

## Myotonic Dystrophy Medical Glossary

**Accommodations.** Supports that are provided to a child throughout the school day that do not significantly alter what is being taught or how the child participates in school activities. Examples include preferential seating, extended time on tests, daily communication logs to share information between school and home, use of spell check or a computer, enlarged print, and books on tape.

**Activities of Daily Living (ADL).** Normal day-to-day activities such as walking, going to the bathroom, eating, dressing, bathing, etc.

**Americans with Disabilities Act (ADA).** The ADA prohibits discrimination against people with disabilities in employment, transportation, public accommodation, communications, and governmental activities.

**Amniocentesis.** A method of prenatal diagnosis at 15 weeks in which a fluid sample is removed from the womb and grown in tissue culture; it takes another 2-3 weeks for results; a fetus can be tested for genetic diseases this way.

**Ankle Foot Orthosis (AFO).** An apparatus used to support, align, prevent, or correct deformities or to improve the function of the ankle and foot.

**Anticipation.** The tendency in certain genetic disorders-like myotonic dystrophy-for individuals in successive generations to present with symptoms at an earlier age and/or with more severe manifestations; often observed in disorders resulting from the expression of a trinucleotide repeat mutation that tends to increase in size and have a more significant effect when passed from one generation to the next.

**Anticholinesterases.** (Example: Neostigmine) One of the drugs that myotonic dystrophy patients should avoid; can adversely affect diameter of blood vessels, function of the intestines, and part of the nervous system that controls smooth muscle, heart muscle and gland cells.

**Apnea.** Periodic absence of breathing while sleeping.

**Armodafinil.** A drug used to treat excessive daytime sleepiness (brand name is Nuvigil).

**Arrhythmia.** An irregular heart beat.

**Aspiration pneumonia.** Serious form of pneumonia resulting from inhalation of foreign material, usually food particles or vomit, into the bronchi.

**Asymptomatic.** Without symptoms, at least in the mind of the person himself.

**Atrial fibrillation.** Abnormal heartbeat in which the normal rhythmical contractions of the upper chambers of the heart (cardiac atria) are replaced by rapid irregular twitchings of the muscular wall.



**Attention Deficit Hyperactivity Disorder.** A behavior disorder originating in childhood in which the essential features are signs of developmentally inappropriate inattention, impulsivity and hyperactivity. Although most individuals have symptoms of both inattention and hyperactivity-impulsivity, one or the other pattern may be predominant. The disorder is more frequent in males than females. Onset is in childhood. Symptoms often attenuate during late adolescence, although a minority experiences the full complement of symptoms into mid-adulthood.

**Autosomal dominant.** A pattern of inheritance in which if one parent has a mutated gene, each offspring has a 50% chance of inheriting it.

**Barium swallow test.** A test in which a patient swallows a thick white substance and the swallowing process is filmed to detect possible abnormalities.

**Blepharoplasty.** Any operation for the correction of a defect in the eyelids.

**Bradyarrhythmia.** Any disturbance of the heart's rhythm resulting in a rate of under 60 beats per minute.

**Bulbar weakness.** Presence of weakness in the tongue, lips, palate, pharynx and larynx.

**Cataracts.** A film that can form in the eye and cause complete or partial opacity of the ocular lens, or blurry vision; in myotonic dystrophy patients, often posterior subcapsular iridescent cataracts form; they are sometimes referred to as Christmas-tree cataracts.

**Cardiac conduction.** The electrical impulses that cause the heart to beat.

**Cardiomyopathy.** Damage to the heart muscle that decreases its ability to pump blood effectively.

**Cardiotoxicity.** Substance that is harmful to the heart.

**CCTG.** The abbreviation for "cytosine, cytosine, thymidine, guanine", the chemicals in the DNA that cause myotonic dystrophy type-2 (on chromosome 3) when they are created in repeats greater than the normal number.

**Chorionic villus sampling (CVS).** A method of prenatal diagnosis at around 10 weeks into the pregnancy; a biopsy in which a piece of membrane around the embryo is removed using a needle through the abdomen or vagina; results in 1-2 weeks.

**Chromosome.** One of the bodies (normally 23 pairs) located in the nucleus of a cell that hosts the genes.

**Cisapride.** (Example: propulsid) A prokinetic drug that should be avoided by individuals with myotonic dystrophy.

**Club foot.** An inversion of the foot in which only the outer side of the sole touches the ground; also called talipes equinovarus.

**CNS.** Abbreviation for central nervous system; brain.

**Cognitive problems.** Difficulties with thinking, learning and memory.



**Cohort study.** Particular form of longitudinal study that samples a cohort, meaning a group of people who share a defining characteristic, typically those who experienced a common event in a selected period.

**Conduction defects.** Problems with the electrical impulses that regulate the heart beat.

**Congenital.** Present at birth.

**Contractures.** Permanent tightening of muscles causing abnormal joint rigidity.

**Copayment.** A fixed amount (\$20 for example) you pay for a covered healthcare service after you've paid your deductible.

**CPAP.** Acronym for continuous positive airway pressure; a device that delivers air to the nose for easier breathing; often used at night for those with sleep apnea.

**Creatine Kinase (CK) levels.** An important enzyme in muscle contraction.

**CTG.** The abbreviation for "cytosine, thymidine, guanine", the 3 chemicals in the DNA that cause myotonic dystrophy type-1 (on chromosome 19) when they sputter beyond the normal 5-37 repeats found along the rung-looking parts in the double helix that resembles a gracefully twisted ladder. CTG repeat causes DM1.

**Deductible.** The amount you pay for covered healthcare services before your insurance plan starts to pay. With a \$2,000 deductible for example, you pay the first \$2,000 of covered services yourself. Once you exceed \$2,000, your insurance company will pay for additional medical costs.

**Degenerative.** Deteriorating, getting worse.

**Depolarizing neuromuscular blocking agents.** (Example: suxamethonium) Drugs causing skeletal muscle paralysis.

**Distal.** Situated away from the trunk of the body, at the end of the limbs toward the feet and hands.

**DM1.** The abbreviation for the Latin name for myotonic dystrophy (dystrophia myotonica) type 1, the more severe form of myotonic dystrophy with the mutation found on chromosome 19.

**DM2.** The abbreviation for the Latin name for myotonic dystrophy (dystrophia myotonica) type 2, with the mutation found on chromosome 3.

**DMPK.** The abbreviation for the myotonic dystrophy gene, the Myotonic Dystrophy Protein Kinase gene that causes DM1; it is located on chromosome 19.

**Double helix.** Two strands of DNA held together by hydrogen bonds; when enlarged it resembles a tiny ladder (with many rungs) that has been gracefully twisted; it is along these rungs that the chemical repeats stutter beyond their normal number and cause the mutation, or change, in the gene that causes myotonic dystrophy.

**Dominant inheritance.** As in myotonic dystrophy, the expression of a gene where if one parent carries the mutated gene, the children have a 50% chance of getting it.

**Dysphagia.** Difficulty swallowing.



**Dysphasia.** Difficulty speaking.

**Dyspnea.** Shortness of breath.

**Dystrophy.** An inherited muscle disorder in which the muscles become weaker.

**Ectopic.** Occurring in the wrong place in the body, such as the development of an impregnated egg outside the cavity of the uterus; or, a cardiac beat originating elsewhere other than at the sinoatrial node

**EDS.** Acronym for excessive daytime sleepiness

**EKG or ECG.** Electrocardiogram, a test that prints out a graphic record of a person's heart beat

**Endocrine system.** The body system that secretes hormones that allow the body perform many of its functions

**Epidemiology.** The study of the distribution of health-related states (for a specific disease, like myotonic dystrophy) or events in specified populations

**Esophagus.** The portion of the digestive canal between the pharynx and stomach

**Expansion.** On FAQ's page, referring to enlargement of the myotonic dystrophy genetic mutation, or abnormality, as it passes to offspring; also refers to the enlargement of mutations within a given organ or system over the life of an affected individual (see somatic mosaicism); happens often in myotonic dystrophy.

**Foot drop.** Partial or total inability to dorsiflex (turn upward) the foot.

**Formulary.** A list of prescription drugs covered by a prescription drug plan or another insurance plan offering prescription drug benefits. Also called a drug list.

**Gait.** Manner of walking.

**Gastroenterologist.** A doctor focusing on the function and disorders of the stomach, intestines and assorted organs that are often referred to as the GI tract.

**Gene.** A functional unit of heredity (like eye and hair color, height, among many other characteristics including inherited diseases) that occupies a specific place on a chromosome; it is capable of reproducing itself at each cell division and directs the formation of an enzyme or protein.

**Genetic.** Of or pertaining to genes; inherited.

**Genetic counseling.** A meeting with a medical professional, often a geneticist, to learn how a possible inherited disease can affect you, and how you can choose to avoid passing it to your offspring if you so choose.

**Genomic background.** Referring to the complete set of genes inherited from one's parents.

**Genotypes.** The sum total of the genetic material transmitted from a person's parents.

**GI tract.** Bodily system referring to the stomach, intestines and related organs.

**Gonadal (or testicular) atrophy in men.** A medical condition in which the male reproductive organs (the testes) diminish in size and fail to function.



**G-Tube.** Implanted feeding tube to supply sustenance when person is unable to safely swallow on his own impotence.

**Haplotype analysis.** Molecular genetic testing to identify a set of closely linked segments of DNA.

**Health Maintenance Organization (HMO).** A type of health insurance plan that usually limits coverage to care from doctors who work for or contract with the HMO. It generally won't cover out-of-network care except in an emergency.

**Heterotropia.** Inability of one eye to attain binocular vision with the other because of imbalance of the muscles of the eyeball--called also strabismus, squint.

**Hydramnios.** Excessive amniotic fluid build-up during pregnancy.

**Hyperkalemia.** A greater than normal concentration of potassium ions in the circulating blood.

**Hyperostosis.** Excessive growth of bony tissue.

**Hypersomnia.** Excessive daytime sleepiness.

**Hypertrophy.** General increase in bulk or a part of an organ.

**Hypothermia.** A body temperature significantly below 98.6.

**Hypotonia.** Low muscle tone causing floppiness, as in a child with the congenital form of myotonic dystrophy.

**Implanted Cardioverter Defibrillator (ICD).** A cardiac device implanted in the chest; a combination pace-maker and defibrillator designed to regulate the heart beat, to keep it from beating too fast or too slow.

**Impulse inhibition.** The inability to control one's impulses.

**Incidence.** New disease diagnoses in a time period.

**In vitro fertilization.** Eggs are obtained from the female after drugs have been used to stimulate ovarian production. While under sedation and with the use of ultrasound guidance, a needle is inserted into the ovaries and eggs are aspirated. These eggs are then fertilized in the laboratory (in-vitro) with the partner's sperm and the developing embryos are cultured from three to six days.

**Incentive spirometry.** A breathing device to help exercise breathing muscles and help maximize lung capacity.

**Individualized Education Program (IEP).** A written plan for each student in special education describing the student's present levels of performance, annual goals including short-term objectives, specific special education and related services, dates for beginning and duration of services, and how the IEP will be evaluated.

**Induction drugs.** Drugs used to "knock a patient out" prior to surgery and certain other procedures.

**Insulin resistance.** Diminished effectiveness of insulin in lowering blood sugar levels.

**Intercostal muscles.** Muscles between the ribs.

**Intubation.** The insertion of a tube into the lungs to provide pulmonary ventilation, or to assist with breathing.



**Late onset.** Diagnosis later in life, typically after age 50.

**Mexiletine.** A drug used to treat myotonia (delayed muscle relaxation after contraction) in muscle diseases such as myotonic dystrophy and myotonia congenital (brand name is Mexitil).

**Modafinil.** A drug is used to treat excessive daytime sleepiness (brand name is Provigil).

**Motility.** The power of spontaneous movement.

**Multisystemic disorder.** A disease that can affect many different organs and systems in the body.

**Mutation.** As used on this website, a change in the normal chemistry of a gene.

**Myotonia.** The inability of contracted muscles to relax on command; or, a special kind of muscle stiffness.

**Myopathy.** Muscle weakness.

**Newborn screening.** Public health program of screening in infants shortly after birth for conditions that are not clinically evident in the newborn period.

**NICU.** Neonatal (new born) intensive care unit.

**Opiates.** Any preparation or derivative of opium.

**Oro.** Referring to the mouth.

**Oropharyngeal muscle weakness.** Reduced strength in the upper expanded portion of the digestive tube, between the esophagus below the mouth and nasal cavities above and in front.

**PCR - polymerase chain reaction.** A procedure that produces millions of copies of a short segment of DNA; the amplified product, doubled each cycle for 30 more cycles, can then be subjected to further testing; it is a common procedure in molecular genetic testing in order to generate enough DNA to perform the test; in individuals suspected of having myotonic dystrophy, it can be used to determine the number of trinucleotide repeats in the DMPK gene on the 19th chromosome.

**PGD.** Abbreviation for preimplantation genetic diagnosis achieved through in vitro fertilization where analysis of embryos is done prior to being implanted by a doctor into the uterus of a woman.

**Pacemaker.** An implanted heart device to correct a very slow or irregular heart beat.

**Pathogenesis.** Biological mechanism that leads to a diseased state

**Perioperative.** Around the time of surgery.

**Pharynx.** The upper expanded portion of the digestive tube, between the esophagus below the mouth and nasal cavities above and in front.

**Phenotype.** The observable signs, symptoms and other aspects of a person's outward appearance and behavior.

**PICU.** Pediatric intensive care unit.

**Pilomatrixoma.** Benign skin tumors under the skin; associated with hair follicles.

**Placenta.** Organ formed inside the lining of the womb that provides nourishment for fetus and elimination of its waste products.



**Placenta accreta.** Condition in pregnancy in which the placenta (see definition) has an abnormally deep attachment through the endometrium and into the myometrium (the middle layer of the uterine wall), causing full or partial placental retention. Condition typically requires surgery to prevent abnormal post-partum bleeding and fully remove the placenta. In severe cases can lead to a hysterectomy or can be fatal.

**Placenta previa.** Condition in pregnancy in which the placenta (see definition) is implanted in lower segment of womb close to the internal opening of the cervix, or sometimes completely covering that internal opening.

**Polyhydramnios.** Excessive amniotic fluid build-up during pregnancy.

**Postoperative apnea.** Absence of breathing after surgery.

**Postpartum hemorrhage.** Heavy bleeding from the birth canal after vaginal delivery of a baby.

**Preferred Provider Organization (PPO).** A type of health plan where you pay less if you use providers in the plan's network. You can use doctors, hospitals, and providers outside the network without a referral for an additional cost.

**Preimplantation genetic diagnosis (PGD).** A form of genetic testing that analyzes and examines embryos during in vitro fertilization (IVF) before fertilized eggs are transferred back to the uterus in order to determine if the embryo has certain genetic conditions.

**Premium.** The amount you pay for your health insurance every month. In addition to your premium, you often must pay other costs for your healthcare, including a deductible, copayments, and coinsurance.

**Pre-mutation.** The presence of slightly more than the normal number of nucleotide repeats in the genetic mutation, e.g. in DM1, somewhere between 38 and 50; CTG repeats; the person exhibits no symptoms but are at risk of having affected children.

**Prenatal diagnosis.** A process of determining whether a child in the womb has a specific inherited disorder.

**Prevalence.** Counting existing disease diagnosis in a single point in time.

**Prognosis.** Forecast of the probable course and outcome of a disease.

**PROMM.** Abbreviation for proximal myotonic myopathy or DM2.

**Proximal.** In medicine, it refers to a part of the body that is nearest to the trunk of the body, such as thighs and upper arms.

**Ptosis.** Droopy eye-lids due to muscle atrophy.

**Pulse oximetry.** A test to measure oxygen levels in the blood.

**Respiratory function test.** A test that measures the amount of air a person can blow out.

**Repeat expansion.** A genetic marker which provides a rough measure of potential severity for DM1 which tends to increase with each generation. See Trinucleotide repeats below.

**Section 504.** Individuals with disabilities may not be excluded from participating in programs and services receiving federal funds. It also prohibits job discrimination against people with disabilities in any program receiving federal financial assistance. Students not eligible for an Individualized Education Program (IEP) may be eligible for a 504 plan.

**Single-nucleotide polymorphism.** DNA sequence variation occurring when a single nucleotide adenine (A), thymine (T), cytosine (C), or guanine (G) in the genome (or other shared sequence) differs between members of a species or paired chromosomes in an individual.

**Smooth muscles.** Muscles that are part of or surround internal organs, as along the gastrointestinal tract.

**Social Security Disability Insurance (SSDI).** A federal insurance program that provides cash assistance for individuals who have worked under Social Security long enough to have “insured status” and who are disabled. SSDI also includes disability benefits and other kinds of benefits for some family members of individuals who are insured and who have died, retired, or become disabled. Most individuals who qualify for SSDI also qualify for Medicare after they have been entitled to benefits for two years. For more information, visit: [www.socialsecurity.gov/disability](http://www.socialsecurity.gov/disability).

**Somatic.** Physical.

**Somatic mosaicism.** In DM1, the presence of different numbers of CTG repeats (the abnormality) found in different organs and systems within the same person; in DM2, the presence of different numbers of CCTG repeats found in different organs and systems within the same person.

**Sonogram.** An image created by ultrasound obtained by a computerized instrument; it can reveal internal parts of the body, such as thyroid gland or fetus in utero.

**Southern blot.** Method used in molecular biology for detection of a specific DNA sequence in DNA samples.

**Steinert’s disease.** The first name given to myotonic dystrophy when it was identified as a disease by Dr. Hans Steinert of Germany in 1909.

**Strabismus.** Inability of one eye to attain binocular vision with the other because of imbalance of the muscles of the eyeball--called also heterotropia, squint.

**Sudden heart block.** A condition of the heart in which the passage of an electrical impulse is arrested, wholly or in part, temporarily or permanently.

**Supplemental Security Income (SSI).** Supplemental Security Income (SSI) is a cash benefit for people who are elderly, blind, and/or disabled and who have very limited income and assets. Unlike Social Security Disability Insurance (SSDI), individuals do not need to have any work history to qualify for SSI assistance. SSI payments do not come from Social Security taxes and there is no insured status requirement as in SSDI.

**Tachyarrhythmia.** Very rapid heart beats.



**Talipes equinovarus.** An inversion of the foot in which only the outer side of the sole touches the ground; also called club foot.

**Testicular (or gonadal) atrophy.** Condition in men in which the reproductive organs (testes) shrink and may lose function.

**Tetranucleotide repeats.** As related to myotonic dystrophy, the series of 4 chemicals (abbreviated CCTG and found in the DNA of the ZNF9 gene, on the 3rd chromosome) that repeats itself more times than normal and causes myotonic dystrophy type 2.

**Tracheotomy.** The implantation of a tube into the trachea to assist patient with breathing; inserted through neck just below the thyroid gland.

**Trinucleotide repeats.** As related to myotonic dystrophy, the series of 3 chemicals (abbreviated CTG and found in the DNA of the DMPK gene, on the 19th chromosome) that repeats itself more times than normal and causes myotonic dystrophy type 1.

**ZNF9.** The mutated gene on chromosome 3 that causes DM2; sometimes called the zinc finger gene.