



A.

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

B.

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

Bradycardia

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

C.

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

abbreviaton for central nervous system; brain

Cognitive problems

difficulties with thinking, learning and memory

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

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

D.

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

E.

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

F.

Foot drop

partial or total inability to dorsiflex (turn upward) the foot

G.

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

H.

Haplotype analysis

molecular genetic testing to identify a set of closely linked segments of DNA

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

I.

Implanted Cardioconverter 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

In vitro fertilization

eggs are obtained from the female after her ovaries have been stimulated with infertility drugs. 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

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

J.

N/a

K.

N/a

L.

N/a

M.

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



N.

NICU

neonatal (new born) intensive care unit

O.

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

P.

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

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

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

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

Q.

N/a

R.

Respiratory function test

a test that measures the amount of air a person can blow out

S.

Smooth muscles

muscles that are part of or surround internal organs, as along the gastrointestinal tract

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

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

T.

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

Z.

ZNF9

the mutated gene on chromosome 3 that causes DM2; sometimes called the zinc finger gene