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**disease** /dis·ease/ (dī-zēz') any deviation from or interruption of the normal structure or function of any body part, organ, or system that is manifested by a characteristic set of symptoms and signs and whose etiology, pathology, and prognosis may be known or unknown. See also entries under [syndrome](#).

**acquired cystic disease of kidney** the development of cysts in the formerly noncystic failing kidney in [end-stage renal disease](#).

**Addison's disease** bronzelike pigmentation of the skin, severe prostration, progressive anemia, low blood pressure, diarrhea, and digestive disturbance, due to adrenal hypofunction.

**Albers-Schönberg disease** [osteopetrosis](#).

**allogeneic disease** graft-versus-host reaction occurring in immunosuppressed animals receiving injections of allogeneic lymphocytes.

**Alpers' disease** a rare disease of young children, characterized by neuronal deterioration of the cerebral cortex and elsewhere, progressive mental deterioration, motor disturbances, seizures, and early death.

**alpha chain disease** heavy chain disease characterized by plasma cell infiltration of the lamina propria of the small intestine resulting in malabsorption with diarrhea, abdominal pain, and weight loss, possibly accompanied by pulmonary involvement.

**Alzheimer's disease** progressive degenerative disease of the brain, of unknown cause; characterized by diffuse atrophy throughout the cerebral cortex with distinctive histopathological changes.

**Andersen's disease** [glycogen storage d., type IV](#).

**apatite deposition disease** a connective tissue disorder marked by deposition of hydroxyapatite crystals in one or more joints or bursae.

**Aran-Duchenne disease** [spinal muscular atrophy](#).

**arteriosclerotic cardiovascular disease** (ASCVD) atherosclerotic involvement of arteries to the heart and to other organs, resulting in debility or death; sometimes used specifically for ischemic heart disease.

**arteriosclerotic heart disease** (ASHD) [ischemic heart d.](#)

**autoimmune disease** any of a group of disorders in which tissue injury is associated with humoral or cell-mediated responses to the body's own constituents; they may be systemic or organ-specific.

**Ayerza's disease** [polycythemia vera](#) with chronic cyanosis, dyspnea, bronchitis, bronchiectasis, hepatosplenomegaly, bone marrow hyperplasia, and pulmonary artery sclerosis.

**Banti's disease** [congestive splenomegaly](#).

**Barlow disease** scurvy in infants.

**Barraquer's disease** [partial lipodystrophy](#).

**Basedow's disease** [Graves' d.](#)

**Batten disease** , **Batten-Mayou disease**

1. [Vogt-Spielmeyer d.](#)

2. more generally, any or all of the group of disorders constituting [neuronal ceroid lipofuscinosis](#).

**Bayle's disease** [general paresis](#).

**Bazin's disease** [erythema induratum](#).

**Bekhterev's (Bechterew's) disease** [ankylosing spondylitis](#).

**Benson's disease** [asteroid hyalosis](#).

**Berger's disease** [IgA glomerulonephritis](#).

**Bernhardt's disease** , **Bernhardt-Roth disease** [meralgia paresthetica](#).

**Besnier-Boeck disease** [sarcoidosis](#).

**Best's disease** [congenital macular degeneration](#).

**Bielschowsky-Janský disease** [Janský-Bielschowsky d.](#)

**Binswanger's disease** a degenerative dementia of presenile onset caused by demyelination of the subcortical white matter of the brain.

**black disease** a fatal disease of sheep, and sometimes of humans, in the United States and Australia, due to [Clostridium novyi](#), marked by necrotic areas in the liver.

**Blocq's disease** [astasia-abasia](#).

**Blount disease** [tibia vara](#).

**Boeck's disease** [sarcoidosis](#).

**Bornholm disease** [epidemic pleurodynia](#).

**Bowen's disease** a squamous cell carcinoma in situ, often due to prolonged exposure to arsenic; usually occurring on sun-exposed areas of skin. The corresponding lesion on the glans penis is termed erythroplasia of Queyrat.

**Brill's disease** [Brill-Zinsser d.](#)

**Brill-Symmers disease** [giant follicular lymphoma](#).

**Brill-Zinsser disease** mild recrudescence of [epidemic typhus](#) years after the initial infection, because [Rickettsia prowazekii](#) has persisted in body tissue in an inactive state, with humans as the reservoir.

**broad beta disease** familial dysbetalipoproteinemia; named for the electrophoretic mobility of the abnormal chylomicron and very-low-density lipoprotein remnants produced.

**Busse-Buschke disease** [cryptococcosis](#).

**Caffey's disease** [infantile cortical hyperostosis](#).

**calcium hydroxyapatite deposition disease** [apatite deposition d.](#)

**calcium pyrophosphate deposition disease** (CPDD) an acute or chronic inflammatory arthropathy caused by deposition of calcium pyrophosphate dihydrate (CPPD) crystals in the joints, chondrocalcinosis, and crystals in the synovial fluid. Acute attacks are sometimes called [pseudogout](#).

**Calvé-Perthes disease** osteochondrosis of capitular epiphysis of femur.

**Camurati-Engelmann disease** [diaphyseal dysplasia](#).

**Canavan disease** , **Canavan-van Bogaert-Bertrand disease** spongy degeneration of the central nervous system.

**Carrión's disease** [bartonellosis](#).

**Castleman disease** a benign or premalignant condition resembling lymphoma but without recognizable malignant cells; there are isolated masses of lymphoid tissue and lymph node hyperplasia, usually in the abdominal or mediastinal area.

**cat-scratch disease** a usually benign, self-limited disease of the regional lymph nodes, caused by [Bartonella henselae](#) and characterized by a papule or pustule at the site of a cat scratch, subacute painful regional lymphadenitis, and mild fever.

**celiac disease** a malabsorption syndrome precipitated by ingestion of gluten-containing foods, with loss of villous structure of the proximal intestinal mucosa, bulky, frothy diarrhea, abdominal distention, flatulence, weight loss, and vitamin and electrolyte depletion.

**Chagas disease** trypanosomiasis due to [Trypanosoma cruzi](#); its course may be acute, subacute, or chronic.

**Charcot-Marie-Tooth disease** muscular atrophy of variable inheritance, beginning in the muscles supplied by the peroneal nerves and progressing to those of the hands and arms.

**cholesteryl ester storage disease** (CESD) a lysosomal storage disease due to deficiency of lysosomal cholesterol esterase, variably characterized by some combination of hepatomegaly, hyperbetalipoproteinemia, and premature atherosclerosis.

**Christmas disease** [hemophilia B](#).

**chronic granulomatous disease** frequent, severe infections of the skin, oral and intestinal mucosa, reticuloendothelial system, bones, lungs, and genitourinary tract associated with a genetically determined defect in the intracellular bactericidal function of leukocytes.

**chronic obstructive pulmonary disease** (COPD) any disorder marked by persistent obstruction of bronchial air flow.

**Coats' disease** [exudative retinopathy](#).

**collagen disease** any of a group of diseases characterized by widespread pathologic changes in connective tissue; they include lupus erythematosus, dermatomyositis, scleroderma, polyarteritis nodosa, thrombotic purpura, rheumatic fever, and rheumatoid arthritis. Cf. [collagen disorder](#).

**communicable disease** a disease the causative agents of which may pass or be carried from one person to another directly or indirectly.

**Concato's disease** progressive malignant polyserositis with large effusions into the pericardium, pleura, and peritoneum.

**constitutional disease** one involving a system of organs or one with widespread symptoms.

**Cori's disease** [glycogen storage d., type III](#).

**coronary artery disease** (CAD) atherosclerosis of the coronary arteries, which may cause angina pectoris, myocardial infarction, and sudden death; risk factors include hypercholesterolemia, hypertension, smoking, diabetes mellitus, and low levels of high-density lipoproteins.

**coronary heart disease** (CHD) [ischemic heart d.](#)

**Cowden disease** a hereditary disease marked by multiple ectodermal, mesodermal, and endodermal nevoid and neoplastic anomalies.

**Creutzfeldt-Jakob disease** a rare [prion disease](#) existing in sporadic, familial, and infectious forms, with onset usually in middle life, and having a wide variety of clinical and pathological features. The most commonly seen are spongiform degeneration of neurons, neuronal loss, [gliosis](#), and amyloid plaque formation, accompanied by rapidly progressive dementia, [myoclonus](#), motor disturbances, and encephalographic changes, with death occurring usually within a year of onset.

**Crigler-Najjar disease** see under [syndrome](#).

**Crohn's disease** [regional enteritis](#); a chronic granulomatous inflammatory disease usually in the terminal ileum with scarring and thickening of the wall, often leading to intestinal obstruction and formation of fistulas and abscesses.

**Crouzon's disease** [craniofacial dysostosis](#).

**Cruveilhier's disease** [spinal muscular atrophy](#).

**Cushing's disease** Cushing's syndrome in which the hyperadrenocorticism is secondary to excessive pituitary secretion of adrenocorticotrophic hormone.

**cystic disease of breast** mammary dysplasia with formation of [blue dome cysts](#).

**cytomegalic inclusion disease** , **cytomegalovirus disease** an infection due to cytomegalovirus and marked by nuclear inclusion bodies in enlarged infected cells. In the congenital form, there is hepatosplenomegaly with cirrhosis, and microcephaly with mental or motor retardation. Acquired disease may cause a clinical state similar to infectious mononucleosis. When acquired by blood transfusion, postperfusion syndrome results.

**deficiency disease** a condition caused by dietary or metabolic deficiency, including all diseases due to an insufficient supply of essential nutrients.

**degenerative joint disease** [osteoarthritis](#).

**Dejerine's disease** , **Dejerine-Sottas disease** [progressive hypertrophic neuropathy](#).

**demyelinating disease** any condition characterized by destruction of the myelin sheaths of nerves.

**disappearing bone disease** gradual resorption of a bone or group of bones, sometimes associated with multiple hemangiomas, usually in children or young adults and following trauma.

**diverticular disease** a general term including the prediverticular state, diverticulosis, and diverticulitis.

**Duchenne's disease**

1. [spinal muscular atrophy](#).
2. [progressive bulbar paralysis](#).
3. [tabes dorsalis](#).
4. [Duchenne's muscular dystrophy](#).

**Duchenne-Aran disease** [spinal muscular atrophy](#).

**Duhring's disease** [dermatitis herpetiformis](#).

**Dukes' disease** a febrile disease of childhood marked by an exanthematous eruption, probably due to a virus of the Coxsackie-ECHO group.

**Durand-Nicolas-Favre disease** [lymphogranuloma venereum](#).

**Duroziez's disease** [congenital mitral stenosis](#).

**Ebola virus disease** fatal acute hemorrhagic fever resembling [Marburg virus disease](#) but caused by Ebola virus, seen in the Sudan and Zaire.

**Ebstein's disease** see under [anomaly](#).

**end-stage renal disease** chronic irreversible [renal failure](#).

**Erb's disease** [Duchenne's muscular dystrophy](#).

**Erb-Goldflam disease** [myasthenia gravis](#).

**Eulenburg's disease** [paramyotonia congenita](#).

**extrapyramidal disease** any of a group of clinical disorders marked by abnormal involuntary movements, alterations in muscle tone, and postural disturbances; they include parkinsonism, chorea, athetosis, etc.

**Fabry's disease** an X-linked lysosomal storage disease of glycosphingolipid catabolism resulting from deficiency of  $\alpha$ -galactosidase A and leading to accumulation of ceramide trihexoside in the cardiovascular and renal systems.

**Farber's disease** a lysosomal storage disease due to defective ceramidase and characterized by hoarseness, aphonia, dermatitis, bone and joint deformities, granulomatous reaction, and psychomotor retardation.

**Fazio-Londe disease** a rare type of progressive bulbar palsy occurring in childhood.

**Feer disease** [acrodynia](#).

**fibrocystic disease of breast** a form of mammary dysplasia with formation of cysts of various size containing a semitransparent, turbid fluid that imparts a brown to blue color to the unopened cysts; believed due to abnormal hyperplasia of the ductal epithelium and dilatation of the ducts of the mammary gland, resulting from exaggeration and distortion of normal menstrual cycle-related breast changes.

**fibrocystic disease of the pancreas** [cystic fibrosis](#).

**fifth disease** [erythema infectiosum](#).

**flint disease** [chalicosis](#).

**floating beta disease** [familial dysbetalipoproteinemia](#).

**focal disease** a localized disease.

**foot-and-mouth disease** an acute, contagious viral disease of wild and domestic cloven-footed animals and occasionally humans, marked by vesicular eruption on the lips, buccal cavity, pharynx, legs, and feet.

**Forbes' disease** [glycogen storage d., type III](#).

**fourth disease** [Dukes' d](#).

**fourth venereal disease** [granuloma inguinale](#).

**Fox-Fordyce disease** a persistent and recalcitrant, itchy, papular eruption, chiefly of the axillae and pubes, due to inflammation of apocrine sweat glands.

**Freiberg's disease** osteochondrosis of the head of the second metatarsal bone.

**Friedländer's disease** [endarteritis obliterans](#).

**Friedreich's disease** [paramyoclonus multiplex](#).

**functional disease** see under [disorder](#).

**Garré's disease** [sclerosing nonsuppurative osteomyelitis](#).

**gastroesophageal reflux disease** (GERD) any condition resulting from gastroesophageal reflux, characterized by heartburn and regurgitation; see also [reflux esophagitis](#).

**Gaucher's disease** a hereditary disorder of glucocerebroside metabolism, marked by the presence of Gaucher's cells in the marrow, and by hepatosplenomegaly and erosion of the cortices of long bones and pelvis. The adult form is associated with moderate anemia and thrombocytopenia, and yellowish pigmentation of the skin; in the infantile form there is, in addition, marked central nervous system impairment; in the juvenile form there are rapidly progressive systemic manifestations but moderate central nervous system involvement.

**genetic disease** a general term for any disorder caused by a genetic mechanism, comprising chromosome aberrations (or anomalies), mendelian (or monogenic or single-gene) disorders, and multifactorial disorders.

**gestational trophoblastic disease** see under [neoplasia](#).

**Gilbert disease** a familial, benign elevation of bilirubin levels without evidence of liver damage or hematologic abnormalities.

**Gilles de la Tourette's disease** see under [syndrome](#).

**Glanzmann disease** see [thrombasthenia](#).

**glycogen storage disease** any of a number of rare inborn errors of metabolism caused by defects in specific enzymes or transporters involved in the metabolism of glycogen.

**type I [glucose-6-phosphatase](#)** deficiency: a severe hepatorenal form due to deficiency of the hepatic enzyme [glucose-6-phosphatase](#), resulting in liver and kidney involvement, with hepatomegaly, hypoglycemia, hyperuricemia, and gout

**type IA [glycogen storage d.](#), [type I](#).**

**type IB** a form resembling [type I](#) but additionally predisposing to infection due to neutropenia and to chronic inflammatory bowel disease; due to a defect in the transport system for glucose 6-phosphate.

**type II** a disorder due to deficiency of the lysosomal enzyme  $\alpha$ -1,4,-glucosidase, the severe infant form resulting in generalized glycogen accumulation, with cardiomegaly, cardiorespiratory failure, and death, and a milder adult form being a gradual skeletal myopathy that sometimes causes respiratory problems.

**type III** a form due to deficiency of [debrancher enzyme \(amylo-1,6-glucosidase\)](#) in muscle, liver, or both; defects in the liver enzyme are characterized by hepatomegaly and hypoglycemia while defects in the muscle enzyme are characterized by progressive muscle wasting and weakness.

**type IV [brancher enzyme](#)** deficiency; cirrhosis of the liver, hepatosplenomegaly, progressive hepatic failure, and death due to deficiency of the [glycogen brancher enzyme](#) (1,4- $\alpha$ -glucan branching enzyme).

**type V** muscle cramps and fatigue during exercise due to a defect in the skeletal muscle isozyme of glycogen [phosphorylase \(muscle phosphorylase\)](#).

**type VI** hepatomegaly, mild to moderate hypoglycemia and mild ketosis, due to deficiency of the liver isozyme of glycogen [phosphorylase \(hepatic phosphorylase\)](#).

**type VII** muscle weakness and cramping after exercise due to deficiency of the muscle isozyme of [6-phosphofructokinase](#).

**type VIII [phosphorylase](#).**

**graft-versus-host (GVH) disease** disease caused by the immune response of histoincompatible, immunocompetent donor cells against the tissue of immunocompromised host, as a complication of bone marrow transplantation, or as a result of maternal-fetal blood transfusion, or therapeutic transfusion to an immunocompromised recipient.

**Graves' disease** an association of hyperthyroidism, goiter, and exophthalmos, with accelerated pulse rate, profuse sweating, nervous symptoms, psychic disturbances, emaciation, and elevated basal metabolism.

**Greenfield's disease** former name for the late infantile form of metachromatic leukodystrophy.

**Gull's disease** atrophy of the thyroid gland with myxedema.

**Günther disease** [congenital erythropoietic porphyria](#).

**H disease** [Hartnup d.](#)

**Hailey-Hailey disease** [benign familial pemphigus](#).

**Hallervorden-Spatz disease** an autosomal recessive disorder caused by decreased numbers of myelin sheaths of the globus pallidus and substantia nigra, with accumulation of iron pigment, progressive rigidity beginning in the legs, choreoathetoid movements, dysarthria, and mental deterioration.

**Hand's disease** [Hand-Schüller-Christian d.](#)

**hand-foot-and-mouth disease** a mild, highly infectious viral disease of children, with vesicular lesions in the mouth and on the hands and feet.

**Hand-Schüller-Christian disease** a chronic, progressive form of multifocal [Langerhans cell histiocytosis](#), sometimes with accumulation of cholesterol, characterized by the triad of calvarial bone defects, exophthalmos, and diabetes insipidus.

**Hansen's disease** [leprosy](#).

**Hartnup disease** a hereditary disorder of intestinal and renal transport of neutral  $\alpha$ -amino acids, marked by a pellagra-like skin rash, with transient cerebellar ataxia, constant renal aminoaciduria, and other biochemical abnormalities.

**Hashimoto's disease** a progressive disease of the thyroid gland with degeneration of its epithelial elements and replacement by lymphoid and fibrous tissue.

**heavy chain diseases** a group of malignant neoplasms of lymphoplasmacytic cells marked by the presence of immunoglobulin heavy chains or heavy chain fragments; they are classified according to heavy chain type, e.g., alpha chain disease.

**Heine-Medin disease** the major form of poliomyelitis.

**hemoglobin disease** any of various hereditary molecular diseases characterized by abnormal hemoglobins in the red blood cells; the homozygous form is manifested by hemolytic anemia.

**hemolytic disease of the newborn** [erythroblastosis fetalis](#).

**hemorrhagic disease of the newborn** a self-limited hemorrhagic disorder of the first few days of life, due to deficiency of vitamin K–dependent coagulation factors II, VII, IX, and X.

**Hers' disease** [glycogen storage d., type VI](#).

**Heubner-Herter disease** the infantile form of celiac disease.

**hip-joint disease** tuberculosis of the hip joint.

**Hippel's disease** [von Hippel's d.](#)

**Hirschsprung's disease** [congenital megacolon](#).

**His disease** , **His-Werner disease** [trench fever](#).

**Hodgkin's disease** a form of malignant lymphoma marked clinically by painless, progressive enlargement of lymph nodes, spleen, and general lymphoid tissue; other symptoms may include anorexia, lassitude, weight loss, fever, pruritus, night sweats, and anemia. Reed-Sternberg cells are characteristically present. Four types have been distinguished on the basis of histopathologic criteria.

**hoof-and-mouth disease** [foot-and-mouth d.](#)

**hookworm disease** infection with the hookworm [Ancylostoma duodenale](#) or [Necator americanus](#), whose larvae enter the body through the skin or in contaminated food or water and migrate to the small intestine where, as adults, they attach to the mucosa and ingest blood; symptoms may include abdominal pain, diarrhea, colic or nausea, and anemia.

**hyaline membrane disease** a type of [respiratory distress syndrome of the newborn](#) in which there is formation of a hyaline-like membrane lining the terminal respiratory passages; extensive atelectasis is attributed to lack of surfactant.

**hydatid disease** an infection, usually of the liver, due to larval forms of tapeworms of the genus [Echinococcus](#), marked by development of expanding cysts.

**hypophosphatemic bone disease** an inherited disorder resembling a mild form of X-linked hypophosphatemia, similarly due to a defect in renal tubular function but usually showing osteomalacia without radiographic evidence of rickets.

**immune complex disease** local or systemic disease caused by the formation of circulating immune complexes and their deposition in tissue, due to activation of complement and to recruitment and activation of leukocytes in type III hypersensitivity reactions.

**infectious disease** one due to organisms ranging in size from viruses to parasitic worms; it may be contagious in origin, result from nosocomial organisms, or be due to endogenous microflora from the nose and throat, skin, or bowel.

**inflammatory bowel disease** any idiopathic inflammatory disease of the bowel, such as Crohn's disease and ulcerative colitis.

**intercurrent disease** one occurring during the course of another disease with which it has no connection.

**iron storage disease** [hemochromatosis](#).

**ischemic bowel disease** [ischemic colitis](#).

**ischemic heart disease** (IHD) any of a group of acute or chronic cardiac disabilities resulting from insufficient supply of oxygenated blood to the heart.

**Janský-Bielschowsky disease** the late infantile form of [neuronal ceroid lipofuscinosis](#), occurring between two and four years of age, characterized by abnormal accumulation of lipofuscin; beginning as myoclonic seizures and progressing to neurologic and retinal deterioration and death by age 8 to 12.

**jumping disease** any of several culture-specific disorders characterized by exaggerated responses to small stimuli, muscle tics including jumping, obedience even to dangerous suggestions, and sometimes coprolalia or echolalia.

**juvenile Paget disease** [hyperostosis corticalis deformans juvenilis](#).

**Kashin-Bek (Kaschin-Beck) disease** a disabling degenerative disease of the peripheral joints and spine, endemic in northeastern Asia; believed to be caused by ingestion of cereal grains infected with the fungus *Fusarium sporotrichiella*.

**Katayama disease** [schistosomiasis japonica](#).

**Kawasaki disease** a febrile illness usually affecting infants and young children, with conjunctival injection, changes to the oropharyngeal mucosa, changes to the peripheral extremities including edema, erythema, and desquamation, a primarily truncal polymorphous exanthem, and cervical lymphadenopathy. It is often associated with vasculitis of the large coronary vessels.

**Kienböck's disease** slowly progressive osteochondrosis of the lunate bone; it may affect other wrist bones.

**kinky hair disease** [Menkes' syndrome](#).

**Köhler's bone disease**

1. osteochondrosis of the tarsal navicular bone in children.
2. thickening of the shaft of the second metatarsal bone and changes about its articular head, with pain in the second metatarsophalangeal joint on walking or standing.

**Krabbe's disease** a lysosomal storage disease beginning in infancy, due to deficiency of  $\beta$ -galactosidase. Pathologically, there is rapidly progressive cerebral demyelination and large globoid bodies (swollen with accumulated cerebroside) in the white substance.

**Kufs' disease** the adult form of [neuronal ceroid lipofuscinosis](#), with onset prior to age 40; characterized by progressive neurologic deterioration but not blindness, excessive storage of lipofuscin, and shortened life expectancy;

**Kümmell's disease** compression fracture of vertebra, with symptoms a few weeks after injury, including spinal pain, intercostal neuralgia, lower limb motor disturbances, and kyphosis.

**Kyasanur Forest disease** a fatal viral disease of monkeys in the Kyasanur Forest of India, communicable to humans, in whom it produces hemorrhagic symptoms.

**Kyrle's disease** a chronic disorder of keratinization marked by keratotic plugs that develop in hair follicles and eccrine ducts, penetrating the epidermis and extending down into the corium, causing foreign-body reaction and pain.

**Lafora's disease** see under [epilepsy](#).

#### **Leber's disease**

1. [Leber's hereditary optic neuropathy](#).
2. [Leber's congenital amaurosis](#).

**legionnaires' disease** an often fatal bacterial infection caused by *Legionella pneumophila*, not spread by person-to-person contact, characterized by high fever, gastrointestinal pain, headache, and pneumonia; there may also be involvement of the kidneys, liver, and nervous system.

**Leiner's disease** a disorder of infancy characterized by generalized seborrhea-like dermatitis and erythroderma, severe intractable diarrhea, recurrent infections, and failure to thrive.

**Leriche disease** [post-traumatic osteoporosis](#).

**Letterer-Siwe disease** a [Langerhans cell histiocytosis](#) of early childhood, of autosomal recessive inheritance, characterized by cutaneous lesions resembling seborrheic dermatitis, hemorrhagic tendency, hepatosplenomegaly, lymphadenitis, and progressive anemia. If untreated it is rapidly fatal. Called also [acute disseminated Langerhans cell histiocytosis](#).

**Libman-Sacks disease** see under [endocarditis](#).

**Lindau's disease** , **Lindau-von Hippel disease** [von Hippel-Lindau d.](#)

**Little's disease** congenital spastic stiffness of the limbs, a form of cerebral palsy due to lack of development of the pyramidal tracts.

**Lobstein's disease** see [osteogenesis imperfecta](#).

**Lou Gehrig disease** [amyotrophic lateral sclerosis](#).

**Lowe disease** [oculocerebrorenal syndrome](#).

**Lutz-Splendore-Almeida disease** [paracoccidioidomycosis](#).

**Lyme disease** a recurrent multisystemic disorder caused by the spirochete *Borrelia burgdorferi*, the vectors being the ticks *Ixodes scapularis* and *I. pacificus*; usually initially characterized by lesions of erythema chronicum migrans, followed by various manifestations including arthritis of the large joints, myalgia, and neurologic and cardiac abnormalities.

**lysosomal storage disease** an inborn error of metabolism with (1) a defect in a specific lysosomal enzyme; (2) intracellular accumulation of an unmetabolized substrate; (3) clinical progression affecting multiple tissues or organs; (4) considerable phenotypic variation within a disease.

**MAC disease** [complex d.](#)

**McArdle disease** [glycogen storage d., type V](#).

**mad cow disease** [bovine spongiform encephalopathy](#).

#### **Madelung's disease**

1. see under [deformity](#).
2. see under [neck](#).

**maple bark disease** [hypersensitivity pneumonitis](#) in logging and sawmill workers due to inhalation of spores of a mold, *Cryptostroma corticale*, growing under the maple bark.

**maple syrup urine disease** (MSUD) a hereditary enzyme defect in metabolism of branched chain amino acids, marked clinically by mental and physical retardation, severe ketoacidosis, feeding difficulties, and a characteristic maple syrup odor in the urine and on the body.

**Marburg virus disease** a severe, often fatal, viral hemorrhagic fever first reported in Marburg, Germany, among laboratory workers exposed to African green monkeys.

**Marchiafava-Micheli disease** [paroxysmal nocturnal hemoglobinuria](#).

**Marie-Bamberger disease** [hypertrophic pulmonary osteoarthropathy](#).

**Marie-Strümpell disease** [ankylosing spondylitis](#).

**Marie-Tooth disease** [Charcot-Marie-Tooth d.](#)

**Mediterranean disease** [thalassemia major](#).

**medullary cystic disease** [familial juvenile nephronophthisis](#).

**Meniere's disease** deafness, tinnitus, and dizziness, in association with nonsuppurative disease of the labyrinth.  
**mental disease** see under [disorder](#).

**Merzbacher-Pelizaeus disease** [Pelizaeus-Merzbacher d.](#)

**metabolic disease** one caused by a disruption of a normal metabolic pathway because of a genetically determined enzyme defect.

**Meyer's disease** adenoid vegetations of the pharynx.

**Mikulicz's disease** benign, self-limited lymphocytic infiltration and enlargement of the lacrimal and salivary glands of uncertain etiology.

**Milroy disease** hereditary permanent lymphedema of the legs due to lymphatic obstruction.

**Minamata disease** a severe neurologic disorder due to alkyl mercury poisoning, with permanent neurologic and mental disabilities or death; once prevalent among those eating contaminated seafood from Minamata Bay, Japan.

**minimal change disease** subtle alterations in kidney function demonstrable by clinical albuminuria and the presence of lipid droplets in cells of the proximal tubules, seen primarily in young children.

**mixed connective tissue disease** a combination of scleroderma, myositis, systemic lupus erythematosus, and rheumatoid arthritis, and marked serologically by the presence of antibody against extractable nuclear antigen.

**Möbius disease** [ophthalmoplegic migraine](#).

**molecular disease** any disease in which the pathogenesis can be traced to a single molecule, usually a protein, which is either abnormal in structure or present in reduced amounts.

**Mondor's disease** phlebitis affecting the large subcutaneous veins normally crossing the lateral chest wall and breast from the epigastric or hypochondriac region to the axilla.

**Monge's disease** [chronic mountain sickness](#).

**Morquio's disease** , **Morquio-Ullrich disease** see under [syndrome](#).

**motor neuron disease** , **motor system disease** any disease of a motor neuron, including spinal muscular atrophy, progressive bulbar paralysis, amyotrophic lateral sclerosis, and lateral sclerosis.

**Mycobacterium avium complex disease** [MAC disease](#); systemic disease caused by infection with organisms of the [Mycobacterium avium-intracellulare](#) complex in patients with human immunodeficiency virus infection.

**Newcastle disease** a viral disease of birds, including domestic fowl, transmissible to humans, characterized by respiratory, gastrointestinal or pulmonary, and encephalitic symptoms.

**new variant Creutzfeldt-Jakob disease** (nvCJD) a variant of [Creutzfeldt-Jakob disease](#) having a younger age of onset than is seen in [Creutzfeldt-Jakob disease](#), and caused by the same agent that causes [bovine spongiform encephalopathy](#).

**Nicolas-Favre disease** [lymphogranuloma venereum](#).

**Niemann's disease** , **Niemann-Pick disease** a lysosomal storage disease due to sphingomyelin accumulation in the reticuloendothelial system; there are five types distinguished by age of onset, amount of central nervous system involvement, and degree of enzyme deficiency.

**nil disease** [minimal change d.](#)

**Norrie's disease** an X-linked disorder consisting of bilateral blindness from retinal malformation, mental retardation, and deafness.

**notifiable disease** one required to be reported to federal, state, or local health officials when diagnosed, because of infectiousness, severity, or frequency of occurrence.

**oasthouse urine disease** [methionine malabsorption syndrome](#).

**obstructive small airways disease** chronic bronchitis with irreversible narrowing of the bronchioles and small bronchi with hypoxia and often hypercapnia.

**occupational disease** disease due to various factors involved in one's employment.

**Oguchi's disease** a form of hereditary night blindness and fundus discoloration following light adaptation.

**organic disease** one associated with demonstrable change in a bodily organ or tissue.

**Osgood-Schlatter disease** osteochondrosis of the tuberosity of the tibia.

**Osler's disease**

1. [polycythemia vera](#).
2. [hereditary hemorrhagic telangiectasia](#).

**Owren's disease** [parahemophilia](#).

**Paget's disease**

1. (of bone) [osteitis deformans](#).
2. (of breast) an intraductal inflammatory carcinoma of the breast, involving the areola and nipple.
3. an extramammary counterpart of Paget's disease (2), usually involving the vulva, and sometimes other sites, as the perianal and axillary regions.

**Parkinson's disease** a slowly progressive form of [parkinsonism](#), usually seen late in life, marked by masklike facies, tremor of resting muscles, slowing of voluntary movements, festinating gait, peculiar posture, muscular weakness, and sometimes excessive sweating and feelings of heat.

**Parrot's disease** see under [pseudoparalysis](#).

**parrot disease** [psittacosis](#).

**Parry's disease** [Graves' d.](#)

**Pelizaeus-Merzbacher disease** a progressive familial form of leukoencephalopathy, marked by nystagmus, ataxia, tremor, parkinsonian facies, dysarthria, and mental deterioration.

**Pellegrini's disease** , **Pellegrini-Stieda disease** calcification of the medial collateral ligament of the knee due to trauma.

**pelvic inflammatory disease** (PID) any pelvic infection involving the upper female genital tract beyond the cervix.

**periodontal disease** any disease or disorder of the periodontium.

**Perthes' disease** osteochondrosis of capitular femoral epiphysis.

**Peyronie's disease** induration of the [corpora cavernosa](#) of the penis, producing a painful fibrous [chordee](#) and penile curvature.

**Pfeiffer's disease** [infectious mononucleosis](#).

#### **Pick's disease**

1. progressive atrophy of the cerebral convolutions in a limited area (lobe) of the brain, with clinical manifestations and course similar to Alzheimer's disease.

2. [Niemann-Pick d.](#)

**polycystic kidney disease** , **polycystic disease of kidneys** either of two unrelated heritable disorders marked by cysts in both kidneys: the *autosomal dominant* or *adult* form is more common, appears in adult life, and is marked by loss of renal function that can be either rapid or slow; the *autosomal recessive* or *infantile* form is more rare, may be congenital or may appear later in childhood, and almost always progresses to renal failure.

**polycystic renal disease** [polycystic kidney d.](#)

**Pompe's disease** [glycogen storage d., type II](#).

**Pott's disease** [spinal tuberculosis](#).

**primary electrical disease** serious ventricular tachycardia, and sometimes ventricular fibrillation, in the absence of recognizable structural heart disease.

**prion disease** any of a group of fatal, transmissible neurodegenerative diseases, which may be sporadic, familial, or acquired, caused by abnormalities of [prion protein](#) metabolism resulting from mutations in the [prion protein](#) gene or from infection with pathogenic forms of the protein.

**pulseless disease** [Takayasu's arteritis](#).

**Raynaud's disease** a primary or idiopathic vascular disorder, most often affecting women, marked by bilateral attacks of Raynaud's phenomenon.

#### **Recklinghausen's disease**

1. [neurofibromatosis](#).

2. (of bone) [osteitis fibrosa cystica generalisata](#).

**Refsum's disease** an inherited disorder of lipid metabolism, characterized by accumulation of phytanic acid, chronic polyneuritis, retinitis pigmentosa, cerebellar ataxia, and persistent elevation of protein in cerebrospinal fluid.

**remnant removal disease** [familial dysbetalipoproteinemia](#).

**reversible obstructive airway disease** a condition characterized by [bronchospasm](#) reversible by intervention, as in [asthma](#).

**rheumatic heart disease** the most important manifestation and sequel to rheumatic fever, consisting chiefly of valvular deformities.

**rheumatoid disease** a systemic condition best known by its articular involvement (rheumatoid arthritis) but emphasizing nonarticular changes, e.g., pulmonary interstitial fibrosis, pleural effusion, and lung nodules.

**Ritter's disease** [dermatitis exfoliativa neonatorum](#).

**Roger's disease** a ventricular septal defect; the term is usually restricted to small, asymptomatic defects.

**runt disease** a graft-versus-host disease produced by immunologically competent cells in a foreign host that is unable to reject them, resulting in gross retardation of host development and in death.

**Salla disease** an inherited disorder of sialic acid metabolism characterized by accumulation of sialic acid in lysosomes and excretion in the urine, mental retardation, delayed motor development, and ataxia.

**Sandhoff's disease** a type of GM2 gangliosidosis resembling Tay-Sachs disease, seen in non-Jews, marked by a progressively more rapid course, and due to a defect in hexosaminidase, both isozymes A and B.

**Schamberg's disease** a slowly progressive purpuric and pigmentary disease of the skin affecting chiefly the shins, ankles, and dorsa of the feet.

**Schilder's disease** subacute or chronic leukoencephalopathy in children and adolescents, similar to [adrenoleukodystrophy](#); massive destruction of the white substance of the cerebral hemispheres leads to blindness, deafness, bilateral spasticity, and mental deterioration.

**Schönlein's disease** see under [purpura](#).

#### **secondary disease**

1. one subsequent to or as a consequence of another disease.
2. one due to introduction of incompatible, immunologically competent cells into a host rendered incapable of rejecting them by heavy exposure to ionizing radiation.

**self-limited disease** one that runs a limited and definite course.

**serum disease** see under [sickness](#).

**severe combined immunodeficiency disease** (SCID) see under [immunodeficiency](#).

**sexually transmitted disease** [venereal disease](#); any of a diverse group of infections transmitted by sexual contact; in some this is the only important mode of transmission, and in others transmission by nonsexual means is possible.

**sickle cell disease** any disease associated with the presence of hemoglobin S.

**Simmonds' disease** see [panhypopituitarism](#).

**sixth disease** [exanthema subitum](#).

**small airways disease** chronic obstructive bronchitis with irreversible narrowing of the bronchioles and small bronchi. See also [obstructive small airways d.](#)

**Smith-Strang disease** [methionine malabsorption syndrome](#).

**Spielmeier-Vogt disease** [Vogt-Spielmeier d.](#)

**Steinert's disease** [myotonic dystrophy](#).

**Still's disease** [juvenile rheumatoid arthritis](#).

**storage disease** a metabolic disorder in which a specific substance (a lipid, a protein, etc.) accumulates in certain cells in unusually large amounts.

**storage pool disease** a blood coagulation disorder due to failure of the platelets to release adenosine diphosphate (ADP) in response to aggregating agents; characterized by mild bleeding episodes, prolonged bleeding time, and reduced aggregation response to collagen or thrombin.

#### **Strümpell's disease**

1. hereditary lateral sclerosis with the spasticity mainly limited to the legs.
2. [cerebral poliomyelitis](#).

**Strümpell-Leichtenstern disease** [hemorrhagic encephalitis](#).

**Strümpell-Marie disease** [ankylosing spondylitis](#).

**Sutton's disease**

1. [halo nevus](#).
2. [periadenitis mucosa necrotica recurrens](#).
3. [granuloma fissuratum](#).

**Swift's disease** , **Swift-Feer disease** [acrodynia](#).

**Takayasu's disease** see under [arteritis](#) .

**Tangier disease** a familial disorder characterized by a deficiency of high-density lipoproteins in the blood serum, with storage of cholesteryl esters in tissues.

**Tarui's disease** [glycogen storage d., type VII](#).

**Tay-Sachs disease** (TSD) the most common GM2 gangliosidosis, seen almost exclusively in northeastern European Jews, characterized by infantile onset, doll-like facies, cherry-red macular spot, early blindness, hyperacusis, macrocephaly, seizures, hypotonia, and death in early childhood.

**Thomsen's disease** [myotonia congenita](#).

**thyrotoxic heart disease** heart disease associated with hyperthyroidism, marked by atrial fibrillation, cardiac enlargement, and congestive heart failure.

**transmissible neurodegenerative disease** [prion d.](#)

**trophoblastic disease** [gestational trophoblastic neoplasia](#).

**tsutsugamushi disease** [scrub typhus](#).

**tunnel disease** [decompression sickness](#).

**uremic bone disease** [renal osteodystrophy](#).

**venereal disease** [sexually transmitted d.](#)

**venoocclusive disease of the liver** symptomatic occlusion of the small hepatic venules caused by ingestion of Senecio tea or related substances, by certain chemotherapy agents, or by radiation.

**vinyl chloride disease** acro-osteolysis resulting from exposure to vinyl chloride, characterized by Raynaud's phenomenon and skin and bony changes on the limbs.

**Vogt-Spielmeyer disease** the juvenile form of [neuronal ceroid lipofuscinosis](#) with onset between ages 5 and 10 years; characterized by rapid cerebretinal degeneration, excessive neuronal storage of lipofuscin, and death within 10 to 15 years.

**Volkman's disease** congenital deformity of the foot due to tibiotarsal dislocation.

**von Hippel's disease** hemangiomas confined principally to the retina; when associated with hemangioblastoma of the cerebellum, it is known as [von Hippel-Lindau d.](#)

**von Hippel-Lindau disease** a hereditary condition marked by hemangiomas of the retina and hemangioblastomas of the cerebellum, sometimes with similar lesions of the spinal cord and cysts of the viscera; there may be neurologic symptoms such as seizures and mental retardation.

**von Willebrand's disease** an autosomal dominant bleeding disorder characterized by prolonged bleeding time, deficiency of [von Willebrand's factor](#), and often impairment of adhesion of platelets on glass beads, associated with epistaxis and increased bleeding after trauma or surgery, menorrhagia, and postpartum bleeding.

**Waldenström's disease** osteochondrosis of the capitular femoral epiphysis.

**Weber-Christian disease** [nodular nonsuppurative panniculitis](#).

**Werlhof's disease** [idiopathic thrombocytopenic purpura](#).

**Wernicke's disease** see under [encephalopathy](#).

**Westphal-Strümpell disease** [hepatolenticular degeneration](#).

**Whipple's disease** a malabsorption syndrome marked by diarrhea, steatorrhea, skin pigmentation, arthralgia and arthritis, lymphadenopathy, central nervous system lesions, and infiltration of the intestinal mucosa with macrophages containing PAS-positive material.

**Whitmore's disease** [melioidosis](#).

**Wilson's disease** an inherited, progressive disorder of copper metabolism, with accumulation of copper in liver, brain, kidney, cornea, and other tissues; it is characterized by cirrhosis in the liver, degenerative changes in the brain, and a pigmented ring at the outer margin of the cornea.

**Wolman's disease** a lysosomal storage disease due to deficiency of the lysosomal sterol esterase, occurring in infants, and associated with hepatosplenomegaly, adrenal steatorrhea, calcification, abdominal distention, anemia, and inanition.

**woolorter's disease** [inhalational anthrax](#).