

## Looking into it Glossary of Terms Related to Neuromuscular Conditions

Click on one of the words below and be taken directly to all the words that begin with that letter [Ctrl + click the word]

|                                    |                       |                                |                       |
|------------------------------------|-----------------------|--------------------------------|-----------------------|
| <b>A</b> etiology                  | <b>B</b> iopsy        | <b>C</b> ardiac                | <b>D</b> iaphragm     |
| <b>E</b> lectrocardiogram          | <b>F</b> atigue       | <b>G</b> ene                   | <b>H</b> omozygous    |
| <b>K</b> yphosis (kyphotic curves) | <b>L</b> ocus         | <b>M</b> echanical ventilation | <b>N</b> euromuscular |
| <b>O</b> phthalmoplegia            | <b>P</b> hysiotherapy | <b>R</b> ecessive gene         | <b>S</b> coliosis     |
| <b>T</b> alipes                    | <b>W</b> ild-type     |                                |                       |

| <b>Term</b>                             | <b>Definition</b>  |
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| <a href="#"><u><b>Aetiology</b></u></a> | The cause of a disease.  |
| <b>Allele</b>                           | There are usually two copies of a gene. These two copies are called alleles.   |
| <b>Amniocentesis</b>                    | Cells from an unborn baby can be found in the fluid that surrounds it in the womb, the amniotic fluid. Amniocentesis means removing a sample of this fluid to test the cells from the unborn baby for specific conditions. |
| <b>Anticholinesterase Drugs</b>         | Any drug which prevents the enzyme acetylcholinesterase from working in the nerves. Acetylcholinesterase destroys the neurotransmitter acetylcholine which is necessary for the nerves to tell muscles to move.            |

| Term                                   | Definition   |
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| <b>Aphagia</b>                         | Unable to eat or swallow. Comes from the Greek terms a- (not) and phago (to eat).  |
| <b>Apnoea</b>                          | When someone stops breathing for a short amount of time. Sleep apnoea is when someone stops breathing for a short amount of time while they are asleep.  |
| <b>Aspiration Pneumonia</b>            | Pneumonia which has been caused by breathing in bits of food, fluid or other objects into the lungs, causing infection. It is more common in people who have weakened chest muscles.   |
| <b>Assisted Cough</b>                  | Applying pressure to someone's abdomen to help them cough. This helps to bring up secretions and mucous in the lungs. There are also mechanical devices to help with this.   |
| <b>Asymptomatic</b>                    | Showing no symptoms.   |
| <b>Atrophy</b>                         | The wasting away of a body organ. Muscle atrophy is the wasting away of muscles.   |
| <b>Autosomal Dominant Inheritance</b>  | An autosome is any of the chromosomes that is not a sex (X or Y) chromosome. When a disease or genetic trait is dominantly inherited only one copy of the gene (i.e one copy from one parent) is required for that disease or genetic trait to be expressed. |
| <b>Autosomal Recessive Inheritance</b> | An autosome is any of the chromosomes that is not a sex (X or Y) chromosome. When a disease or genetic trait is recessively inherited two copies of the gene are required (i.e one copy from each parent) for the disease or genetic trait to be expressed.  |
| <b>Autosome</b>                        | Any chromosome which is not one of the two sex chromosomes (X or Y). Humans have 22 pairs of autosomes (44 in all) and two sex chromosomes XX in females and XY in males.  |
| <b><a href="#">Biopsy</a></b>          | The removal of tissue from the body so that it can be analysed. A muscle biopsy involves removal of muscle tissue from the body.   |
| <b><a href="#">Cardiac</a></b>         | Related to, or affecting the heart.  |

| <b>Term</b>                            | <b>Definition</b>  |
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| <b>Carrier</b>                         | Someone who carries a normal version of the gene which is dominant, and a mutated form of the gene which is recessive. The mutated form will not be expressed and they will have either no or minimal symptoms, but they can pass the mutated gene on to their children.   |
| <b>Cell</b>                            | The basic subunit of any living tissue.  |
| <b>Cell Membrane</b>                   | A lipoprotein envelope that surrounds a cell.  |
| <b>Central Nervous System</b>          | The brain and the spinal cord.   |
| <b>Chorionic Villus Sampling (CVS)</b> | The chorion develops into the placenta. Chorionic cells have the same genetic composition as cells of the fetus. CVS is a procedure for obtaining cells of the chorion to enable testing of the foetus for specific abnormalities. Samples of cells may be taken through the vagina or through the abdomen of the pregnant mother – usually in the 10 <sup>th</sup> to 12 <sup>th</sup> week of pregnancy. |
| <b>Chronic</b>                         | When an illness or condition persists for a long time.   |
| <b>Chromosome</b>                      | Very long strands of DNA contained in the nucleus of cells. These strands contain the genes which carry most of someone's hereditary information. Humans have 23 pairs of chromosomes (46 in all).   |
| <b>Congenital</b>                      | Any abnormality or condition that is present from birth.   |
| <b>Contractures</b>                    | An abnormal shortening of muscle tissue. It is often caused by muscle weakness.  |
| <b>Creatine Kinase</b>                 | A type of protein found in muscle. When injury or disease causes damage to a muscle this protein leaks into the blood stream. A blood test is used to measure the level of creatine kinase in the blood.   |
| <b>Cyanosis</b>                        | A blue colour of the skin caused by a lack of oxygen in the blood.   |
| <b><u>Diaphragm</u></b>                | A large muscle used for breathing that is located between the chest and abdominal cavities.  |

| <b>Term</b>                              | <b>Definition</b>  |
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| <b>Diplopia</b>                          | Double vision.   |
| <b>Distal</b>                            | Away from the centre of the body, for example the hands and the feet.  |
| <b>DNA</b>                               | Stands for Deoxyribonucleic Acid. It is a twisted strand (double helix) of molecules that codes for hereditary information. Chromosomes and genes are made of strands of DNA. DNA is found mainly in the nucleus of the cell but there is also a small amount found in mitochondria. |
| <b>Dominant gene</b>                     | A gene that if present in someone will defiantly be expressed. If the dominant gene has a mutation that will cause a particular condition then the person will have the condition.   |
| <b>Dysarthria</b>                        | Difficulty speaking because the muscles that are needed to speak are too weak or not functioning properly.   |
| <b>Dysphagia</b>                         | Difficulty in swallowing.  |
| <b>Dyspnoea</b>                          | Hard to breath; short of breath.   |
| <b>Dystrophin</b>                        | The protein missing in people who have Duchenne MD and reduced in those who have Becker MD. The dystrophin protein normally sits in the membrane that surrounds muscle fibres like a skin. Lack of dystrophin leads to membrane damage and eventually muscle fibres death.           |
| <a href="#"><u>Electrocardiogram</u></a> | Using electrode stickers on the skin to measure the electrical activity of the heart. Used to diagnose heart abnormalities.  |
| <b>Electromyography</b>                  | An electrical test of the activity of the muscles and the nerves controlling the muscles. It is used to help diagnose MD.  |
| <b>Exacerbation</b>                      | Increase of the severity of a disease.   |
| <a href="#"><u>Fatigue</u></a>           | Great tiredness, both emotionally and physical, caused especially by hard work or effort.  |
| <a href="#"><u>Gene</u></a>              | Genes are made of DNA and each carries instructions for the production of a specific protein. Proteins produced by genes build up our cell structure and carry out cell functions. Genes usually come in pairs, one inherited from each parent. They are                             |

| Term  | Definition  |
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|   | passed on from one generation to the next, and are the basic units of inheritance. Any faults in genes can cause inherited disorders.   |
| <b>Genetic Counselling</b>                        | Information and support provided by a specialist, usually a doctor or a nurse, to people who have genetic conditions in their families or who are concerned about transmitting the condition to future children.  |
| <b>Genetic Disorders</b>                          | Conditions that result from alteration in the genetic make-up of an individual. They may be caused by defects in genes or chromosomes, or parts that may be lost, duplicated, misplaced or replaced. One or several genes can be affected.  |
| <b>Gowers' Sign</b>                               | A clinical sign named after the English physician who first described it in 1879. Whenever there is a weakness in the muscles around the hips, rising from the floor becomes increasingly difficult. The person has to press on his thighs and then climbs up them in order to extend the hips and straighten up. |
| <b>Heterozygous</b>                               | Describing an individual in whom members of a pair of genes (alleles) determining a particular characteristic are dissimilar  |
| <u><a href="#">Homozygous</a></u>                 | Describing an individual in whom members of a pair of genes (alleles) determining a particular characteristic are identical   |
| <b>Hypotonia</b>                                  | Floppiness, reduced tone of muscle.   |
| <u><a href="#">Kyphosis (kyphotic curves)</a></u> | Kyphotic curves refer to the outward curve of the thoracic spine (at the level of the ribs). A small degree of kyphotic curvature is normal. Too much kyphotic curving cause round shoulders or hunched shoulders (Scheuermann's disease).  |
| <u><a href="#">Locus</a></u>                      | In genetics it refers to the specific area on a chromosome where a gene is located.   |
| <b>Lordosis (lordotic curves)</b>                 | Refers to the inward curve of the lumbar spine (just above the buttocks). Too much lordotic curving is called swayback (lordosis). A small degree of lordotic curvature is normal.  |
| <u><a href="#">Mechanical ventilation</a></u>     | A medical procedure used to aid or replace breathing when a patient is unable to breathe adequately them selves. This usually involved a ventilator machine or manual bag   |

| Term                          | Definition   |
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|                               | compression to maintain the flow of air into and out of the lungs.   |
| <b>Muscle Cell</b>            | The basic unit of muscle fibre.  |
| <b>Muscle Fibre</b>           | The basic unit of muscle tissue formed by the fusion of groups of muscle cells.  |
| <b>Muscular Dystrophy</b>     | A group of muscle disorders characterized by progressive weakness and wasting of the muscles that control body movement.   |
| <b>Mutation</b>               | A permanent change in a gene. If the mutation occurs in the germ line cells (sperm or egg) it is then able to be inherited. Mutations can occur naturally or spontaneously or they may be due to exposure to mutagens. |
| <b>Myopathy</b>               | A disease of muscle that control body movement.  |
| <b>Myotonia</b>               | Inability to relax a muscle following contraction. It is often noted in the handgrip, or by trouble opening the eyes after forceful eye closure.   |
| <b><u>Neuromuscular</u></b>   | Anything that pertains to the nerves, muscles, or the nerve-muscle junction.   |
| <b>Nucleus</b>                | The control centre of a cell, which contains the cell's DNA.   |
| <b><u>Ophthalmoplegia</u></b> | Paralysis of the muscles of the eye that causes difficulty in moving the eyes.   |
| <b><u>Physiotherapy</u></b>   | The physical and therapeutic techniques commonly used to maintain strength and maximize range of motion, posture and comfort.  |
| <b>Prenatal testing</b>       | Testing for diseases or conditions in a foetus before it is born. This is done by analysing the DNA in sample of the fluid or tissue surrounding the foetus. See amniocentesis and chorionic villi sampling.           |
| <b>Prognosis</b>              | An assessment of the predicted course or outcome of a disorder based on knowledge of the course of the disease in other patients together with the general health and age.   |
| <b>Protein</b>                | It is a gene product responsible for cell structure and functions  |

| Term                                   | Definition  |
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|  | from eye colour to muscle functions.  |
| <b>Proximal</b>                        | Close to the centre of the body.  |
| <b>Pseudo hypertrophy</b>              | Increase in the size of an organ or structure caused by excessive growth of cells but this does not contribute directly to its functioning. The result is usually a decline in the efficiency. This is a common characteristic of calf muscles in Duchenne muscular dystrophy.  |
| <b>Ptosis</b>                          | Drooping of upper eye lid.  |
| <b>Pulmonary</b>                       | Related to, or affecting the lungs.   |
| <a href="#"><u>Recessive gene</u></a>  | Refers to a characteristic that is presented only when both of a pair of the genes coding for the characteristic are defected.  |
| <a href="#"><u>Scoliosis</u></a>       | Curvature of the spine, caused by abnormalities of the vertebrae, muscles and nerves. In neuromuscular disorders it most often results from muscular weakness or localised muscle imbalance. It can be treated with surgery, but there is varying degrees of success.   |
| <b>Sex Chromosome</b>                  | A chromosome that is involved in the determination of the sex of the individual. Women have two X chromosomes; men have one X chromosome and one Y chromosome.  |
| <b>Sex-linked</b>                      | Describing genes that are carried on the sex chromosome, usually the X chromosome. Since these sex-linked genes are recessive, men are more likely to have the diseases since they have only one X chromosome; women can carry the genes but their harmful effects are usually masked by the dominant alleles on their second X chromosome. |
| <b>Skeletal Muscle/striated muscle</b> | Refers to the muscles used in movement or posture. It does not include muscles of the digestive tract, or the heart muscle. They are also called voluntary muscles because they are under voluntary control.  |
| <b>Spinal fusion</b>                   | A surgical procedure carried out to correct scoliosis where the vertebrae in the spine are surgically joined.   |
| <a href="#"><u>Talipes</u></a>         | Also known as club-foot. A congenital deformity of one or both feet in which the patient cannot stand with the sole of the foot   |

| Term                             | Definition  |
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|                                  | flat on the ground.   |
| <b>Tracheostomy</b>              | A surgical opening of the trachea/windpipe through which a tube is inserted to assist with breathing.   |
| <u><a href="#">Wild-type</a></u> | In genetics, this refers to the healthy, un-mutated copy of a gene. This “normal” gene is the one that does not cause a disorder, and also is found most often in a population. |