. An abnormal immune response has been implicated. See also [sicca s.](#)

Sjögren-Larsson syndrome, a congenital syndrome of mental retardation, ichthyosis, and spastic pyramidal symptoms.

sleep apnea syndrome, [sleep apnea](#).

sleeper syndrome, [thromboembolic meningoencephalitis](#).

SLE-like syndrome, see [systemic lupus erythematosus](#), under lupus.

slipping rib syndrome, weakness or rupture of the medial fibrous attachments of the eighth, ninth, or tenth ribs, so that their cartilage tip slips upward and impinges on the intercostal nerve, causing chest pain.

Sluder syndrome, see under [neuralgia](#).

Sly syndrome, a mucopolysaccharidosis caused by deficiency of β -glucuronidase and characterized biochemically by excretion of dermatan sulfate, heparan sulfate, and chondroitin sulfates A and C in the urine and by granular inclusions in granulocytes. Onset is between 1 and 2 years with mild to moderate Hurler-like features including dysostosis multiplex, pectus carinatum, visceromegaly, cardiac murmurs, short stature, and moderate mental retardation. Milder forms exist. Called also [mucopolysaccharidosis VII](#).

Smith-Lemli-Opitz syndrome, a hereditary syndrome, transmitted as an autosomal recessive trait, characterized by multiple congenital anomalies, including microcephaly, mental retardation, hypotonia, incomplete development of male genitalia, short nose with anteverted nostrils, and syndactyly of second and third toes.

Sneddon syndrome, a rare condition in which cerebral arteriopathy and ischemia are accompanied by diffuse noninflammatory [livedo](#)

[reticularis](#).

SO syndrome, [orbital apex s.](#)

social breakdown syndrome, deterioration of social and interpersonal skills, work habits, and behavior in persons confined in institutions for extended periods with little productive activity, such as long-term psychiatric patients or prisoners; it is caused by the effects of long-term institutionalization rather than any diagnosed mental disorder. Symptoms include excessive passivity, assumption of the chronic sick role, withdrawal, and apathy.

Sohval-Soffer syndrome, a congenital syndrome in males, consisting of [hypogonadism](#) with abnormalities of the cervical spine and ribs, as well as mental retardation.

solitary rectal ulcer syndrome, prolapse or internal intussusception of the rectal mucosa with a single ulcer on the anterior aspect 4 to 12 cm from the anal verge, accompanied by constipation and hematochezia; the condition may sometimes be confused with malignancy, infection, or Crohn disease. It is most common in persons 20 to 40 years old who have a history of straining at evacuation.

Solomon syndrome, [sebaceous nevus s.](#)

somnolence syndrome, a transient condition of drowsiness, lethargy, anorexia, and irritability with electroencephalographic changes, occurring in children after irradiation of the head in the treatment of brain tumors, acute leukemia, or [non-Hodgkin lymphoma](#).

Sorsby syndrome, an autosomal dominant condition consisting of bilateral macular [colobomas](#) associated with dystrophic changes of the hands and feet, usually [brachydactyly](#) of the distal two phalanges of certain fingers or toes.

Sotos syndrome, **Sotos syndrome of cerebral gigantism**, [cerebral gigantism](#).

space adaptation syndrome, a form of motion sickness occurring in a weightless environment during space flight, with nausea, vomiting, anorexia, headache, malaise, drowsiness, and lethargy. It is probably caused by conflicting signals concerning motion from the otolith (whose proper function depends on the presence of gravity) and the visual system (which affects the autonomic nervous system). Called also [space sickness](#).

Spanish toxic oil syndrome, name given to an epidemic of acute pneumonia with pulmonary edema, fever, and [pseudoscleroderma](#) (rash, myalgia, and eosinophilia), sometimes with neuromuscular damage or fatal respiratory failure. It occurred in Spain in 1981 after contaminated cooking oil was sold by traveling salesmen. The toxin has not been identified. Called also [toxic oil s.](#)

Spens syndrome, [Adams-Stokes s.](#)

sphenoidal fissure–optic canal syndrome, [orbital apex s.](#)

spherophakia-brachymorphia syndrome, [Weill-Marchesani s.](#)

splenic flexure syndrome, constipation with left upper quadrant pain attributed to kinking of an adhesion between the transverse and descending colon with obstruction; probably a manifestation of the [irritable colon syndrome](#) rather than an organic lesion. Called also [Payr disease](#).

split brain syndrome, an association of symptoms produced by disruption of or interference with the connection between the hemispheres of the brain. See also [split brain](#), under brain.

Sprinz-Dubin syndrome, **Sprinz-Nelson syndrome**, [Dubin-Johnson s.](#)

spun glass hair syndrome, [uncombable hair s.](#)

stagnant loop syndrome, [stasis s.](#)

staphylococcal scalded skin syndrome, an infectious disease of infants, young children, and occasionally older children and adults, seen after infection with certain strains of [Staphylococcus aureus](#) (phage group II), which elaborate [exfoliatin](#) (q.v.). Clinical signs are localized or widespread fine vesicles and bullae that rupture easily to cause exfoliation of large sheets of skin, leaving raw, denuded areas that make the skin surface look scalded. Called also [dermatitis exfoliativa neonatorum](#) and [Ritter disease](#). Cf. [toxic epidermal necrolysis](#).



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Staphylococcal scalded skin syndrome.

stasis syndrome, overgrowth of bacteria within the small intestine resulting from a variety of conditions causing [stasis](#). The most common causes are disturbances to intestinal [motility](#) or decreased acid secretion; it may also be caused by structural abnormalities such as [diverticula](#), [fistulae](#) between the colon and small intestine, or chronic obstruction. Characteristics include malabsorption of vitamin B₁₂,

[steatorrhea](#), and [anemia](#). Called also [bacterial overgrowth s.](#), [blind loop s.](#), and [stagnant loop s.](#)

Stauffer syndrome, a paraneoplastic syndrome seen in patients with [renal cell carcinoma](#), marked by biochemical hepatic abnormalities without hepatic metastasis of the tumor.

Steele-Richardson-Olszewski syndrome, [progressive supranuclear palsy](#).

steely hair syndrome, [Menkes disease](#).

Stein-Leventhal syndrome, [polycystic ovary s.](#)

Steinbrocker syndrome, [shoulder-hand s.](#)

Steiner syndrome, [Curtius s.](#)

Stevens-Johnson syndrome, a syndrome traditionally considered to be a severe form of [erythema multiforme](#). A respiratory prodrome precedes characteristic mucocutaneous lesions and other symptoms. Large areas of the skin and oronasal, genital, and colonic mucous membranes develop macules and become necrotic; hemorrhagic crusts appear on the lips. Ocular lesions may include conjunctivitis, iritis, keratitis, and corneal perforations and opacities leading to blindness. The lungs, heart, kidneys, and gastrointestinal system may also become involved, sometimes with a fatal outcome. Called also [erythema multiforme majus](#).

Stewart-Treves syndrome, lymphangiosarcoma which occurs as a late complication of severe lymphedema of the arm following excision of lymph nodes, usually associated with radical mastectomy.

Stickler syndrome, [hereditary progressive arthro-ophthalmopathy](#).

stiff heart syndrome, any cardiac disease characterized by restrictive hemodynamics; it may result from any pathologic process that renders

the myocardial fibers abnormally rigid or that externally applies a constricting pressure and as a consequence impedes flow of blood into the ventricular cavities.

stiff man syndrome, stiff person syndrome, a condition of unknown etiology characterized by progressive fluctuating rigidity of axial and limb muscles in the absence of signs of cerebral and spinal cord disease but with continuous electromyographic activity; some cases have been linked to autoimmune conditions.

Stilling syndrome, Stilling-Türk-Duane syndrome, Duane s.

Stokes syndrome, Stokes-Adams syndrome, Adams-Stokes s.

Stokvis-Talma syndrome, enterogenous cyanosis.

stomatitis-pneumoenteritis syndrome, peste des petits ruminants.

Strachan syndrome, Strachan-Scott syndrome, a nutritional polyneuropathy of unknown etiology found in impoverished areas of Jamaica and other countries, possibly due to a deficiency in dietary thiamine or riboflavin; characterized by amblyopia, paresthesias, dizziness, glossitis, stomatitis, lesions of the sensory pathways, and various other symptoms.

straight back syndrome, a skeletal deformity characterized by loss of the anterior concavity of the vertebral column in the upper thoracic region, with consequent reduction in the anteroposterior diameter of the thorax and compression of the heart between the dorsal spine and the sternum.

stroke syndrome, a condition with sudden onset caused by acute vascular lesions of the brain, such as infarction from hemorrhage, embolism, or thrombosis, or rupturing aneurysm. It may be marked by any of a variety of symptoms reflecting the focus of infarction or hemorrhage, including hemiparesis, vertigo, numbness, aphasia, and dysarthria; it is often followed by permanent neurologic damage. Called also [cerebrovascular accident](#) and [stroke](#).

Sturge syndrome, Sturge-Kalischer-Weber syndrome, Sturge-Weber s.

Sturge-Weber syndrome, a congenital syndrome of unknown etiology consisting of a [port-wine stain](#) distributed over the [trigeminal nerve](#), usually unilaterally, with a similar vascular disorder of underlying meninges and cerebral cortex. Called also [encephalofacial](#) or [encephalotrigeminal angiomatosis](#), [Sturge](#) or [Sturge-Kalischer-Weber s.](#), and [Weber disease](#).



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Sturge-Weber syndrome.

subclavian steal syndrome, cerebral or brainstem ischemia resulting from vertebrobasilar insufficiency in cases of subclavian steal.

sudden adult death syndrome, sudden arrhythmia death syndrome, sudden arrhythmic death syndrome, any of various conditions in which [sudden cardiac death](#) results from an [arrhythmia](#), usually tachycardia but occasionally severe bradycardia.

sudden infant death syndrome, the sudden and unexpected death of an apparently healthy infant, typically occurring between the ages of three weeks and five months, and not explained by careful postmortem studies; called also [cot death](#) and [crib death](#).

sudden unexplained death syndrome, death for which no underlying cause can be found of a person two years old or older. [Brugada s.](#)

Sudeck-Leriche syndrome, post-traumatic osteoporosis associated with vasospasm.

Sulzberger-Garbe syndrome, exudative discoid and lichenoid dermatitis.

sump syndrome, an occasional complication of [choledochoduodenostomy](#) when the anastomosis becomes stenosed and the [common bile duct](#) acts as a [diverticulum](#); food and other debris collects in the duct, causing infection.

sundown syndrome, sundowning.

superior mesenteric artery syndrome, compression of the third, or transverse, portion of the duodenum against the aorta by the [superior mesenteric artery](#), resulting in complete or partial obstruction that may be chronic, intermittent, or acute; symptoms range from mild to severe, including nausea and vomiting, pain, and extreme distention of the stomach and duodenum.

superior orbital fissure syndrome, deep orbital and unilateral frontal headache with progressive sixth, third, and fourth cranial nerve palsies, with oculomotor paralysis, diminution of the field of vision, and other ocular changes; it occurs either as a result of a meningioma of the sphenoid bone that compresses nearby nerves or as an extension of infection from sphenoid sinusitis into the superior orbital fissure. Cf. [Tolosa-Hunt s.](#)

superior sulcus tumor syndrome, Pancoast s. (def. 1).

superior thoracic aperture syndrome, thoracic outlet s.

superior vena cava syndrome, a complex of symptoms caused by compression of the superior vena cava, such as by a bronchial tumor or by metastatic mediastinal lymph nodes in lung cancer; characteristics include suffusion and brawny edema of the face, neck, or upper arms; central nervous system disturbances; cyanosis; conjunctival edema; and edema of the trachea and esophagus leading to dyspnea and dysphagia.

supine hypotension syndrome, supine hypotensive syndrome, partial occlusion of the [inferior vena cava](#) and the [descending aorta](#) by the uterus, especially when a woman is pregnant, resulting in hypotension when in a [supine position](#); it is corrected by assuming a side-lying position. Called also [vena caval s.](#)

supraspinatus syndrome, tenderness over the supraspinatus tendon, a painful arc on movement of the arm, and a reversal of scapulohumeral rhythm.

supravalvular aortic stenosis syndrome, Williams s.

sweat retention syndrome, the dermatologic symptoms caused by occlusion of [sweat ducts](#); dependent on factors such as extent of blockage, environmental temperature, and duration of the sweating stimulus. See also [miliaria.tropical anhidrotic asthenia](#).

Sweet syndrome, acute febrile neutrophilic dermatosis.

Swyer syndrome, [46,XY gonadal dysgenesis](#).

Swyer-James syndrome, acquired unilateral emphysema, with severe airway obstruction during exhalation, oligemia, and a small hilum; called also [Macleod s.](#)

Sylvian syndrome, **Sylvian aqueduct syndrome**, impairment of vertical gaze, retraction nystagmus, convergence nystagmus, convergence spasms, and poor or absent reaction of the pupils (which are usually of normal size) to light or near vision. It is caused by a neoplasm, inflammation, or vascular lesion adjacent to the periductal gray matter of the aqueduct of Sylvius. Called also [Koeber-Salus-Elschnig s.](#) and [retraction nystagmus](#).

syringomyelic syndrome, [syringomyelia](#).

systolic click-murmur syndrome, [mitral valve prolapse s.](#)

Takayasu syndrome, see under [arteritis](#).

Tapia syndrome, unilateral paralysis of the tongue and larynx, the soft palate being unaffected. It follows injury to the vagus and hypoglossal nerves, most often from trauma or a tumor.

TAR syndrome, [thrombocytopenia-absent radius s.](#)

tarsal tunnel syndrome, a syndrome of overuse injury with a complex of symptoms resulting from compression of the posterior tibial nerve or of the plantar nerves in the tarsal tunnel, with pain, numbness, and tingling paresthesia of the sole of the foot.

Taussig-Bing syndrome, a rare congenital malformation of the heart characterized by transposition of the great vessels and a ventricular septal defect straddled by a large pulmonary artery; hemodynamically it is characterized by pulmonary hypertension, pulmonary plethora, cyanosis, and greater O₂ saturation of blood in the pulmonary artery than in the aorta.

Tay syndrome, a rare autosomal recessive syndrome characterized by [trichothiodystrophy](#), [ichthyosis](#), infertility, developmental delay, and short stature; called also [IBIDS s.](#)

tegmental syndrome, [Benedikt s.](#)

temporomandibular joint syndrome, [temporomandibular disorder](#).

Terry syndrome, [retinopathy of prematurity](#).

Terson syndrome, [vitreous hemorrhage](#) caused by spreading of an [intracranial](#), [subarachnoid](#), or [subdural hemorrhage](#).

testicular feminization syndrome, [complete androgen insensitivity s.](#)

tethered cord syndrome, a congenital anomaly resulting from defective closure of the neural tube; the conus medullaris is abnormally low and is tethered by one or more forms of intradural abnormality such as a short, thickened filum terminale, fibrous bands or adhesions, or an intraspinal lipoma.

thalamic syndrome, **thalamic pain syndrome**, a syndrome caused by a lesion in the thalamus and characterized by contralateral hemianesthesia; some later develop persistent severe pain and choreoathetoid movements on the affected side, mild hemiataxia, and astereognosis. Called also [Dejerine-Roussy s.](#) and [thalamic hyperesthetic anesthesia](#).

Thibierge-Weissenbach syndrome, [calcinosis](#).

Thiele syndrome, tenderness and pain in the region of the lower portion of the sacrum and coccyx, or in contiguous soft tissues and muscles.

Thiemann syndrome, see under [disease](#).

thin ewe syndrome, chronic caseous lymphadenitis in a ewe, with weight loss and reproductive failure.

thoracic insufficiency syndrome, inability of the thorax to support normal respiration and lung growth, resulting from a variety of conditions that cause thoracic deformities, such as progressive [scoliosis](#) or [asphyxiating thoracic dystrophy](#).

thoracic outlet syndrome, any of a variety of neurovascular syndromes resulting from compression of the subclavian artery, the brachial plexus nerve trunks, or less often the axillary vein or subclavian vein, by thoracic outlet abnormalities such as a drooping shoulder girdle, a cervical rib or fibrous band, an abnormal first rib, or occasionally compression of the edge of the scalenus anterior muscle. Continual hyperabduction of the arm may cause another variety ([hyperabduction s.](#)). Arterial compression leads to ischemia, paresthesias, numbness, and weakness of the affected arm, sometimes with Raynaud phenomenon of the arm. Nerve compression causes atrophy and weakness of the muscles of the hand and, in advanced cases, of the forearm, with pain and sensory disturbances in the arm. Venous obstruction usually takes the form of the [Paget-Schroetter syndrome](#). Other types include [cervical rib s.](#), [costoclavicular s.](#), and [scalenus anticus s.](#)

Thorn syndrome, [salt-losing nephropathy](#).

thrombocytopenia-absent radius syndrome, an autosomal recessive syndrome consisting of thrombocytopenia associated with absence or hypoplasia of the radius and sometimes congenital heart disease and renal anomalies. Called also [TAR s.](#)

thromboembolic syndrome, the association between the formation of thrombi in the deep veins of the leg and pulmonary embolism.

Tietze syndrome, idiopathic painful nonsuppurative swellings of one or more costal cartilages, especially of the second rib; the anterior chest pain may mimic that of coronary artery disease. Called also [costal chondritis](#). albinism, except for normal eye pigment, deafness, and hypoplasia of the eyebrows.

TINU syndrome, a rare syndrome of [tubulointerstitial nephritis](#) and [uveitis](#), often with immunologic alterations; called also [Dobrin s.](#)

TMJ syndrome, [temporomandibular disorder](#).

TNF-receptor-associated periodic syndrome, an [autoinflammatory disease](#) (q.v.) of autosomal dominant inheritance, caused by mutations in the gene for [tumor necrosis factor](#) and characterized by febrile attacks lasting one or two days to two weeks. It may be accompanied by myalgia, abdominal pain and gastrointestinal disturbances, headache, testicular pain, conjunctivitis, periorbital edema, pleuritis, erythematous skin lesions, and arthralgia. There is neutrophilia, and serum [C-reactive protein](#) is elevated while the soluble type 1 receptor for [TNF-α](#) is decreased. Called also [tumor necrosis factor receptor-associated periodic s.](#) and [familial Hibernian fever](#).

Tolosa-Hunt syndrome, unilateral ophthalmoplegia associated with pain behind the orbit and in the area supplied by the first division of the trigeminal nerve; it is thought to be due to nonspecific inflammation and granulation tissue in the superior orbital fissure or cavernous sinus. Cf. [cavernous sinus s.](#) and [superior orbital fissure s.](#)

Tommaselli syndrome, see under [disease](#).

TORCH syndrome, any of a group of infections seen in neonates that occurs when organisms causing one of the infections cross the placental barrier; they all have similar symptoms in babies and may be clinically silent in the mothers. Called also [TORCH infection](#).

Torre syndrome, multiple carcinomas, primarily of the gastrointestinal tract, in association with large numbers of sebaceous neoplasms ranging from [sebaceous hyperplasia](#) to [sebaceous carcinoma](#). Called also [Muir-Torre s.](#)

Touraine-Solente-Golé syndrome, [pachydermoperiostosis](#).

Tourette syndrome, [Gilles de la Tourette s.](#)

Townes syndrome, an autosomal dominant syndrome of auricular anomalies, anal defects, limb and digit—particularly thumb—abnormalities, and renal deficiencies; it occasionally includes cardiac disease, deafness, or cystic ovary.

toxic fat syndrome, toxicity in 3- to 10-week old chickens that have been fed diets supplemented with fat containing any of several toxins; symptoms are edema of the pericardium and abdomen, waddling gait, and sudden death.

toxic hypoglycemic syndrome, [Jamaican vomiting sickness](#).

toxic oil syndrome, [Spanish toxic oil s.](#)

toxic shock syndrome, a severe illness caused by a bacterial infection, characterized by high fever of sudden onset, vomiting, diarrhea, and myalgia, followed by hypotension and, in severe cases, [shock](#). A sunburnlike rash with peeling of the skin, especially of the palms and soles, occurs during the acute phase. It was originally observed almost exclusively in menstruating women using tampons, with the infective agent being [Staphylococcus aureus](#), but a nearly identical syndrome has subsequently been seen in males and females of different ages infected with [group A streptococcus](#).

translocation Down syndrome, [Down syndrome](#) in which the excess chromosomal material (the long arm of chromosome 21) is translocated

to another acrocentric chromosome (in standard trisomy 21 there is an additional chromosome 21). A carrier of the translocation chromosome has 45 chromosomes including the translocation chromosome and may be at increased risk of having a child with Down syndrome.

transurethral resection syndrome, severe **hyponatremia** caused by absorption of fluids used to irrigate the bladder during **transurethral resection of the prostate**. Called also **TUR s.**

Treacher Collins syndrome, the incomplete form of **mandibulofacial dysostosis**.

Treacher Collins–Franceschetti syndrome, **mandibulofacial dysostosis**.

trichorhinophalangeal syndrome, an autosomal recessive syndrome consisting of sparse, slowly growing hair, pear-shaped nose with high philtrum, and brachyphalangia with deformity of the fingers and wedge-shaped epiphyses.

triparanol syndrome, alopecia, poliosis, ichthyosis, irreversible cataracts, and impotence, due to the use of triparanol, a drug formerly used to depress the synthesis of cholesterol.

triple-A syndrome, **Allgrove s.**

trismus-pseudocamptodactyly syndrome, a rare autosomal dominant disorder caused by mutation in the MYH8 gene (locus: 17p13.1), which encodes a myosin heavy chain, characterized by inability to open the mouth fully, facultative camptodactyly resulting from shortened finger-flexor tendons, and short stature. Called also **distal arthrogryposis type 7** and **Hecht, Hecht-Beals**, or **Hecht-Beals-Wilson s.**

trisomy 8 syndrome, a syndrome associated with an extra chromosome 8, usually mosaic (trisomy 8/normal), characterized by mild to severe mental retardation, prominent forehead, deep-set eyes, thick lips, prominent ears, and camptodactyly.

trisomy 11q syndrome, a rare syndrome resulting from the presence of an extra long arm of chromosome 11; because different segments may be involved, the associated anomalies are highly variable and include preauricular fistulas, hypoplasia of the gallbladder, micropenis, bicornuate uterus, microphthalmos, malformations of the heart, lung, and brain, seizures, and recurrent infection.

trisomy 13 syndrome, a chromosome aberration in which an extra chromosome 13 causes severe central nervous system defects, such as **arhinencephalia** and **holoprosencephaly**, and mental retardation, together with cleft palate and lip, polydactyly, dermal pattern anomalies, and abnormalities of the heart, viscera, and genitalia. Called also **Patau s.**

trisomy 18 syndrome, a condition characterized by mental retardation, scaphocephaly or other skull abnormality, micrognathia, blepharoptosis, low-set ears, corneal opacities, deafness, webbed neck, short digits, ventricular septal defects, Meckel diverticulum, and other deformities. It is due to the presence of an extra chromosome 18. Called also **Edwards** or **trisomy E s.**

trisomy 21 syndrome, **Down s.**

trisomy 22 syndrome, a rare syndrome due to an extra chromosome 22, characterized typically by mental and growth retardation, microcephaly, low-set or malformed ears, micrognathia, long philtrum, preauricular skin tag or sinus, and congenital heart disease. In males, there is often a small penis or undescended testes.

trisomy C syndrome, **trisomy 8 s.**

trisomy D syndrome, **trisomy 13 s.**

trisomy E syndrome, **trisomy 18 s.**

Troisier syndrome, bronzed cachexia occurring in diabetes associated with hemochromatosis.

tropical splenomegaly syndrome, a syndrome of massive **splenomegaly**, hepatomegaly, anemia, and elevated serum IgM levels that occurs in some areas where malaria is endemic, such as parts of sub-Saharan Africa and New Guinea. Lymphocytic infiltrates are present in hepatic sinusoids, and the association with chronic malarial infection is suggested by a polyclonal increase in IgM, a very high titer of IgM antibodies to **Plasmodium falciparum**, and often by a therapeutic response to malarial chemoprophylaxis. Called also **tropical splenomegaly**.

Trousseau syndrome, **thrombophlebitis migrans** occurring primarily as a **paraneoplastic syndrome** in association with carcinoma of the abdominal viscera, but also with some types of metastatic neoplasms and chemotherapy.

tumor lysis syndrome, severe hyperphosphatemia, hyperkalemia, hyperuricemia, and hypocalcemia occurring after effective induction chemotherapy of rapidly growing malignant neoplasms; thought to be due to release of intracellular products after cell lysis.

tumor necrosis factor receptor–associated periodic syndrome, **TNF-receptor–associated periodic syndrome**.

TUR syndrome, **transurethral resection s.**

Turcot syndrome, familial adenomatous polyposis of the colon associated with malignant tumors (gliomas) of the central nervous system.

Turner syndrome, a disorder of gonadal differentiation in patients phenotypically female, marked by short stature, undifferentiated (streak) gonads, and variable abnormalities that may include webbing of the neck, low posterior hairline, cubitus valgus, and cardiac defects; it is typically associated with absence of the second sex chromosome (XO or 45,X), although structural abnormality of one X chromosome or mosaicism (e.g., XX/XX or X/XXX) may also be responsible. Called also **gonadal dysgenesis**.



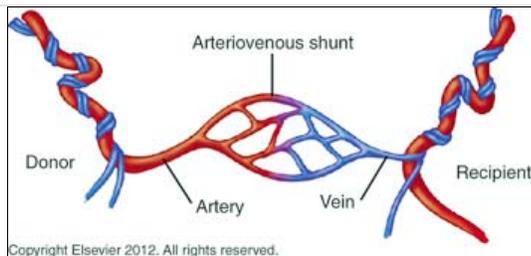
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Turner syndrome; newborn showing webbed neck with low hairline, shield-shaped chest with widespread nipples, abnormal ears, and micrognathia.

Turner syndrome, male, **Noonan s.**

twiddler's syndrome, dislodgement, breakdown, or other malfunction of an artificial cardiac pacemaker, chemotherapy port, drip infusion valve, or similar implanted diagnostic or therapeutic device as a result of unconscious or habitual manipulation by the patient.

twin transfusion syndrome, **twin?twin transfusion syndrome**, a syndrome caused by **twin-to-twin transfusion** (q.v.); the donor twin develops hypovolemia, hypotension, anemia, microcardia, and growth retardation, while the recipient develops hypervolemia, hypertension, polycythemia, cardiomegaly, and congestive heart failure; polyhydramnios frequently occurs.



Twin transfusion syndrome characterized by arteriovenous shunt at a shared placental cotyledon in diamniotic monozygotic twins.

tying-up syndrome, [azoturia](#) (def. 2).

Uberreiter syndrome, [chronic superficial keratitis](#).

Ullrich-Feichtiger syndrome, a condition of micrognathia, hexadactyly, and genital abnormalities, with depressed nose, small eyes, hypertelorism, and protuberant ears, along with other defects.

Ullrich-Turner syndrome, [Noonan s.](#)

uncombable hair syndrome, an abnormality of the hair inherited as an autosomal dominant trait, in which the individual hairs are triangular in cross section, with a longitudinal groove; the hair has a spun-glass appearance and is arranged in bundles that stand out in different directions. Called also [pili canaliculi](#), [pili trianguli et canaliculi](#), and [spun glass hair s.](#)

unilateral nevoid telangiectasia syndrome, [unilateral nevoid telangiectasia](#).

Unna-Thost syndrome, [nonepidermolytic palmoplantar keratoderma](#).

upper airway resistance syndrome, an incomplete form of [obstructive sleep apnea](#) in which the upper airway resists air flow and becomes partially obstructed during sleep.

uremic syndrome, the spectrum of symptoms accompanying [uremia](#); see [uremia](#) (def. 2).

urethral syndrome, a nonspecific group of symptoms considered to arise from a urethral problem other than urinary tract infection; it may include suprapubic aching and cramping, [urinary frequency](#), and such bladder complaints as [dysuria](#), straining on urination, and low back pain.

urofacial syndrome, a rare hereditary syndrome, usually autosomal recessive, consisting of hydronephrosis, hydroureter, and an inverted facial expression, so that affected children appear to be crying when they try to smile. Called also [Ochoa s.](#)

Usher syndrome, an autosomal recessive syndrome in which congenital deafness is accompanied by retinitis pigmentosa, often ending in blindness; sometimes mental retardation and disturbances of gait also occur.

vagoaccessory syndrome, [Schmidt s.](#) (def. 1).

vagoaccessory-hypoglossal syndrome, [Jackson s.](#)

Vail syndrome, [vidian neuralgia](#).

van Bogaert-Nyssen syndrome, **van Bogaert-Nyssen-Peiffer syndrome**, the adult form of [metachromatic leukodystrophy](#).

van Buchem syndrome, [hyperostosis corticalis generalisata](#).

Van der Woude syndrome, an autosomal dominant syndrome consisting of cleft lip with or without cleft palate, with cysts of the lower lip.

vanishing bile duct syndrome, progressive destruction of intrahepatic bile ducts with cholestasis; some cases are a reaction to medications, and others are idiopathic.

vanishing testes syndrome, a disorder in males characterized by absence of testes and gonadal tissue (usually unilateral but sometimes bilateral) and a small penis; when it is bilateral the individual will not undergo puberty or adolescent [masculinization](#) without [testosterone](#) supplements. The testes are thought to have been present in the embryo but to have "vanished" before completion of male sexual differentiation. Called also [embryonic testicular regression s.](#)

vanishing twin syndrome, the disappearance of one fetus following the sonographic diagnosis of a twin pregnancy, with only one twin eventually being delivered; the vanished twin may be resorbed or incorporated into the placental membrane.

Van Lohuizen syndrome, [cutis marmorata telangiectatica congenita](#).

vascular syndrome, any syndrome due to occlusion or stenosis of vessels supplying the nervous system.

vascular leak syndrome, a disorder caused by extravasation of plasma through vessel walls, causing edema in surrounding tissues; the most common cause is a reaction to therapy with interleukin-2.

VCF syndrome, **velocardiofacial syndrome**, a rare autosomal dominant syndrome of cardiac defects and characteristic craniofacial abnormalities including cleft palate, jaw abnormalities, and prominent nose. Learning disabilities occur often; short stature, slender hyperextensible hands and digits, scoliosis, mental retardation, inguinal hernia, auricular abnormalities, and microcephaly occur less frequently. It is a phenotype of [22q11 deletion syndrome](#) (q.v.). Called also [Shprintzen](#) or [Shprintzen-Goldberg s.](#)

vena caval syndrome, [supine hypotension s.](#)

Verner-Morrison syndrome, a rare syndrome of profuse watery diarrhea, hypokalemia, and achlorhydria, usually associated with excess levels of [vasoactive intestinal polypeptide](#) resulting from a [VIPoma](#) in the pancreas; called also [diarrheogenic s.](#), [pancreatic cholera](#), [pancreatic cholera s.](#), and [WDHA s.](#)

Vernet syndrome, paralysis of the ninth, tenth, and eleventh cranial nerves due to a lesion in the region of the jugular foramen, and marked by paralysis of the superior constriction of the pharynx and difficulty in swallowing solids; paralysis of the soft palate and fauces with anesthesia of these parts and of the pharynx, and loss of taste in the posterior third of the tongue; paralysis of the vocal cords and anesthesia of the larynx; and paralysis of the sternocleidomastoid and trapezius muscles. Called also [jugular foramen s.](#)

vertebasilar syndrome, see under [insufficiency](#).

Villaret syndrome, unilateral paralysis of the ninth, tenth, eleventh, and twelfth cranial nerves and sometimes the seventh, due to a lesion behind the parotid glands. This causes paralysis of the superior constriction of the pharynx and difficulty swallowing solids; paralysis of soft palate and fauces with anesthesia there and in the pharynx; loss of taste in the posterior third of the tongue; paralysis of the vocal cords with anesthesia of the larynx; paralysis of the sternocleidomastoid and trapezius; and paralysis of the cervical sympathetic nerves ([Horner syndrome](#)).

Vinson syndrome, [Plummer-Vinson s.](#)

Virchow-Seckel syndrome, [Seckel s.](#)

vitamin E-selenium deficiency syndrome, a deficiency disease of pigs whose diet is low in vitamin E and selenium, most commonly rapidly growing, recently weaned piglets. It usually manifests as either hepatitis dietetica or mulberry heart disease, and affected animals may die suddenly during exercise.

Vogt syndrome, a syndrome associated with birth trauma, characterized by bilateral athetosis, walking difficulties, spasmodic outbursts of laughing or crying, speech disorders, excessive myelination of the nerve fibers of the corpus striatum, giving it a marbled appearance (status marmoratus), and sometimes mental deficiency. Called also [s. of corpus striatum](#).

Vogt-Koyanagi syndrome, uveomeningitis characterized by exudative iridocyclitis and choroiditis associated with patchy depigmentation of the skin and hair; the lashes and eyebrows also become whitened, and there may also be retinal detachment and associated deafness and tinnitus. Cf. [Vogt-Koyanagi-Harada s.](#)

Vogt-Koyanagi-Harada syndrome, bilateral uveitis with exudative iridocyclitis, choroiditis, meningism, and temporary or permanent retinal

detachment, occurring in association with alopecia, vitiligo, poliosis, loss of visual acuity, headache, vomiting, deafness, and sometimes vertigo or glaucoma. The syndrome may be an inflammatory autoimmune disorder. Called also [Harada s.](#)

Vohwinkel syndrome, an autosomal dominant, progressive form of [palmoplantar keratoderma](#) that begins in childhood, characterized by a stellate pattern of [hyperkeratosis](#) on the backs of the hands and feet, linear keratoses on elbows and knees, and annular constrictions of digits like those of [ainhum](#). In some patients it is associated with scarring alopecia and deafness. Called also [keratoderma](#) or [keratoma hereditarium mutilans](#).

Volkman syndrome, see under [contracture](#).

vulvar vestibulitis syndrome, severe pain and tenderness of the vulvar vestibule upon touch or attempted vaginal entry; a major cause of [dyspareunia](#).

Waardenburg syndrome, any of several hereditary conditions characterized principally by a combination of auditory and pigmentary abnormalities, including hypopigmented or heterochromatic irides and sometimes hypopigmented fundi; pigmentation defects of the skin and hair, particularly [white forelock](#), white eyelashes, and [leukoderma](#); and [cochlear hearing loss](#). The underlying cause is disruption of various genes associated with the development, migration, or differentiation of melanocytes and neural crest cells; four types have been distinguished on clinical and genetic bases.

type 1, an autosomal dominant form caused by mutation in the transcription factor gene [PAX3](#), and additionally characterized by wide bridge of the nose due to [dystopia canthorum](#).



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Waardenburg syndrome type 1.

type 2, an autosomal dominant form similar to [type 1](#), but lacking [dystopia canthorum](#). It is subdivided into types 2A and 2B; mutation in the transcription factor gene MITF causes type 2A, but the mutation associated with type 2B has not been identified.

type 3, a form clinically similar to [type 1](#) but characterized also by upper limb defects. It is caused by mutation of the transcription factor gene [PAX3](#), and autosomal dominant and recessive inheritance have both been observed. Called also [Klein-Waardenburg s.](#)

type 4, a form clinically similar to [type 2](#), but characterized also by [Hirschsprung disease](#). It may be either autosomal dominant, caused by mutation in the transcription factor gene [SOX10](#), or autosomal recessive, caused by mutation in the gene encoding either [endothelin-3](#) or a specific receptor for it.

Waardenburg-Shah syndrome, [Waardenburg s. type 4](#).

WAGR syndrome, a syndrome of Wilms' tumor, aniridia, genitourinary abnormalities or gonadoblastoma, and mental retardation, due to a small interstitial deletion of the p13 region of chromosome 11.

Walker-Warburg syndrome, a congenital syndrome, usually fatal before the age of one year, consisting of hydrocephalus, agyria, various ocular anomalies such as retinal dysplasia, corneal opacity, and microphthalmia, and sometimes an encephalocele. Called also [Walker lissencephaly](#), [HARD s.](#), and [Warburg s.](#)

Wallenberg syndrome, a syndrome due usually to occlusion of the vertebral artery, and less often to occlusion of its branch, the posterior inferior cerebellar artery; marked by ipsilateral loss of temperature and pain sensations of the face and contralateral loss of these sensations of the extremities and trunk, ipsilateral ataxia, dysphagia, dysarthria, nystagmus, and [Horner syndrome](#). Called also [lateral medullary s.](#) and [posterior inferior cerebellar artery s.](#)

Warburg syndrome, [Walker-Warburg s.](#)

Ward-Romano syndrome, [Romano-Ward s.](#)

wasting syndrome, the [wasting](#) condition seen in persons infected with the [human immunodeficiency virus](#) (HIV), consisting of weight loss, muscle wasting, and often fever, due to a variety of causes including nutritional deficiencies and chronic diarrhea. Called also [HIV wasting s.](#)

Waterhouse-Friderichsen syndrome, a fulminating complication of [meningococemia](#), with bilateral adrenal hemorrhages, characterized by sudden onset and short course, cyanosis with petechial hemorrhages of the skin and mucous membranes, fever, and hypotension that can lead to [shock](#) and coma.

Watson syndrome, a rare, autosomal dominant condition characterized by [pulmonary stenosis](#), [café au lait spots](#), subnormal intelligence, and sometimes [neurofibromas](#); some authorities consider it a variant of [neurofibromatosis 1](#).

WDHA syndrome, [Verner-Morrison s.](#)

WDHH syndrome, [Verner-Morrison s.](#)

Weber syndrome, a syndrome caused by a lesion in a cerebral peduncle, with paralysis of the [oculomotor nerve](#) on the same side as the lesion, producing ptosis, strabismus, and loss of light reflex and of accommodation, as well as spastic hemiplegia on the side opposite the lesion with increased reflexes and loss of superficial reflexes. Called also [alternating oculomotor hemiplegia](#) and [Weber paralysis](#).

Weber-Cockayne syndrome, the localized form of [epidermolysis bullosa simplex](#).

Weber-Gubler syndrome, [Weber-Leyden syndrome](#), [Weber s.](#)

Wegener syndrome, see under [granulomatosis](#).

Weil syndrome, a severe form of [leptospirosis](#) characterized by jaundice usually with azotemia, hemorrhages, anemia, disturbances of consciousness, and fever. It is usually caused by [Leptospira interrogans serovar icterohaemorrhagiae](#) but may be caused by other serovars. Called also [Fiedler disease](#), [icterohaemorrhagic leptospirosis](#), and [infectious or leptospiral jaundice](#).

Weill-Marchesani syndrome, a congenital disorder of connective tissue transmitted as an autosomal dominant or recessive trait, characterized by brachycephaly, brachydactyly, short stature with a broad chest and heavy musculature, reduced joint mobility, spherophakia, ectopia lentis, myopia, and glaucoma; called also [dystrophia mesodermalis congenita hyperplastica](#), [Marchesani s.](#), and [spherophakia-brachymorphia s.](#)

Weingarten syndrome, [tropical eosinophilia](#).

Wellens syndrome, electrocardiographic signs indicating critical left anterior descending artery stenosis in patients with unstable angina: biphasic or symmetric, often deeply inverted, T waves in the anterior precordial leads, no pathological precordial Q waves, little or no ST segment elevation, no loss of precordial R waves, and little or no cardiac enzyme elevation.

Wells syndrome, [cellulitis](#) with erythema, edema, and often blistering of the skin accompanied by eosinophilia, [flame figures](#), and a mild fever;

a single episode lasts 2 to 6 weeks and recurrences or exacerbations are common. Called also [eosinophilic cellulitis](#).

Wermer syndrome, [multiple endocrine neoplasia, type 1](#).

Werner syndrome, an autosomal recessive syndrome of premature aging in the adult, caused by mutations in the RECQL2 gene (locus: 8p12-p11.2), which encodes a helicase important in maintaining genome stability. Characteristics include short stature with slender limbs and a stocky trunk, scleroderma-like skin changes (especially on the limbs), cataracts, subcutaneous calcification, muscular atrophy, a tendency to diabetes mellitus, prematurely aged face, canities, baldness, and a high incidence of malignancy.

Wernicke-Korsakoff syndrome, the behavioral disorder caused by thiamine deficiency, most commonly due to chronic alcohol abuse and associated with other nutritional polyneuropathies. [Wernicke encephalopathy](#) (confusion, ataxia of gait, nystagmus, and ophthalmoplegia) occurs as an acute attack and is reversible, except for some residual ataxia or nystagmus, by administration of thiamine; [Korsakoff syndrome](#) (severe [anterograde](#) and [retrograde amnesia](#)) may occur in conjunction with Wernicke encephalopathy or may become apparent later; only about 20 per cent of patients recover completely from the amnesia.

West syndrome, [infantile spasms](#).

Weyers oligodactyly syndrome, a congenital syndrome consisting of deficiency of the ulna and ulnar rays, antecubital pterygia, reduced sternal segments, malformations of the kidney and spleen, and cleft lip and palate.

whiplash shake syndrome, a constellation of injuries to the brain and eye that may occur when a very young child is shaken vigorously while being held by the trunk or limbs with the head unsupported. This causes stretching and tearing of the cerebral blood vessels and brain substance, often with subdural hematomas and retinal hemorrhages, and sometimes with cerebral contusion. It may result in blindness and other visual disturbances, as well as paralysis, convulsions, and even death.

whistling face syndrome, whistling face?windmill vane hand syndrome, [Freeman-Sheldon s.](#)

white clot syndrome, [heparin-induced thrombocytopenia](#).

Widal syndrome, former name for [hemolytic anemia](#).

Wilkie syndrome, [superior mesenteric artery s.](#)

Willebrand syndrome, [von Willebrand disease](#).

Williams syndrome, Williams-Beuren syndrome, a neurodevelopmental disorder caused by a deletion on chromosome 7, characterized by [supravalvular aortic stenosis](#), mental retardation, [elfin facies](#), transient [idiopathic hypercalcemia](#), musculoskeletal defects, growth deficiency, hypersensitivity to sound, visual impairment, and a hoarse voice; sometimes inherited in an autosomal recessive pattern. Called also [elfin facies s.](#)

Williams-Campbell syndrome, congenital bronchomalacia and bronchiectasis, resulting from absence of annular cartilage distal to the first division of the peripheral bronchi.

Wilson-Mikity syndrome, a rare form of pulmonary insufficiency in low-birth-weight infants, marked by hyperpnea and cyanosis of insidious onset during the first month of life and often resulting in death. Radiographically, there are multiple cystlike foci of hyperaeration throughout the lung with coarse thickening of the interstitial supporting structures. The disorder has been attributed to disparity of maturation of parenchymal elements, especially of alveoli proliferation, and hence has been called [pulmonary dysmaturity](#).

Winchester syndrome, an autosomal recessive syndrome consisting of short stature, joint contractures, osteoporosis, corneal opacities, and changes in the joints resembling rheumatoid arthritis.

Winter syndrome, a congenital syndrome consisting of renal hypoplasia or aplasia, anomalies of the internal genitalia, especially vaginal atresia, and anomaly of the ossicles of the middle ear.

Wiskott-Aldrich syndrome, an X-linked [immunodeficiency](#) syndrome characterized by [eczema](#), [thrombocytopenia](#), and recurrent pyogenic infection. Patients cannot produce antibodies to polysaccharide antigens and have increased susceptibility to infection with encapsulated bacteria ([Haemophilus influenzae](#), [meningococcus](#), [pneumococcus](#)). Typically IgM is low, IgA and IgE are elevated, and there is [anergy](#) of the skin. Many affected persons also have [lymphoreticular disorders](#). Called also [Aldrich s.](#)

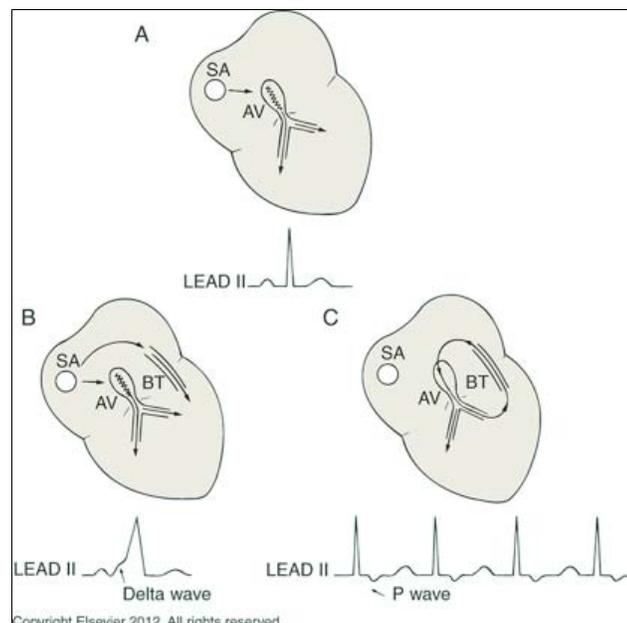
withdrawal syndrome, [substance withdrawal](#).

wobbler syndrome, in large to giant dogs, malformation of the lower cervical vertebrae with compression of the spinal cord so that the animal has ataxia of the hind limbs and a swaying gait; it may progress to paralysis. Called also [wobbles](#).in young horses, incoordination of the hind legs with a swaying gait progressing to stumbling, inability to walk, and sometimes paralysis; causes are varied and include stenosis of the spinal canal, malformation of the cervical vertebrae with demyelination, and inflammation of the cord. Called also [equine sensory ataxia](#) and [wobbles](#).

Wohlfart-Kugelberg-Welander syndrome, [Kugelberg-Welander s.](#)

Wolf-Hirschhorn syndrome, a syndrome associated with partial deletion of the short arm of chromosome 4, characterized by microcephaly, [ocular hypertelorism](#), epicanthus, cleft palate, micrognathia, low-set ears simplified in form, cryptorchidism, and hypspadias.

Wolff-Parkinson-White syndrome, the association of [paroxysmal tachycardia](#) or [atrial fibrillation](#) with [preexcitation](#); the electrocardiogram displays a short P-R interval and a wide QRS complex, usually with an early QRS vector ([delta wave](#)). The term is sometimes used synonymously with [preexcitation s.](#) Called also [WPW s.](#)



Wolff-Parkinson-White (WPW) syndrome. (A), Normal sinus rhythm, traveling from the sinoatrial (SA) node to the atrioventricular (AV) node and then down the bundle branches, with physiologic slowing in the AV node (jagged line). (B), In sinus rhythm in WPW syndrome, conduction down the bypass tract (BT) preexcites the ventricles ahead of impulse arrival at the AV node; the PR interval is short and the QRS complex is wide, with slurring at its onset (delta wave). (C), Atrioventricular reentrant tachycardia (AVRT) in WPW syndrome, caused by spread of the premature atrial beat back up the bypass tract to create a repeating reentrant loop; characterized by a normal QRS complex and a negative P wave in lead II.

Wolfram syndrome, an autosomal recessive syndrome, first evident in childhood, consisting of diabetes mellitus, diabetes insipidus, optic atrophy, and neural deafness. Called also [DIDMOAD s.](#)

WPW syndrome, [Wolff-Parkinson-White s.](#)

Wright syndrome, [hyperabduction s.](#) a condition marked by multifocal areas of osteitis fibrosa, patchy cutaneous pigmentation, and precocious puberty.

Wyburn-Mason syndrome, arteriovenous aneurysms on one or both sides of the brain, with ocular anomalies, especially in the retina, facial nevi, and sometimes mental retardation.

syndrome X, [cardiac s. X. metabolic s.](#)

X-linked lymphoproliferative syndrome, an X-linked immunodeficiency disorder caused by mutation in either the SH2D2A or XIAP gene (both at locus Xq25), which are involved in T-cell development and apoptosis; the disorder is characterized by defective cellular or humoral immune response to infection with [Epstein-Barr virus](#). Fulminant [infectious mononucleosis](#), fatal B cell malignancies, or [hypogammaglobulinemia](#) can result from viral infection. Called also [Duncan s.](#) and [Purtilo s.](#)

XXY syndrome, [Klinefelter s.](#)

yellow nail syndrome, a syndrome consisting of a yellow to greenish discoloration of the nails, which may be smooth, thickened, excessively curved on the long axis, and slow growing, and may become loose and be shed. It is often associated with [lymphedema](#), especially of the lower limbs.

Young syndrome, obstructive [azoospermia](#) associated with chronic respiratory infections of sinuses and lungs.

Zellweger syndrome, [cerebrohepato renal s.](#)

Zieve syndrome, a syndrome of [hypercholesterolemia](#), [hepatosplenomegaly](#), [fatty liver](#), [hemolytic anemia](#), and [hypertriglyceridemia](#) seen in alcoholics.

Zinsser-Cole-Engman syndrome, [dyskeratosis congenita](#).

Zollinger-Ellison syndrome, a triad comprising extreme [gastric hyperacidity](#); [peptic ulcers](#) that are intractable and sometimes fulminating; and [gastrinomas](#) (gastrin-secreting [islet cell tumors](#)) that may appear outside the pancreas, such as in the duodenum. See also [multiple endocrine neoplasia, type 1](#).

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