

Medical Terminology in Plain English



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This glossary helps you to understand some of the words which are often used in respect of genetic conditions or inborn conditions. Please also see our: [Demystifying Medical Terminology](#) document.

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What are Genes?

The genes are the individual instructions written in genetic code that we pass on to our children. There are estimated to be around 60,000 individual genes. Our genes are fixed when the egg and sperm unite and remain the same throughout our lives.

What are Chromosomes?

The chromosomes are like the wrapping around the genes. We cannot see individual genes under the microscope but we can see the chromosomes. There are 23 pairs of chromosomes making 46 chromosomes in all. Men have an X and a Y chromosome while women have two X chromosomes. The other chromosomes are the same in men and women.

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What is a Genetic Mutation?

Genetic disorders have got to start somewhere and in most cases this happens by chance when we copy the genetic material. The amount of information we pass on in each egg or sperm is equivalent to many volumes of an encyclopaedia and, if you imagine photocopying this, you will see how mistakes in copying could arise. As we make billions of sperm and millions of eggs you will see that the copying is usually very good, but occasionally by chance, a new mutation will arise.

What is X Linked Inheritance?

On some occasions disorders will only affect men and can be carried by women. These are X linked or sex linked disorders.

What is Autosomal Dominant Inheritance?

In this form of inheritance the genetic disorder can be passed on from generation to generation. If an individual is affected they have a 50:50 chance of passing on the disorder in each pregnancy. Autosomal dominant disorders can affect both men and women.

What is Autosomal Recessive Inheritance?

Our genes come in pairs and in Autosomal Recessive Inheritance both parents have one normal gene and one abnormal gene which means they are carriers. If they both pass on an abnormal gene then the child will inherit a "double dose" and would be affected. The chances of this happening again in future pregnancies is 1 in 4 or 25%.

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What is Sporadic Inheritance?

A genetic condition that occurs in a random or isolated manner and describes the occurrence of a mutant gene.

What is Multifactorial Interference?

These include conditions that have a genetic element, but do not cover Mendelian patterns of inheritance. A combination of genetic and environmental factors, e.g. cleft lip and palate.

Chromosome Terminology:

Aneuploidy

Individual whole chromosomes are missing or extra; i.e. having less than or more than the normal diploid set of 46 chromosomes.

Autosome

The term used to denote any of the 22 paired chromosomes, excepting the sex chromosomes.

Biparental

Involving, or inherited from both parents.

Centromere

The specialised area of a chromosome at which it is divided into its short and long arms.

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Chromosome Abnormality, Nomenclature

Descriptions of particular chromosome formations are written in a shortened form which indicates the total number of chromosomes, the sex of the individual and the number of the abnormal chromosome.

The short arm of a chromosome is designated by a letter 'p'.

The long arm by a letter 'q'.

An additional chromosome or portion of a chromosome is designated by a plus sign (+).

The absence of a chromosome or portion of a chromosome by a minus sign (-).

A ring formation is designated by a letter 'r'.

EXAMPLES

Deletion example 1:

46, XY 18p- - Denotes a male (one X and one Y chromosome) with the correct number of chromosomes (46) but with a missing short arm (p) of one number 18 chromosome.

Deletion example 2:

45, XO Syndrome - Denotes a female with absence of one of her X chromosomes.

Ring formation example:

46, XXr13 - Denotes a female (two X chromosome) with the correct total number of chromosomes (46) but with the ends of a number 13 chromosome curved to form a ring.

Trisomy example 1:

47, XY +21 - Denotes a male (one X and one Y chromosome) with a total chromosome count of 47. The extra chromosome is an additional chromosome 21.

Trisomy example 2:

47, XXY Syndrome - Denotes a male with an additional X chromosome.

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Deletion

The absence of a chromosome, or part of a chromosome. The absence of a whole autosome is usually lethal but absence of a short arm or long arm, or part of an arm is fairly common.

Diploid

The term for a cell with a normal chromosome number, made up of two sets of parental chromosomes (23 from each parent).

Disomy

A normal chromosome pair, i.e. 23 chromosomes from each parent.

Dominant Gene

A gene which can produce its effect over its partner gene; i.e. it can produce its effect by 'dominating' its normal partner, whereas a recessive gene can only act when paired with another gene for the same characteristic.

Duplication

A double copy of part of a chromosome, resulting in an extra (abnormal) dose of the duplicated material. Also termed trisomy.

Homologous Chromosomes

A pair of chromosomes having the same structure and gene sequence, each derived from one parent. Humans normally have 22 pairs of homologous (autosomes) and two sex chromosomes, XX (homologous) in females and an X and a Y chromosome (non-homologous) in males.

Non-Homologous Chromosomes

A pair of chromosomes which are not identical in gene structures, e.g. XY in males.

Interstitial

Any part of a chromosome between its ends.

Interstitial Deletion

A situation in which two breaks occur in a chromosome and the intervening material is lost, resulting in an unbalanced amount of chromosome material.

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Inversion

A chromosome may break in two places and the middle piece re-attach itself after turning upside down. Providing no chromosome material is lost, this abnormality should have no effect on an individual. However, he or she may carry the risk of producing eggs or sperm with the incorrect amount of chromosome material.

Pericentric Inversion

An inversion of chromosome material, involving the centromere.

Paracentric Inversion

An inversion of chromosome material not involving the centromere.

Karyotype

A record of the chromosomes of a body cell. A photograph is taken using a microscope, and then arranged so that the chromosomes are in pairs of decreasing size. Used for the detection of abnormality in the number, size or structures of chromosomes.

Meiosis

Division of the sex cells, whereby the chromosome number is halved, as distinct from mitosis.

Mitosis

Division of all cells, except the sex cells, whereby identical cells are duplicated, as distinct from meiosis.

Mixoploidy

A mixture of normal diploid and abnormal polyploid cells, resulting in mosaicism.

Monosomy

A single copy of a chromosome or part of a chromosome, instead of the normal pair.

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Mosaicism

A situation in which some cells have the correct number of chromosomes (46) and others have an incorrect number; i.e. too many, or too few.

For example, in the mosaic form of Down Syndrome (Trisomy 21) a proportion of the body cells carry 47 chromosomes and the remainder carry the correct number of 46. In a mosaic disorder, the extent and severity of features depends upon the proportion of normal cells to abnormal ones and upon which tissues are involved.

Polyploidy

Having three or more sets of homologous chromosomes.

Recessive Gene

A gene which is the weaker of a pair and whose characteristic is only expressed if paired with another recessive gene for the same characteristic.

Ring Formation

The two ends of a chromosome may become damaged and curl over so that the ends of the short arm and the long arm almost meet, forming a ring shape, resulting in the loss of chromosome material.

Sex Chromosomes

The X and Y chromosomes which are responsible for sexual development. In humans, females have two X chromosomes and males have an X and a Y. Genes on the Y chromosome are responsible for male sexual development.

Sex Chromosome Abnormality Disorders

Disorders of fetal development caused by defects in the sex chromosomes, as distinct from the autosomes.

Tetrasomy

The presence of four copies of a chromosome, or part of a chromosome, instead of the normal two.

Tetraploidy

The presence of four extra sets of chromosomes, giving a total chromosome count of 92.

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Triplication

Two extra copies of a chromosomal segment. Also termed partial tetrasomy.

Triploidy

The presence of a complete set of extra chromosomes, giving a total chromosome count of 69.

Trisomy

The addition of a complete extra chromosome, or part of a chromosome, to a pair, resulting in three copies of the chromosomal material at the site. Where there is a complete extra chromosome the total chromosome count is 47.

Uniparental Disomy

Both members of a chromosome pair are contributed by one parent rather than one from each parent.





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