Great Ormond Street Hospital for Children NHS Foundation Trust: Information for Families

Glossary of terms used in the Neuromuscular Disorders Service

This glossary has been put together to help you during appointments and when reading the reports and letters about your child's visit to the Neuromuscular Clinic.

Letters and reports may have medical words and information, which are difficult to understand. If you are unable to find the definition from the list below or would like more specific details about a particular condition, contact the Muscular Dystrophy Campaign (telephone 020 7720 8055 or www.muscular-dystrophy.org or Contact a Family (telephone 0808 808 3555 or www.cafamily.org.uk)

Whenever you see a word in *italics*, it means that there is a separate definition for this particular word elsewhere in the glossary.

If you have difficulty understanding any of the words used, please ask a member of the Neuromuscular Team to explain during your child's outpatient appointment.

If you have any more questions, please ring the Children's Neuromuscular Centre on 020 7405 9200 ext 0517 or 1195 or bleep 2123 or 0228. You can also send an email to muscle.service@gosh.nhs.uk.

Abductor – Muscles which move a part of the body away from the midline (an imaginary line running through the centre of the body), such as raising your arm out from your side.

Acetylcholine (ACh) – A chemical substance involved in the transmission of a signal from the ***nerve*** to the muscle, causing it to contract.

Acetylcholine receptors – Acetylcholine receptors (AChRs) are part of the ***membrane*** around the ***muscle cell***, which binds to the ***acetylcholine*** that is released when we want to use a muscle.

Amino acids – The 'building blocks' of ***proteins***. The sequence of amino acids determines the shape, properties and role of the protein.

Amniocentesis – Removal of a sample of amniotic fluid during pregnancy to be tested for certain abnormalities. Cells from the unborn child can be extracted from this fluid, which surrounds the baby in the womb.

Antibodies – ***Proteins*** produced by the body's immune system in response to 'foreign material', such as germs.

Aspiration – Food, liquid or saliva entering the trachea (airway) instead of the oesophagus (gullet or food pipe). Can cause choking.

Atrophy – Muscle wasting.

Autoimmune disorders – Medical conditions where your immune system produces ***antibodies*** that attack your own cells. For example, Dermatomyositis, Polymyositis and Myasthenia Gravis.

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Autosomal inheritance – This means that an abnormal ***gene*** can affect either sex. It can be ***dominant*** or ***recessive***. See also ***autosomes***.

Autosomes – All the ***chromosomes*** other than the X and Y (the sex chromosomes) are known as autosomes. Also see ***autosomal inheritance***.

Axial – This word is often used to indicate the trunk and the neck. For example, axial weakness refers to weakness of the muscles in the neck and trunk.

Axon – An extension of a ***neuron*** that carries the signal to target ***cells***, such as a muscle cell.

Bilateral – Relates to both sides. For example, bilateral ankle tightness means both right and left ankles are tight.

Biopsy – Removal of a small amount of tissue, muscle or fluid for examination in the laboratory.

Calpain 3 – This is lacking or deficient in individuals with Limb Girdle muscular dystrophy Type 2A.

Carbohydrate – Carbohydrates (such as sugars and starches) are mainly used by the body to store energy or convert food into energy.

Cardiac – To do with the heart.

Cardiomyopathy – A heart condition. The most common type in children or young people with a neuromuscular condition is dilated cardiomyopathy. This involves enlargement and thinning of the heart muscle, deterioration of the muscle and loss or reduced movement of the heart wall. In rare cases, the cardiomyopathy can be 'hypertrophic', which involves significant thickening of the heart walls.

Carrier – Someone who has one normal and one abnormal copy of a pair of genes for a genetic disorder. A carrier of a ***gene*** for a ***recessive*** disorder will usually remain unaffected throughout life. A carrier can pass the faulty gene on to their children.

Carrier testing – Genetic testing to find out if a person who does not show symptoms of a condition nevertheless 'carries' a copy of a faulty ***gene*** which could be passed on to his or her children.

Cell – The basic structural unit of all living organisms. The cell is surrounded by a ***membrane***. Inside the cell is a structure called the ***nucleus***, which contains ***DNA***, arranged into ***chromosomes***. These chromosomes carry the ***genes*** that we inherit.

Central nervous system – Also known as CNS, this refers to the brain and spinal cord.

Cerebrospinal fluid (CSF) – Fluid found in the spaces inside the brain, surrounding the folds of the outside of the brain and in the spinal cord.

Chorionic villus sampling – Also known as CVS, this refers to the removal of chorionic villi for testing during pregnancy. Chorionic villi are cells situated on the wall of the uterus (womb) which form the early placenta (afterbirth). They have the same genetic make-up as the unborn baby and can be tested for certain abnormalities.

Chromosomes – These are cylinder-shaped bundles in the cell ***nucleus***, made of a very long thin strand of ***DNA***, coiled upon itself many times. Humans have 46 ***chromosomes*** (23 pairs) in most cells of their bodies. The sex cells (the unfertilised egg and sperm) contain only 23 unpaired chromosomes each. Fertilisation of a 23-chromosome egg by a 23-chromosome sperm produces a new 46-chromosome cell, which grows into a new individual. This means that one of each chromosome pair is inherited from each parent. The ***genes*** we inherit are located on the chromosomes.

Chromosome Karyotype – The chromosomal characteristics of a ***cell***.

Cobb Angle – Refers to the angle of curve in the spine. This is measured on an x-ray of the spine and indicates how mild or severe the curve is.

Congenital – Present at birth, or soon afterwards.

Cognitive – Relating to thinking, reasoning, remembering, imagining and learning words.

Consanguineous – Relating to or involving individuals that are closely related, such as first cousins.

Contractures – Shortening of muscles or tendons that prevents the associated joints from moving freely.

Convexity – Describes a curved or rounded shape like a sphere. May be used to describe the shape of the spine, such as 'right-sided convexity'.

Creatine kinase – A type of ***protein*** found in muscle. Some forms of muscular dystrophy are associated with raised levels of this protein and it can be used as an indicator of muscle damage.

Deletion – The loss of a piece of genetic material from a ***chromosome*** or ***gene***.

Desmin – A ***protein*** characteristically found in ***muscle cells***. In individuals with Desmin Myopathy there is an abnormal assembly of this protein.

Distal – Away from the centre of the body.

DNA (Deoxyribonucleic Acid) – the chemical composition of which ***genes*** are made. Each cell's ***chromosomes*** contain about two metres of DNA, but it is so thin that it is barely visible, even with the most powerful microscope. If your body's entire DNA were stretched end to end, it would be long enough to reach the moon and back about 10,000 times.

Dominant – In dominant ***inheritance*** if one of a pair of ***genes*** is altered, the person with that gene will show signs of the relevant disorder. In any pregnancy, there is a 50 per cent or 1 in 2 chance that the mother or father will pass their altered gene on to the child, who will also be affected.

Dorsiflexion – The foot or toes turning upwards.

Duplication – Where a part of a ***chromosome*** or ***gene*** is copied.

Dysferlin – A ***protein*** which is missing from people with Limb Girdle muscular dystrophy type 2B and Miyoshi myopathy.

Dysphagia – Difficulty in swallowing.

Dystonia – Problems due to muscle tone, such as writhing, twisting or spasms.

Dystrophin – This is a ***protein*** which is missing in boys with Duchenne muscular ***dystrophy*** and reduced in boys with Becker muscular dystrophy. Dystrophin binds to other proteins, which may also be missing in other forms of muscular dystrophy.

Dystrophy – Dystrophy comes from two Greek words – 'dys' meaning faulty and 'trophe' meaning nourishment. Muscular dystrophy refers to a group of disorders characterised by progressive muscle weakness and a loss of muscle tissue.

Echocardiogram (Echo) – The use of ***ultrasound*** to examine and measure the structure and function of the heart.

Emerin – A ***protein*** which is absent in people with the X-linked ***recessive*** form of Emery-Dreifuss muscular dystrophy. It is found in the nuclear ***membrane*** of ***muscle cells***.

Enzyme – A ***protein*** that triggers chemical reactions. For example, amylase in the intestine that breaks down the carbohydrates we eat (such as starch) to produce energy.

Exon – A section of a ***gene*** that contains the code for producing a protein. Also known as 'coding ***DNA***'.

Extensor – A muscle which allows any part of the body (such as an arm or finger) to extend or straighten.

Fasciculation – Muscle twitching causing rhythmic movements. For example, with the tongue.

Fatigability – When a person is tends to become extremely tired or exhausted during or after normal activity.

Flexor – A muscle which bends or flexes any part of the body, such as at the elbow or wrist.

Gastrostomy – An opening through the abdominal wall into the stomach. A feeding device is inserted through this opening, which allows a person to be fed directly into his or her stomach, bypassing the mouth and throat.

Gastro-oesophageal reflux – Backward flow of stomach contents into the oesophagus (gullet or food tube).

Genes – The coded instructions that control the make-up of every human being. Genes are made of ***DNA***. Each gene carries instructions for the production of a specific ***protein***. Genes usually come in pairs, one inherited from each parent. They are passed on from one generation to the next and are the basic units of ***inheritance***. Alterations in genes (mutations) can cause inherited disorders.

Gene therapy – A potential method of treatment for ***genetic disorders***. Only gene therapy aimed at somatic (non-reproductive) cells is allowed in humans. This means that the changes cannot be passed on to future generations.

Genetic counselling – Information and support provided by a specialist (usually a doctor or nurse) to people who have genetic conditions in their families, or who are concerned about the possibility of genetically transmitted conditions.

Genetic disorders – Conditions that result from alterations in the genetic make-up of an individual. They may be the consequences of defects in single ***genes***, or in whole ***chromosomes***, parts of which may be lost, duplicated, misplaced or replaced. Alternatively, they may result from the interaction of multiple genes and external factors such as the environment.

Genetic testing – Examination of an individual's genetic material to identify faults that may cause a disorder.

Genome – The complete set of ***genes*** of a person or organism.

Gower's Sign – Describes the way a child gets up from the floor by 'climbing up their legs' with their hands.

Habitus – Relating to the 'build' of an individual's body.

Hepatosplenomegaly – Enlargement of the liver and spleen.

Heterozygote – Someone with different alleles (different forms of the same ***gene*** with the same function found at the same place on the ***chromosome***) of one or more specific ***genes***.

Human genome project – This project was started to analyse the ***DNA*** of human beings. Its aims were to identify the location of every human gene on different ***chromosomes***, to determine each gene's chemical structure to show its function in health and disease, and to determine the sequence of ***nucleotides*** of the entire set of genes (the ***genome***).

Hypotonia – Floppiness.

Hypoventilation – Inefficiency of breathing that leads to reduction of oxygen in the blood and an increase in carbon dioxide. If this happens at night during sleep, it is known as 'nocturnal hypoventilation'.

Immune response – The body's reaction to 'foreign' material, such as an infection or a transplanted organ.

Immunocytochemistry – A method of examining ***cells*** in the laboratory.

Immunosuppressive therapy – Medicines that are used to prevent rejection after a transplant or to treat a variety of conditions by reducing or stopping the immune system from producing ***antibodies***.

In-frame mutation – A mutation which does not alter the ***reading frame***. These types of mutations are normally associated with milder symptoms as they usually allow for the production of a shorter version of the protein, such as BMD. See also 'Out of frame mutation'.

Inheritance – Individuals acquiring characteristics from a parent or ancestor.

KAFO (Knee Ankle Foot Orthoses or 'callipers') – They are used particularly by boys with Duchenne Muscular Dystrophy to prolong their ability to walk. They are also used to help children with other conditions to walk.

Lamin A/C – A ***protein*** found in the inside ***membrane*** surrounding the ***nucleus*** of the cell. A lack of lamin A/C is associated with Emery-Dreifuss muscular dystrophy (EMD2), Limb Girdle muscular dystrophy type 1B, Charcot-Marie Tooth disease type II and four other disorders not related to muscular dystrophy or related conditions.

Linkage studies – These are tests carried out on various family members to establish how a particular piece of genetic information runs through a family. It can sometimes be used to predict whether someone has inherited a faulty ***gene***.

Malignant Hyperthermia – An inherited disorder that causes unexpected and very serious reactions to certain anaesthetics. People with Central Core Disease are potentially at risk from this condition.

Manifesting carrier – A female carrier of an X-linked medical condition who has symptoms of that condition.

Medial malleolus – This is the visible pointed bit of bone in the ankle joint.

Membrane – Membranes form a barrier between the inside and outside of a ***cell***. Membranes protect cells and control which substances leave or enter a cell.

Merosin (laminin alpha 2) – The ***protein*** that is absent in Congenital muscular dystrophy type 1A (MDC1A)

Mitochondria – Mitochondria produce energy via a process called the respiratory chain, which involves many different ***proteins***. They have their own ***DNA***, which is inherited only from the mother.

MRI (Magnetic Resonance Imaging) – This technique uses magnetism and radiofrequency waves to collect information about the part of the body being examined. The scanner looks like a tunnel with open ends and the patient is asked to lie still on a table that slowly slides into the tunnel before the scan begins. abies and toddlers are usually given sedation so that they are asleep and not moving during the MRI scan.

Muscle cell – The basic unit of a ***muscle fibre***.

Muscle fibre – The basic unit of muscle tissue, formed by the fusion of groups of ***muscle cells***.

Mutation – When a ***gene*** is changed or altered in some way. Mutations can be passed on to subsequent generations.

Myelin – Insulating material that surrounds ***nerves*** and causes nerve impulses to be conducted faster. See also ***Nerve conduction***.

Myelination studies – Examination of the level of ***myelin*** in the body.

Myopathy – Muscle weakness.

Myositis – Muscle inflammation.

Myotonia – A condition where muscles are slow to relax after contracting (muscle stiffness).

Myotonin protein kinase (DMPK) – one of the ***proteins*** involved in myotonic dystrophy.

Myotubularin – The protein lacking in individuals with myotubular (centronuclear) myopathy.

Nasogastric feeding – The use of a special tube passed via the nose and oesophagus (gullet or food tube) down into the stomach to give liquid feeds to an individual.

Nebulin – A ***protein*** found in skeletal muscle, it is also thought to control the length of thin filaments within the muscle. Production of this protein is reduced in individuals with nemaline mypopathy.

Nerve conduction – A test that measures the electrical activity of muscles by using small discs placed on the leg or arm.

Nerves – Bundles of ***axons*** that transmit signals around the body.

Neuromuscular junction – The point where a ***muscle fibre*** and a ***nerve*** meet in the body.

Neuron – ***Cells*** that produce signals in the form of electrical impulses, which travel via the ***axons***.

Nucleotide – The building block of ***DNA*** and ***RNA***.

Nucleus – The structure in the centre of each ***cell*** which contains their ***chromosomes*** with their genetic material.

Obstructive sleep apnoea – A condition where someone stops breathing for a short time during sleep due to their airway becoming blocked.

Ocular – Relating to the eye.

Ophthalmoplegia – Difficulty in moving the eyes.

Orthoses – Devices or aids to assist movement and/ or positioning of the spine or limbs.

Out of frame mutation or frame-shift mutation – The removal or addition of one or more ***nucleotides*** which alters the ***reading frame***, severely affecting the production of the ***protein***, for example in some DMD ***mutations***. See also ***In-frame mutation***.

Oxygen saturation monitoring – Records oxygen levels in blood and relates to how effectively someone is breathing. The test is usually done for 8 to 10 hours overnight using a machine with a finger clip or pads that attach to a toe or the foot. This is usually used to check that an individual's oxygen levels stay at a good level all night while they are asleep.

Peripheral myelin protein P22 (PMPP22) – This is a ***protein*** lacking in individuals with some forms of Hereditary Motor and Sensory Neuropathies.

Peripheral nervous system – The part of the nervous system that links the spine and brain to the skin, muscles, blood vessels and other organs.

Phenotype – The visible picture (a combination of what a person can do and looks like mentally and physically) of an individual that is produced through interaction with their ***genes***.

Plantar flexion – Turning the foot or toes downwards.

Pneumothorax – Presence of air in the space between the outside of the lung and the ribs.

Podiatrist – Specialist who treats foot problems.

Preclinical diagnosis – Diagnosis of a genetic disorder before it is clinically recognisable. This means before the features of the child's condition show in the child.

Prenatal – Before birth.

Proband – The first family member to be diagnosed with a particular condition.

Prognosis – A prediction of what will happen to someone with a particular medical condition.

Protein – One or a few long chains of ***amino acids*** linked in a sequence.

Proximal – Close to the centre of the body.

Pseudohypertrophy – Muscles that appear to be developed (big), but only look this way due to fat and fibrous tissue.

Ptosis – Droopy eyelids.

Reading frame – The genetic code consists of a sequence of letters which are read in groups of three. This means that there are three possible ways of reading the sequence, depending on the starting point. For example, if the code reads AGCAGCAGC, the three reading frames are AGC AGC, GCA GCA and CAG CAG.

Recessive – A form of ***inheritance*** where a faulty copy of a ***gene*** is inherited from each parent. Therefore, to develop the disorder an individual has to have two copies of the faulty gene. This includes X-linked ***recessive*** and ***autosomal*** recessive modes of inheritance.

Respiration – To do with breathing.

Revertant fibres – Occasional fibres seen in muscle ***biopsies***, which contain forms of ***dystrophin***, even though the ***mutations*** in these individuals should prevent the production of this ***protein***.

RNA (Ribonucleic Acid) – This is a substance very similar to ***DNA***. When a ***gene*** is 'active', RNA copies of the gene's code are made (called 'messenger RNA' or 'mRNA') and move outside the ***cell nucleus*** where they are used to direct the manufacture of ***proteins***.

Sarcoglycan genes – The ***genes*** associated with LGMD type 2C, 2D, 2E and 2F. Mutations in these genes result in the reduction or absence of the ***sarcoglycan proteins***.

Sarcoglycan proteins – There are four sarcoglycan proteins – gamma, alpha, beta and delta. These ***proteins*** link the inside and outside of the cell across the ***membrane***.

Sarcoglycanopathies – Term for a group of Limb Girdle muscular dystrophies involving the ***sarcoglycan genes***.

Sarcolemma – A ***membrane*** or skin which surrounds each ***muscle fibre***.

Scoliosis – Curvature of the spine.

Sex chromosomes – The X and Y ***chromosomes*** determine the sex of an individual. Females have two X chromosomes; males have one X and one Y chromosome.

Silent aspiration – Breathing in of liquids or solids into the lungs without signs of choking or coughing.

Survival Motor Neuron gene (SMN gene) – This ***gene*** occurs in two forms – SMN1 and SMN2. Spinal Muscle Atrophy (SMA) is associated with the loss of the SMN1 genes.

Survival Motor Neuron protein (SMN protein) – produced by the ***SMN genes***. Production is reduced in individuals with Spinal Muscular Atrophy (SMA).

Stop codon – A section of ***DNA*** code that terminates ***protein*** production.

Talipes – Describes a ***congenital*** deformity of the feet. Sometimes called 'clubfeet'.

Tissue – A group of ***cells*** organised to carry out a particular job or function.

Trial – Experimental tests or treatments on animals or humans.

Ultrasound – Ultrasound examinations (scans) are done by building up pictures from the return of sound waves as they bounce back from any resistance they meet. The sound waves can be directed to and bounced back from the surfaces of various structures within the body to form pictures on a television screen. Ultrasound scans can be used to diagnose a variety of conditions. They are commonly used to scan babies during pregnancy.

Utrophin – A very similar ***protein*** to ***dystrophin***. All people (including boys with DMD) have low levels of utrophin in their blood, but not enough to compensate for the loss of dystrophin.

Ventricular dilatation – If referred to the heart, it means enlargement of the ventricle (pumping chamber), for example in dilated ***cardiomyopathy***. It can also refer to enlargement of the brain ventricles, for example in hydrocephalus.

X-linked – A form of ***inheritance*** where the gene in question lies on the X-***chromosome***. X-linked genes can be dominant, but are usually recessive. Girls who carry an altered gene are usually not affected, since they have a second, normal copy of the gene. However, they are carriers and there is a 50% chance that they will pass the affected gene on to their children. Boys, who have only one chromosome, do not have a second normal copy of the gene, so they will be affected by the disorder in question. They will pass on the altered gene to all of their daughters, who will be carriers, but to none of their sons.

If you have any questions, please ask the ward for advice.

Clinical Nurse Specialists in the Dubowitz Neuromuscular Centre – 020 7405 9200 ext 0517/1195 or bleep 2123/0228 or email muscle.service@gosh.nhs.uk

Notes

Compiled by the Dubowitz Neuromuscular Centre
in collaboration with the Child and Family Information Group

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