

## **Glossary & Terminology**

During or following a diagnosis with a rare disease you may hear words or terms that are new or maybe confusing. Here are some helpful definitions for some of these words.

### **Acute condition or acute illness**

A medical condition of short duration that often starts quickly and may only change the person's ability to function for a short period of time. This is different to a chronic condition (see below).

### **Allied Health Professionals**

Health professionals, including psychologists, physiotherapists, pharmacists, social workers, occupational therapists and speech and language therapists.

### **Advance Care Planning**

Advance care planning is a process of discussion and reflection about goals, values and preferences for future treatment in the context of an anticipated deterioration in the child's condition.

### **Advocate**

A person who represents or speaks and may negotiate on behalf of a family while also empowering and supporting people to assert their way through the health care system.

### **Assessment**

The evaluation of the development status and intervention needs of a child.

### **Chromosome**

Chromosomes are minute particles within the cells of our bodies. Very simply, they are the building blocks which determine our individual characteristics such as eye and hair colour. Chromosomes are normally grouped together in 23 pairs (46 in all), half of which come from the mother and the other half from the father.

**Diagnosis**

The process of identifying a condition by its signs or symptoms using various diagnostic procedures, such as reviewing patient history, physical examination, scans, and laboratory tests. Not all illnesses have a complete diagnosis.

**Diagnostic odyssey**

This refers to the significant diagnostic delay and misdiagnosis, that is commonly faced by people when trying to achieve a diagnosis for a medical condition.

**Deletion (chromosome)**

This means that a part of the chromosome is missing. When genes are missing, there may be errors in the development of a baby, since some of the "instructions" are missing.

**DNA (deoxyribonucleic acid)**

DNA is a molecule that contains the genetic code that is unique to every individual. Think of this code as an instruction manual for making all the proteins that form our bodies and help them thrive.

**Enteral Feeding**

Enteral feeding is feeding into the stomach to promote normal growth and development. This can include oral feeding or tube feeding.

In the medical setting, the term Enteral Feeding is most often used to mean tube feeding.

**Epidemiology**

This is the study of how often diseases occur in different groups of people and why.

Epidemiological information is used to plan and evaluate strategies to prevent illness and as a guide to the management of patients in whom disease has already developed.

**Exome Sequencing**

This is also known as whole exome sequencing (WES), which is a genomic technique for sequencing all of the protein-coding regions of genes in a genome (known as the exome).

**Family Centred Care**

A method of service delivery that recognises parents as the experts on their child's needs; it promotes partnership between parents and the service providers and supports the family's role in decision making about services for their child.

**Fine Motor Skills**

Relates to the use of the small muscles of the body such as muscles in the face, hands, feet, fingers and toes.

**Genes**

These are 'the instructions for building our body and telling it how to work'. Genes are located on chromosomes – we normally have 46 chromosomes that are in pairs (for each of the 23 pairs, we get one from our mother and one from our father). Our entire genetic code is called a genome.

**Genetics**

Genetics is 'the study of how genes work and transmit information from parents to offspring. It can help us understand the risk of inheriting a genetic disease.

**Genetic Counselling**

This involves talking about a genetic condition with a health professional who has qualifications in both genetics and counselling. Genetic counselling can help you understand more about an inherited condition (a condition passed down from one or both parents) and what causes it. A genetic counsellor can talk you through genetic testing. They can also help you and your family adjust to living with a genetic condition and plan for the future.

**Genetic Counsellors**

are 'allied health professionals who work as part of a skilled team delivering personalised information and risk assessments to patients and their families with, or at risk of genetic conditions.

**Genetic testing**

This investigates variants in our genes, chromosomes, or proteins. A genetic test might investigate a single gene, search for large changes or rearrangements in our chromosomes or analyse the proteins formed from genes. Genetic testing also encompasses newer forms of investigations called genomic testing.

**Genetic variants**

These are 'changes in our genes that contribute to our uniqueness. Most genetic differences have no impact on our health but there are some variants (often known as a pathogenic variant mutation) that can cause disease. Health conditions that are caused by genetic variants are called 'genetic conditions'.

**Genetic variant of uncertain significance (VUS)**

This is a genetic variant identified in the DNA of an individual with a health condition, but it is not yet known if the variant causes or contributes to that health condition.

**Genomics**

This is 'the study and mapping of genomes – the full set of genetic instructions for an organism. It includes both human and other genomes and how these interact with the environment. Genomics can help doctors identify and diagnose genetic disorders and rare diseases, develop prevention strategies and tailor a patient's treatment.

**Genotype** This is the genetic makeup of an organism. The genetic makeup of an organism is coded for in its DNA, the hereditary material of the cell. Organisms DNA is organized into sections that code for proteins, called genes.

**Gross Motor Skill**

This relates to the use of the large muscles of the body such as muscles in the legs, arms and abdomen.

**Individual Service Plan (ISP)**

The individual service plan is a written document prepared by an interdisciplinary team in partnership with the parents describing the needs of the child and the goals of the service to be provided.

**Inter/multi disciplinary**

This is a team approach provides a formal structure for professionals representing different fields of expertise including doctors, nurses both hospital and community based, social workers, physiotherapist, occupational therapist, speech therapist and psychologists to share information and develop goals for a joint service plan. Family input is encouraged.

**Life Limiting Condition**

A condition from which there is no reasonable hope of cure and from which the person is expected to die. The condition cannot be reversed by treatment. Life limiting conditions/illnesses progress quickly, and others cause slow deterioration over time rendering the child increasingly dependent on parents and carers. Life Limiting also includes life-shortening from complications that are secondary to the condition.

### **Life Threatening Condition**

A condition that actively and continually threaten the person's life; for which there may be considerable and effective treatment, yet cure is either unlikely or not necessarily guaranteed and/or death is inevitable. This means diseases or conditions where the likelihood of death is high.

### **Multi-sensory Therapy**

A specifically designed environment which enables a child with special needs to enjoy a wide range of sensory experiences through vision, sound, touch, smell, and taste for therapy, learning, relaxation and fun.

### **Muscle Tone**

Hypertonic – rigid muscle tone, tension in muscle when resting. Hypotonia – low muscle tone, floppy baby syndrome.

### **Nasogastric tube feeding (NG)**

A NG tube is a plastic tube inserted through the nose and it passes down into the stomach and is used for feeding.

### **Neurodevelopmental Delay**

Neurodevelopmental Delay is failure to meet certain developmental milestones normally achieved during infancy and early childhood usually caused by organic, psychological or environmental factors. Developmental delay indicates a problem in normal development in the central nervous system.

### **Newborn Screening (NBS)**

National Newborn Bloodspot Screening Programme and the National Universal Newborn Hearing Screening Programme. The key focus of both screening programmes is early identification of serious medical conditions and appropriate interventions to reduce mortality and/or morbidity in our population. The HSE Newborn screening programme currently screens for 9 conditions.

## **Occupational Therapy**

Occupational Therapy (OT) looks at how your child is developing the skills which are used in everyday life. This includes his/her ability to carry out self-care tasks such as eating and dressing, playing with toys and other children and as your child grows the possibility of writing and concentration skills. The assessment is done by observation of the child and by discussion with the parents. OT aims to develop fine motor, cognitive and perceptual ability, along with self-care skills.

## **Orphan Drug**

This is a pharmaceutical agent that is developed to treat certain rare medical conditions. An orphan drug would not be profitable to produce without government assistance, due to the small population of patients affected by the conditions. The conditions that orphan drugs are used to treat are referred to as orphan diseases.

## **Palliative Care**

Palliative care is inter-disciplinary care that aims to improve quality of life of individuals with Life Threatening Conditions or Life Limiting Conditions seeking to reduce pain and distressing symptoms while attending to a wide variety of psychological, social and spiritual needs. Palliative Care is patient centred and family focused; it can complement curative and life prolonging interventions from the time of diagnosis onward provided in home, hospital or hospice, while also supporting family members in bereavement.

## **PEG feeding tube (PEG)**

PEG stands for percutaneous endoscopic gastrostomy, 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.

## **Phenotype**

This term refers to 'the observable physical properties of an organism... [including its] appearance, development and behavior. Examples of phenotypes include height, wing length, and hair colour. Phenotypes also include observable characteristics that can be measured in the laboratory, such as levels of hormones or blood cells.

## **Physiotherapy**

Most children with disability will be referred for physiotherapy; first the physiotherapist will access the child and look at his/her ability to hold their head, move, sit and crawl. The Physiotherapist will also look at balance, the way the muscles and joints work, the way the heart and lungs function and how your child responds to sensory information. They identify areas of concern and develop treatment goals and a programme to work with at home. They will support this programme with follow up appointments adjusting and modifying the programme according to your child's needs.

### **Picture Exchange Communication System (PECS)**

Picture exchange is used to help motivate children to request objects/food/activities if they don't have the words to request them through speech.

### **Prognosis**

Prognosis refers to the doctor's expectation of how a child's condition/disease will progress. This judgement is based on their knowledge and experience with children with similar medical conditions.

### **Psycho-Educational Assessment**

A psycho-educational assessment is a comprehensive assessment of your child's level of cognitive functioning (activities associated with learning and problem solving). During the assessment procedure, this may take place over several sessions and it involves a variety of verbal and non-verbal performance tasks to ascertain his/her level of ability. Arising from the findings, specific recommendations regarding the most appropriate school placement are made.

### **Registries**

These are collections of standardised information about a group of patients who share a condition or health experience.

#### **A patient registry**

This is a standardised system that collects uniform information from patients, to evaluate specified outcomes for a population defined by a particular disease, condition, or exposure and that serves predetermined scientific, clinical, or policy.

### **Respite care**

Respite is short term care for the child to allow the carer a rest/break. Respite can be in a hospital, hospice or the home.

**Speech and Language Therapy (SLT)**

Speech and Language Therapy (SLT) aims to help your child to develop his/her communication, eating, drinking and swallowing skills to help reach potential in this area. Early intervention is important so SLT will begin prior to your child's first word. The SLT will assess your child's understanding, expression, how he/she manages sucking, swallowing and chewing. They will also assess how your child plays with objects and people. They then provide an individual programme to work with at home.

**Undiagnosed or Syndromes Without A Name (SWAN).**

This refers to a 'patient for whom a diagnostic test is not yet available since the disease has not been characterised and the cause is not yet identified. This patient can also be misdiagnosed as his/her condition can be mistaken for others. These conditions are also likely to be rare.

**Video fluoroscopy**

A specialised x ray to assess eating, drinking and swallowing.