



Study to Explore Early Development

GLOSSARY OF MEDICAL TERMINOLOGY

Following are definitions and descriptions of each of the conditions listed in the Maternal and Paternal Medical History Forms and in the Autoimmune Survey. If you are still not sure about the meaning of a condition, please mark it and we can go over it with you during a clinic visit.

Addison’s disease	A rare condition that occurs when the adrenal glands are not able to produce enough of the hormones cortisol and aldosterone.
Allergies	The immune system overreacts to certain substances causing inflammation (swelling) or irritation of the eyes, nose, throat, lungs, skin, or digestive system. Examples include hayfever and allergy to mold, dander, pollen, dust, drugs, or food.
Ankylosing spondylitis	A form of arthritis that causes inflammation (swelling) of the joints between the vertebrae (bones) of your spine and the joints between your spine and pelvis (at the hip). The disorder gets worse over time and often causes pain and stiffness in the back, hips, and shoulders. More severe forms of the disease cause fatigue and weight loss and can affect the eyes and heart.
Anxiety disorder	A psychiatric disorder in which a person has feelings of anxiety, fear, or worry. Types of anxiety disorders include panic attacks, separation anxiety, obsessive-compulsive disorder, and posttraumatic stress disorder.
Aplastic anemia	Low levels of red blood cells caused when the bone marrow is unable to produce enough blood cells. Early symptoms include weakness, fatigue, infections that won’t go away or occur again and again, and/or easy bruising and bleeding.
Asperger’s syndrome	One of the autistic spectrum disorders. A pervasive developmental disorder in which a person has a hard time communicating or relating socially with others. Other symptoms may include lack of coordination, unusual interests, repetitive routines or rituals, and different ways of speaking or using language. See autism spectrum disorders.
Asthma	A condition that causes wheezing or difficulty breathing. Treatment may involve using steroid inhalers or tablets. Symptoms may get worse with exposure to pollen, mold, or tobacco smoke.

Attention-deficit/hyperactivity disorder (ADHD/ADD)	A behavior disorder in which a person has great trouble paying attention and is hyperactive or impulsive to a point that causes problems for the individual in school, work, or social situations.
Autism	See autism spectrum disorders.
Autism spectrum disorders	A neurodevelopmental disorder that causes a person to have problems interacting socially with other people; trouble talking or communicating with others; and behavioral development that requires routines, schedules, and predictable conditions. Autism spectrum disorders include autistic disorder, pervasive developmental disorder – not otherwise specified, and Asperger’s syndrome.
Autoimmune hepatitis	A disease in which the body's immune system attacks liver cells causing the liver to become inflamed (swollen, irritated). Symptoms often include excess hair growth, acne, arthritis, and an enlarged liver.
Bipolar disease	A psychiatric disorder that causes mood swings from mania (exaggerated feeling of well-being, energy, and confidence) to depression (sadness, lack of self-esteem).
Birth defect	A malformation or defect that is present when a child is born, usually recognized before or at birth. Examples of birth defects include limb defects, cleft lip and cleft palate, and heart defects.
Bleeding/clotting disorders	Bleeding disorders: A group of conditions of the blood clotting (coagulation) system in which a person bleeds a lot and for a long time after an injury. Clotting disorders: A group of conditions of the blood clotting (coagulation) system in which potentially dangerous blood clots can form.
Cancer	A disease that develops when cells in a part of the body begin to grow out of control. Types of cancer include breast, bone, bladder, brain, leukemia, lymphoma.
Cardiovascular condition	A problem affecting the heart, blood vessels, or circulation of the blood.
Celiac disease (sprue)	Digestive condition in which eating the protein gluten (found in wheat, barley, or rye) causes damage to small intestines.
Cerebral palsy	A disorder that affects a person's ability to move and to maintain balance and posture, usually diagnosed in the first year of life.
Crohn’s disease	Chronic form of inflammatory bowel disease; causes severe irritation in the gastrointestinal tract; usually affects the lower intestine and/or colon.
Childhood disintegrative disorder	In children, a loss of skills after a period of 2 years of normal development; significant loss of skills in at least two of the following areas: use or understanding of language, social skills, bowel or bladder control, or motor skills.
Cystic fibrosis	A chronic disease that causes mucus to become thick and build up, which clogs the passages to organs, mainly the lungs and the pancreas. Blocked pathways to the lungs can cause breathing

	problems. The disease gets worse over time.
Depression	Severe, depressed mood that interferes with normal living. Symptoms include extended sadness, lack of self-esteem, loss of interest or pleasure in normal activities, decreased energy, changes in sleep and appetite, and feelings of guilt or hopelessness that last for at least 2 weeks.
Dermatitis herpetiformis (celiac disease of the skin)	Chronic dermatitis (swelling of the skin) that causes itchy, raised areas of the skin and swollen or infected areas on the skin that look like clusters of hives.
Diabetes	A problem with controlling insulin levels and processing sugars that results in high or low blood sugar. The three main types of diabetes are Type 1, also known as insulin-dependent diabetes mellitus or juvenile (childhood) diabetes; Type 2, also known as adult onset, non-insulin dependent diabetes; and gestational diabetes which usually occurs only during pregnancy.
Down syndrome	Down syndrome is a genetic condition that causes delays in physical and intellectual development.
Eating disorder	A disorder involving unhealthy weight control practices that may include extremely low food or calorie intake (anorexia) or vomiting (bulimia) to lose weight.
Eczema/psoriasis	Chronic skin conditions. Eczema causes the skin to become dry, itchy, or scaly. Psoriasis causes pink or silvery-white scaly patches, often over the scalp and knee and elbow joints.
Endocrine disorder (hormonal disorder)	Problems in organs or glands that make hormones, which cause problems with reproduction, metabolism (food burning and waste elimination), and growth.
Fragile X syndrome	A hereditary condition that causes a wide range of mental impairment, from mild learning disabilities to severe mental retardation.
Gastrointestinal disorder	Condition related to the gastrointestinal tract. Conditions might be related to the small and large intestines, rectum, and stool patterns.
Giant cell arteritis (temporal arteritis and cranial arteritis)	Disorder that results in swelling of arteries in the head (most often the temporal arteries located on each side of the head), neck, and arms.
Graves' disease (diffuse toxic goiter)	Over-activity of the thyroid gland that becomes enlarged into a goiter. Graves' disease is the most common cause of hyperthyroidism.
Guillain-Barré syndrome (Landry's ascending paralysis)	A disorder in which the body's immune system attacks part of the peripheral nervous system. The first symptoms include weakness or tingling sensations in the legs.
Hashimoto's thyroiditis	A type of autoimmune thyroid disease in which the immune system attacks and destroys the thyroid gland. The thyroid helps set the rate of metabolism (how fast the body uses energy).
Hearing impairment	A reduction in the ability to perceive sound; ranges from slight inability to complete deafness.

Hemolytic anemia	Disorder in which the red blood cells are destroyed faster than the bone marrow can produce them. TYPES: intrinsic or extrinsic relating to way the cell is destroyed.
High blood pressure	Occasional or constant rise in systemic arterial blood pressure to a level likely to cause cardiovascular damage or other adverse consequences (usually greater than 140/90).
Hyperthyroidism	Condition in which there is too much thyroid hormone in the body.
Hypothyroidism	Condition in which there is too little thyroid hormone in the body.
Irritable bowel syndrome	Disorder of the large and small intestines that causes abdominal pain or discomfort, constipation, or diarrhea.
Learning disability	A disorder in one or more of the basic cognitive and psychological processes involved in understanding or using written or spoken language.
Lupus, or systemic lupus erythematosus (SLE)	Tissue disease, mostly among women, that causes fever, skin rash, and arthritis, often by anemia, inflamed mucus membrane, and involvement of the heart and kidneys.
Mental retardation	A condition in which a person has both a significantly below-average score on a test of mental ability or intelligence and limitations in the ability to function in areas of daily life, such as communication, self-care, and getting along in social situations and school activities. Mental retardation is sometimes referred to as a cognitive or intellectual disability.
Migraine headaches	Periodic headaches, usually on one side of the head; may occur with nausea, vomiting, increased sensitivity of the eyes to light, and other symptoms.
Mixed connective tissue disease (MCTD)	Syndrome in which a person has a combination of scleroderma, systemic lupus erythematosus, polymyositis, or rheumatoid arthritis. Individuals with MCTD have symptoms of each of these disorders, including painful or swollen joints, heart or lung problems, skin problems, kidney disease, and muscle weakness.
Motor problem/movement or coordination problems	Problems with both fine motor tasks (those that involve skills of the hands and ability to use small objects) and gross motor tasks (such as walking, running, postural control).
Multiple sclerosis	A disorder of the brain and spinal cord (central nervous system) caused by increasing damage to the outer covering of nerve cells (myelin); may result in decreased nerve functioning, fatigue, dizziness, or difficulty walking or using hands.
Myasthenia gravis (muscle fatigue disease)	A disease in which muscles tire easily and become increasingly weak.
Narcolepsy	A sleep disorder that causes a person to fall asleep uncontrollably during the day.
Neurofibromatosis Type 1 (von Recklinghausen's disease)	Rare genetic disorder that causes the development of multiple non-cancerous (benign) tumors of the nerves and skin (neurofibromas) and areas of abnormal skin color, including tan, brown, or white

	spots in unusual places like under the arms and groin.
Neuromuscular disorder	Conditions that involve the weakness or wasting of the body muscles. These problems may arise in the spinal cord, the peripheral nerves, or the muscle fibers. Examples include muscular dystrophy and myasthenia gravis.
Obesity	Excess fat; a body weight that is 30% more than the ideal body weight for a person's height.
Obsessive compulsive disorder	A psychiatric (mental) disorder in which a person gets caught up in certain rituals, either thoughts or ways of doing things, that interfere with normal social interaction with others and cannot be stopped. Examples include constant worrying about germs or concern with the exact order of things.
Optic neuritis (optic nerve inflammation)	Swelling or inflammation of the optic nerve (located in the eye).
Pemphigus	An uncommon blistering disease of the skin and mucous membranes commonly involving the mouth and scalp.
Personality disorders	General term for a group of behavioral diseases characterized by lifelong patterns of thought and action usually lifelong ingrained maladaptive that result in abnormal behavior and interfere with lifestyle and social adjustment.
Pervasive developmental disorder	A group of mental disorders of infancy, childhood, or adolescence characterized by lack of the basic social skills, language skills, imagination, and need for routine.
Pervasive developmental disorder—not otherwise specified	See autism spectrum disorders.
Reading difficulty	Difficulty reading at the level appropriate for a given age.
Recklinghausen's disease	See neurofibromatosis.
Reiter's syndrome (reactive arthritis)	Arthritis of large, weight-bearing joints such as the knee and ankle with involvement of the urethra, eye, and skin; more commonly found among men.
Respiratory condition	Condition related to the lungs, esophagus, or flow of air through the respiratory passageway that may cause difficulty breathing.
Rett's syndrome	A developmental disorder mostly affecting girls after an apparently normal early development. The syndrome is characterized by a slowing of the rate of head growth, severely impaired language and social skills, loss of purposeful hand movements, and a shaky torso.
Rheumatoid arthritis	A relatively common disease in which the tissues lining the joints become inflamed (swollen). Over time, the inflammation may destroy the joint tissues, leading to disability.

Schizophrenia	A chronic mental illness that can cause a person to withdraw from the people and activities in the world around them and retreat into a world of delusions and hallucinations.
Scleroderma	Chronic (lifelong) disease that can cause thickening, hardening, or tightening of the skin, blood vessels, and internal organs.
Self-injuring behavior	Trying to cause bodily harm to oneself. Examples include biting the wrist, banging the head, and cutting the skin.
Seizure disorder/epilepsy	Disorders of the central nervous system that cause sudden seizures, muscle contractions, and partial or total loss of consciousness.
Sickle cell/thalassemia/other hereditary anemias	Inherited, lifelong disease in which the red blood cells, which are normally disc-shaped, become crescent-shaped and do not function normally. Blood clotting occurs frequently. Thalassemias are hereditary disorders characterized by defective production of hemoglobin (protein inside red blood cells that carries oxygen), which leads to low production, and excessive destruction, of red blood cells. This hereditary disorder is characterized by growth failure, bone deformities, and enlarged liver and spleen. Other hereditary anemias may include G-6-PD.
Sjögren’s syndrome	An inflammatory disorder affecting the whole body. Symptoms include dry mouth, decreased tearing, and other dry mucous membranes. It is often associated with autoimmune rheumatic disorders.
Sleep disorder	Difficulty getting to sleep or staying asleep for at least 1 month; associated with daytime fatigue (tired feeling); leads to distress or impairment in social, occupational, or other important areas of functioning.
Speech problems	Problems speaking to communicate with others. Difficulty expressing an understanding of written or spoken language.
Stevens-Johnson syndrome	Skin disorder, of unknown cause, resulting from an allergic reaction to medications, illness, or infections. Symptoms include a rash, skin peeling, or sores on the mucous membranes.
Sydenham’s chorea (St. Vitus’ dance)	Disorder of the central nervous system characterized by emotional instability; purposeless movements; and muscular weakness.
Suicide attempt	Trying to take one’s own life.
Thrombocytopenia (immune, idiopathic)	A condition in which there is an abnormally small number of platelets in the circulating blood.
Tourette’s syndrome	Neurological disorder characterized by tics—involuntary, rapid, sudden movements that occur repeatedly in the same way.
Tuberous sclerosis	Rare genetic, neurological disorder that causes seizures, mental retardation, and skin and eye lesions (damage). Small tumors may grow on the face and eyes and in the brain, kidneys, and other organs.

	Seizures most often start in the first year of life.
Ulcerative colitis	A chronic (lifelong) inflammatory disease of the large intestine and rectum characterized by bouts of bloody diarrhea.
Vision impairment	Impairment in a person's eyesight that cannot be corrected to a “normal” level. It may be caused by a loss of visual acuity, where the eye does not see objects as clearly as usual. It may also be caused by a loss of visual field, where the eye cannot see as wide an area as usual without moving the eyes or turning the head.