Glossary of Terms

ACTH Adrenocorticotropic Hormone:
Adrenocorticotropic hormone is produced by the pituitary gland. Its key function is to stimulate the production and release of cortisol from the cortex of the adrenal gland

Addison's disease
Primary adrenal insufficiency, also called Addison's disease, occurs when adrenal gland production of cortisol is chronically deficient, resulting in chronically elevated ACTH levels. A deficiency of ACTH is a cause of secondary adrenal insufficiency. Primary adrenal insufficiency is found in 85% of young boys who develop the most severe form of ALD

Adrenal Glands:
These sit one top of each kidney and produce (secrete) three types of hormones:

- Glucocorticoid hormones produce cortisol which help maintain sugar (glucose) control, decrease immune response and helps the body to respond to stress
- Mineralocorticoid hormones regulate such things as sodium and potassium
- Sex hormones androgens (male) and estrogens (female)

Allele:

The variant form of a gene. For example, the gene for eye color has several variations (alleles) such as an allele for blue eye colour or an allele for brown eyes.

Allogenic Transplant:
Allogenic means that the transplanted cells are coming from a donor – this may be a sibling, other relative, or someone unrelated to the patient (the cells can even come from umbilical cord blood)

Alopecia:
A general term for hair loss or sparse hair

Amaurosis:
Partial or total vision loss without visible changes to the eye

Anaemia:
Shortage of red blood cells, causing fatigue, weakness and lack of energy as insufficient amounts of oxygen are transported around the body. Iron deficiency anaemia is a common type of anaemia caused by a lack of iron in the blood

Angiokeratomas:
Small dark red spots caused by enlarged blood vessels. These benign spots or lesions are more common in older people

Apnoea:
Temporary interruptions to breathing, especially during sleep – this is also known as Sleep Apnoea or Obstructive Sleep Apnoea (OSA). Over time, those with apnoea can feel increasingly fatigued, as breathing may be interrupted hundreds of times a night. There are a range of treatments available to relieve this

Ataxia:
Ataxia is a lack of muscle coordination which may affect speech, eye movements, the ability to swallow, walking, picking up objects and other voluntary movements. A person with persistent ataxia may have damage in the part of the brain that controls muscle coordination – the cerebellum

Athetosis:
Abnormal muscle contraction causes slow, involuntary writhing of the limbs, possibly continuously

Atrophy:
The wasting away or decrease in size of an organ or tissue in the body. When a body part is affected by an inability to function, the muscles may atrophy through lack of use and especially as a result of the degeneration of cells

Autologous Transplant:
Autologous means structures or cells came from you and your body. In an autologous transplant the patient “donates” their cells to them self

Autonomic dysfunction:
Damage to the nerves of the autonomic nervous system, causing dizziness, problems with digestion, urination, vision and sexual problems

Autosome:
An autosome is any of the numbered chromosomes, as opposed to the sex chromosomes. Humans have 22 pairs of autosomes and one pair of sex chromosomes (the X and Y). Autosomes are numbered roughly in relation to their sizes: chromosome 1 has approximately 2,800 genes, while chromosome 22 has approximately 750 genes.

Autosomal Inheritance:
The gene responsible for the phenotype is located on one of the 22 pairs of autosomes (non-sex determining chromosomes).

Autosomal Dominant:
Dominant conditions are expressed in individuals who have just one copy of the mutant allele. Affected individuals have one normal copy of the gene and one mutant copy of the

gene, thus each offspring has a 50% chance of inheriting the mutant allele. Approximately half of the children of affected parents inherit the condition and half do not.

Autosomal Recessive:
Recessive conditions are clinically manifest only when an individual has two copies of the mutant allele. When just one copy of the mutant allele is present, an individual is a carrier of the mutation, but does not develop the condition. Females and males are affected equally by traits transmitted by autosomal recessive inheritance. When two carriers mate, each child has a 25% chance of being unaffected; a 25% chance of being affected; or a 50% chance of being an unaffected carrier.

Bone Marrow Transplant:
A Bone Marrow Transplant (BMT) replaces bone marrow stem cells in people whose bone marrow has been destroyed by large doses of chemotherapy or radiotherapy. The healthy stem cells are put into your body intravenously, and then find their way into your bones to become healthy marrow. The transplant of healthy stem cells rescues you from your chemotherapy, and/or radiotherapy by enabling your bone marrow to start making new red and white blood cells and platelets.
They can be Allogenic or Autologous. Allogenic means that the transplanted cells are coming from a donor – this may be a sibling, other relative, or someone unrelated to the patient (the cells can even come from umbilical cord blood) or Autologous means structures or cells came from you and your body. In an autologous transplant the patient “donates” their cells to them self

Bradykinesia:
Unusually slow or difficult body movements, making everyday movements take much longer. Parkinson’s Disease is the most common but not only possible cause

Bulbar symptoms:
Symptoms which relate to the function of cranial nerves, including dysphagia, dysphonia and dysarthria

Camptodactyly:
Unusual, permanent curvature of one or more of the fingers, always including the little finger

Cardiomegaly:
Abnormally enlarged heart. This can be temporary or permanent, and can either cause no symptoms or cause shortness of breath, swelling and an irregular heart beat (arrythmia)

Central Nervous System (CNS):
The central nervous system consists of the brain and the spinal cord. The spinal cord carries out automatic motor responses, transmits sensory information to the brain and transmits messages from the brain to the muscles and organs. Each of the spinal cords segments controls sensation and movement in a different part of the body

Cerebellar Ataxia:

This relates to the cerebellum which is that large structure at the back of the brain that receives information from the sensory systems, the spinal cord, and other parts of the brain and then regulates motor movements. The cerebellum coordinates voluntary movements such as posture, balance, coordination, and speech, resulting in smooth and balanced muscular activity.
Cerebellar ataxia can occur as a result of lesions to the cerebellum and many diseases and presents with symptoms of an inability to coordinate balance, gait, extremity and eye movements

Cholestasis:
Reduction or stoppage of bile flow between the liver and small intestine, causing some waste products to accumulate in the bloodstream rather than being expelled in stools or urine. This can cause yellow (jaundiced) skin, light-coloured stools, dark urine and itchiness and can be caused by problems in the liver or in transit between the liver and small intestine

Chorionic Villus Sampling (CVS):
A sample is taken from the lining of the uterus (Chorion) which becomes the placenta 11-13 weeks from gestation. This contains foetal tissue and the DNA which is tested for the known mutation, VLCFA'S and other tests that look at chromosome make up, size etc (Karyotyping) to diagnose leukodystrophies and other conditions

Chromosome:
A chromosome is a long strand of DNA that contains many genes; there are 46 human chromosomes (22 pairs of chromosomes, and the X and Y chromosomes).
Women have 44 autosomes plus two copies of the X chromosome
Men have 44 autosomes plus an X and a Y chromosome

Cleft palate:
An opening in the roof of the mouth (palate) or upper lip, present from birth. This is the most common facial birth defect in the UK and is treatable with surgery. Without or before surgery, this can affect ability to hear and speak as well as creating dental and feeding issues

Coagulopathy:
Impaired ability of the blood to form clots (coagulate). This may mean that prolonged or excessive bleeding occurs, either spontaneously or after injury

Demyelination:
The destruction, removal or loss of the myelin sheath on a nerve or nerves. The myelin sheath is the protective covering around the nerve; damage to this causes nerve impulses to slow or stop, meaning messages sent by the brain cannot be received elsewhere in the body

DNA:
Deoxyribonucleic acid (DNA) is contained in your body's cells. It is a double, long chain of molecules called nucleotides that tell each cell what proteins to make. The **DNA** itself makes up chromosomes

Dysarthria:
Speaking difficulties caused by brain damage. Those with dysarthria may have slurred, nasal-sounding or breathy speech

Dysmetria:
A type of ataxia, dysmetria is a lack of coordination of movement. This can be linked to an inability to judge scale or distance

Dysmyelination:
As opposed to demyelination, where the previously normal myelin sheath is damaged or destroyed, dysmyelination relates to defective or malformed myelin sheath

Dysphagia:
Swallowing difficulties ranging from problems with certain foods to a complete inability to swallow

Dysphonia:
Speaking difficulties caused by physical disorder of the mouth or throat

Dystonia:
A state of abnormal muscle tone resulting in muscular spasm and abnormal posture, typically due to neurological disease. Dystonia is a disorder characterized by involuntary muscle contractions that cause slow repetitive movements or abnormal postures. The movements may be painful, and some individuals with dystonia may have a tremor or other neurologic features.

Encephalopathy:
A general term for brain disease, damage or malfunction, causing an altered mental state

Endocrine System:
The endocrine system is a collection of glands that secrete chemicals directly into the bloodstream. These chemicals are called hormones and some of those are: cortisol, aldosterone, adrenaline, noraadrenaline, testosterone, estrogen, oxytocin and dopamine. The main endocrine glands include: pituitary, pineal and hypothalamus, thyroid, pancreas, adrenal, testis and ovarie

Enzyme:
An enzyme is made from a group of complex proteins that are produced by living cells and act as catalysts in specific biochemical reactions. They speed up the chemical process. Not all enzymes are made from Amino therefore are not proteins. Enzymes are named by what they do

Epidemiology:
The study of patterns, causes, and effects of health and disease conditions in defined population

Equinus:

Limited ankle motion, inability to flex the foot towards the front of the leg

Erythema:
Red skin caused by increased blood flow after injury, infection or inflammation

Fontanelles:
Spaces between the bones of the skull as an infant develops, colloquially known as 'soft spots'

Gadolinium Enhanced MRI:
Gadolinium is used as a contrast agent to improve the visibility of internal body structures in magnetic resonance imaging (MRI). When having an MRI for ALD, gadolinium enhancement can show areas of inflammation/active disease within the brain

Gastrostomy:
A gastrostomy tube, sometimes referred to as a PEG (percutaneous endoscopic gastrostomy), is a procedure in which a flexible feeding tube is placed through the abdominal wall and into the stomach. PEG allows nutrition, fluids and/or medications to be put directly into the stomach, bypassing the mouth and throat

Genotype:
A genotype is an individual's collection of genes. The term also can refer to the two alleles inherited for a particular gene. The genotype is expressed when the information encoded in the gene's DNA is used to make protein and RNA molecules. The expression of the genotype contributes to the individual's observable traits, called the phenotype

Genome:
A genome is the genetic material of an organism

Gene Therapy:
Gene therapy is designed to introduce genetic material into cells to compensate for abnormal genes or to make a beneficial protein. It involves removing a sample of the patient's bone marrow – the building block of all of the body's immune cells – and exposing them to viruses carefully modified to carry a functioning copy of the faulty gene

Glial Cell:
A supportive cell in the central nervous system. Unlike neurons, glial cells do not conduct electrical impulses. The glial cells surround neurons and provide support for and insulation between them. Glial cells are the most abundant cell types in the central nervous system. Types of glial cells include oligodendrocytes, astrocytes, ependymal cells, Schwann cells, microglia, and satellite cells.

Globoid cells:
Large cells which usually have more than one nucleus

Grey Matter:

The “grey matter” of the brain gets its colour from the cell bodies, dendrites and the unmyelinated axons

Graft Versus Host Disease GVHD:
GVHD may occur after a bone marrow or stem cell transplant in which someone receives bone marrow tissue or cells from a donor. This type of transplant is called allogenic. The newly transplanted cells regard the recipient’s body as foreign. When this happens, the newly transplanted cells attack the recipient’s body. GVHD does not occur when someone receives his or her own cells during a transplant. This type of transplant is called autologous

Hematopoietic Stem Cell:
An immature cell that can develop into all types of blood cells, including white blood cells, red blood cells, and platelets. Hematopoietic stem cells are found in the peripheral blood and in the bone marrow. They are used in Bone Marrow Transplants (BMT) because the transplant of healthy stem cells can enable your bone marrow to start making new red and white blood cells and platelets

Hepatomegaly:
Abnormally enlarged liver

Hepatosplenomegaly:
Abnormally enlarged liver and spleen

Heterozygote:
An individual who has two different forms of a particular gene (allele), one inherited from each parent

Homozygote:
An individual who inherits the identical forms of a particular gene (allele) from each parent

Hydrocephalus:
Accumulation of cerebrospinal fluid in the brain, increasing pressure

Hyperextension of the Limbs:
The extension of the limbs beyond their normal movement

Hyperreflexia:
Overactive or over-responsive reflexes, including twitching and spasticity

Hypertelorism:
An abnormally increased distance between two body parts, most commonly the eyes

Hypertrophic cardiomyopathy:
Thickening of heart muscle forcing the heart to work harder

Hypodontia:
Developmental absence of one or more teeth, affecting both childhood and permanent teeth

Hypoglycaemia:
Low blood sugar

Hyporeflexia:
Decreased response of the deep tendon reflexes, usually resulting from injury to the central nervous system or a metabolic disease

Hypotension:
Sudden fall in blood pressure when standing up

Hypotonia:
Hypotonia is an abnormality of the skeletal muscle tone, which is indicative of genetic disorders or nervous system dysfunction. Patients with these tendencies display floppy limbs and an inability to sustain normal head position

Ichthyosis:
Persistent dry, scaly skin covering large areas of the body

Incidence:
Incidence is calculated as the number of new cases of a disease or disorder in a specified time. It is calculated by the number of new cases of a disease or disorder in a specified time, usually a year, divided by the population that is being considered. In rare diseases it can be very small and is often shown as the number of people per 100,000

Kyphoscoliosis:
Abnormal spine curvature in two places, combining outward curvature (kyphosis) and lateral curvature (scoliosis) of the spine

Leukodystrophy:
A genetic disorder primarily affecting central nervous system (brain or spinal cord) white matter

Leukoencephalopathy:
This simply means abnormal white matter in the brain and does not imply a genetic cause

Loes Score:
The Loes score was developed to measure the loss of myelin shown on a brain MRI. The higher the points the more myelin loss and it has a direct correlation to functionality.

Macrocephaly:
Unusually large head

Magnetic Resonance Imaging (MRI):

This procedure uses a very powerful magnetic field and radio waves to contrast the various soft tissues that we have in our bodies. This makes it ideal for looking at our brain. From the results the radiologists can see the difference in the white matter in our brain and where it is deficient and showing irregularities. The MRI scan helps produce a result called the Loes Score which rates the severity of brain damage

Megalencephaly:
Unusually large brain

Microcephaly:
Unusually small head

Microglia:
Microglia are the primary immune cells of the CNS. They act as the major inflammatory cell type in the brain and respond to pathogens (disease carriers) and injury by becoming "activated" – a process whereby they rapidly change form, proliferate and migrate to the site of infection/injury where they eat and destroy pathogens as well as remove the damaged cells

Micrognathism:
Undersized jawbone, also known as mandibular hypoplasia

Microphthalmia:
Developmental disorder of the eye, causing one or both eyes to be abnormally small

Mitochondria:
Mitochondria are the power houses of the cell providing the body with over 90% of the energy it needs to sustain life. Mitochondrial disease is a debilitating and potentially fatal disease that reduces the ability of the mitochondria to produce this energy

Myelin:
Myelin is the insulation material that is made from proteins and lipids (fats etc) that is produced in the neuron to coat and protect the transmission arm of the neuron – the axon. This process is called Myelination. Myelin is a spongy substance which is white in appearance and is referred to as "the white matter" of the brain.
The major purpose of the myelin is to increase the speed of transmission of an impulse. When this process is disrupted or destroyed by a dysfunctional metabolic process, with a genetic cause it can be classified as a Leukodystrophy

Myopathy:
Muscle weakness due to improper functioning of muscle fibres

Neuroimaging:
This term includes CAT Scans (Computerised Axial Tomography) and MRI's (Magnetic Resonance Imaging)

Neuron:

A neuron is a nerve cell. There are three types of neurons: sensory, motor and interneurons

Nystagmus:
Constant uncontrolled eye movements

Ophthalmoparesis:
Weakness of muscles which move the eyes

Optic atrophy:
Abnormality in the optic nerve, which carries information from the eye to the brain

Osteopenia:
Low bone density making bones weaker than normal

Osteoporosis:
Thinning bones, causing a tendency for fractures

Ovarian dysgenesis:
Abnormal development of the ovaries (the term gonadal dysgenesis refers to abnormal development of reproductive systems in both men and women)

Palatal myoclonus:
Spasms in the roof of the mouth (palatal muscles)

Paraparesis:
Paraparesis (paraplegia) refers to partial (-paresis) or complete (-plegia) loss of voluntary motor function in the pelvic limbs.

Paresthesia:
A pricking sensation, numbness or weakness in hands and feet

Paroxysmal kinesigenic dyskinesia:
Episodic abnormal movements affecting one or both sides of the body, ranging from mild to severe. These jerking or shaking movements are induced by sudden motion, such as being startled or standing up quickly

Peripheral Nervous System (PNS):
The peripheral nervous system consists of neurons that carry messages to and from the central nervous system. The PNS sends information to and from the body's internal structure that carry out basic life processes such as digestion and breathing

Peripheral Neuropathy:
Peripheral neuropathy is a condition that develops as a result of damage to the peripheral nervous system — the vast communications network that transmits information between the central nervous system (the brain and spinal cord) and every other part of the body. Symptoms can range from numbness or tingling, to pricking sensations (paresthesia), or muscle weakness

Peroxisomal:
Peroxisomes are a specialist part of a cell containing proteins and performing essential metabolic functions, such as the decomposition of fatty acids.

Phenotype:
A phenotype is an individual's observable traits, such as height, eye colour, and blood type. The genetic contribution to the phenotype is called the genotype. Some traits are largely determined by the genotype, while other traits are largely determined by environmental factors.

Photophobia:
Increased sensitivity to light

Polydactyly:
One or more extra toes or fingers, on one or both feet or hands

Portal hypertension:
High blood pressure to the liver

Prevalence:
Prevalence is the number of living cases of a disease or disorder that exist at any point in time. In rare diseases it can be shown as the number of people per 100,000

Prognathism:
Protruding upper or lower jawbone

Proteins:
Proteins are required for the structure, function, and regulation of the body's cells, tissues, and organs

Quadriplegia:
Quadriplegia is paralysis caused by illness or injury to a human that results in the partial or total loss of use of all their limbs and torso; Paraplegia is similar but does not affect the arms. The loss is usually sensory and motor, which means both sensation and control are lost

Scoliosis:
Scoliosis is a sideways curvature of the spine. Most cases of scoliosis are mild, but some children develop spine deformities that continue to get more severe as they grow. Severe scoliosis can be disabling. An especially severe spinal curve can reduce the amount of space within the chest, making it difficult for the lungs to function properly. In many cases, no treatment is necessary. Some children will need to wear a brace to stop the curve from worsening. Others may need surgery to keep the scoliosis from worsening and to straighten severe cases of scoliosis. Curvature of the spine in two places is called kyphoscoliosis

Seizure:

A seizure is a sudden, uncontrolled surge of electrical activity in the brain which will affect how a person appears or acts for a short time. It may produce a physical convulsion, minor physical signs, thought disturbances, or a combination of symptoms. The type of symptoms and seizures depend on where the abnormal electrical activity takes place in the brain, what its cause is, and such factors as the patient's age and general state of health. Some seizures can hardly be noticed, while others are totally disabling

Spasticity:
A state of increased tone of a muscle (and an increase in the deep tendon reflexes). For example, with spasticity of the legs (spastic paraplegia) there is an increase in tone of the leg muscles, so they feel tight and rigid and the knee jerk reflex is exaggerated

Spondylosis:
Degeneration of the discs of the spine

Strabismus:
This is the medical term for a squint, a condition where the eyes point in different directions. One eye may turn inwards, outwards, upwards or downwards while the other eye looks forward

Syndactyl:
Some or all fingers or toes are wholly or partially united, creating webbed skin. Underlying bones may or may not be fused together

Tinnitus:
Hearing noises not caused by an outside sources, such as a ringing in the ears, either continuously or sporadically

Vertical gaze palsy:
Restricted up-and-down eye movement, also known as Parinaud's Syndrome

Visceromegaly:
Abnormally large abdominal organs

Xanthomata:
Yellowish, cholesterol-rich deposits seen anywhere on the body, showing abnormal storage of fats

X-linked inheritance:
A mode of genetic inheritance where the gene responsible for the disease or disorder is located on the X chromosome. This typically means that the disease or disorder will have a greater effect on males: Males have one X and Y chromosome, whereas females have two X chromosomes. This means that if a female has a mutated gene on one X chromosome, the effects are somewhat balanced by the other X chromosome, while a male does not have this second X