Liverpool Women's NHS Foundation Trust

Dedicated to you

Clinical Genetics

Glossary of Genetic Terms for Patients

ACQUIRED

A condition or disease originating after birth. An acquired condition is not hereditary.

ACUTE

A sudden onset of symptoms or disease which may be severe and/or brief in duration.

ALLELE

Alternative forms of a gene responsible for alternative traits.

AMNIOCENTESIS

Procedure used in prenatal diagnosis to obtain amniotic fluid which can be used for genetic and other diagnostic tests. This test can be performed as early as 16 weeks gestation.

ANEUPLOIDY

The presence of more or fewer than the usual (diploid) number of chromosomes, giving rise to an abnormal chromosome number. There may be partial aneuploidy, where there is the addition or loss of part of a chromosome.

AUTOSOME

Any chromosome other than the sex (X or Y) chromosomes.

AUTOSOMAL DOMINANT

Where an individual possesses an altered gene and demonstrates characteristics of the condition. The condition will usually be transmitted to children with a fifty per cent chance of an affected child for each pregnancy.

AUTOSOMAL RECESSIVE

Where an individual carries an altered gene but it does not affect the individual (they do not display any characteristics of the condition). However, when two carriers with the same altered gene have children there is usually a twenty-five per cent chance of an affected child for each pregnancy.

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NHS

BRCA1/BRCA2:

The first breast cancer genes to be identified.

CARRIER

An individual who carries an altered gene for a specific condition without symptoms.

CELL

The basic structural unit of all living organisms. It is surrounded by a membrane and contains a nucleus that carries genetic material.

CLINICAL GENETICS

Clinical Genetics is the medical specialty, which is concerned with the cause, course, diagnosis and treatment of genetic and part-genetic disorders. Clinical geneticists provide a diagnostic service and genetic counselling for individuals and their families with, or at risk of, conditions which may have a genetic basis.

A genetic counsellor provides information and support to families in making decisions relating to genetic disorders.

CONSANGUINEOUS

Where two people share at least one common ancestor ("blood relatives").

CONSULTAND

A person requesting genetic counseling

CHORIONIC VILLUS SAMPLING

A method of collecting the chorion cells from the pregnant mother. Chorion cells are situated on the wall of the uterus (womb). They have the same origin as the fetal cells and can, therefore, be analysed to detect certain fetal abnormalities. This test is performed in early pregnancy i.e.11-13 weeks.

CHROMOSOME

A rod-like structure present in the nucleus of all body cells (with the exception of the red blood cells), which stores genetic information. Normally, humans have 23 pairs, the unfertilised ova and each sperm carrying a set of 23 chromosomes. On fertilisation the chromosomes combine to give a total of 46 (23 pairs).

CHRONIC

Long-term or prolonged disease or condition that can persist or progress over a long period of time.

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CONGENITAL

A condition that is present at birth, although it can be recognised prenatally, at birth or many years later. A congenital disorder can be genetic or acquired at any time during fetal development.

DEGREE OF RELATIONSHIP

The degree of relationship helps define the proportion of genes shared by two blood relatives. First degree relatives (parents, siblings, children) share 1/2 of their genes, second degree relatives (uncles, aunts, nephews, nieces, grandparents, grandchildren and half- siblings) share 1/4 of their genes and third degree relatives (first cousins, great-grandparents and great-grandchildren share 1/8 of their genes.

DELETION

The absence of genetic material on a chromosome.

DIZYGOTIC

Two cells having been fertilised resulting in twins.

DNA

Deoxyribonucleic acid is the 'building block' for all genetic material.

DUPLICATION

Occurs where a chromosome or part of a chromosome is duplicated. This may happen during cell division prior to fertilisation.

FAMILIAL

Characteristic of some or all members of a family.

FRAGILE SITE

A site on a chromosome where genetic material may be prone to break. In Fragile X syndrome there is a fragile site on the female X chromosome.

GAMETE

A reproductive cell, either the male sperm or the female egg.

GENE

The functional and physical unit of heredity passes from parent and offspring. Genes are pieces of DNA, and most genes contain the information for making a specific protein.

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GENETIC COUNSELLING

The process by which information on genetic disorders is given to a family and supporting them in making their own decisions.

GENOME

All the DNA contained in an organism or a cell, which includes both the chromosomes within the nucleus and the DNA in mitochondria.

GENOTYPE

The genetic identity of an individual that does not show as outward characteristics.

INVERSION

Occurs where a chromosome breaks in two and becomes reattached after turning round 180 degrees. Providing no chromosome material is lost, this abnormality should have no effect on the individual. However, that individual may carry the risk of producing eggs or sperm with the incorrect amount of chromosomal material.

KARYOTYPE

The chromosomal complement of an individual, including the number of chromosomes and any abnormalities. The term is also used to refer to a photograph of an individual's chromosomes.

LOCUS (LOCI)

The precise physical site or location of a specific gene on a chromosome.

p denotes the short arm of the chromosome

q denotes the long arm of the chromosome

MITOCHONDRIA

Refers to the small bodies that are responsible for energy production. Mitochondria also carry their own genes and DNA.

MITOCHONDRIAL INHERITANCE

The condition can only be transmitted by females in the maternal line. Typically a mitochondrial inherited condition can affect both sexes.

MITOSIS

Division of all cells (except the reproductive cells which is called Meiosis) and which results in the daughter cells having the same chromosome number and genetic make-up compared with as the parent cell.

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MONOSOMY

The total loss of one of a pair of chromosomes. This occurs, for example, in Turner syndrome where one X chromosome is lost leaving a total of 45 chromosomes.

MONOZYGOTIC

Two individuals (twins) born together from one sperm and one egg.

MOSAICISM

Where a genetic or chromosomal abnormality does not occur in all body cells. The proportion of normal to abnormal cells will determine the severity of the disorder.

MULTIFACTORIAL INHERITANCE

This type of inheritance is caused by the summation of the effects of multiple genes interacting with environmental factors to produce a liability to a particular disorder. A person is affected if his or her liability is above a certain threshold.

MUTATION

A mutation refers to any change from the normal to an altered form of a particular gene. This may be disease causing or a benign, normal variant.

NUCLEUS

A body in the centre of each cell that contains the chromosomes with their genetic material.

OVUM

The female reproductive cell, also known as an egg, carrying 23 chromosomes.

PATTERN OF INHERITANCE

Describes how the gene is inherited: for example, autosomal dominant, autosomal recessive or X-linked.

PEDIGREE

A simplified diagram of a family's genealogy that shows family member's relationships to each other and how a particular trait of disease might have been inherited.

PHENOTYPE

The observable characteristics and physical appearance of an individual.

PLACENTA

A complex structure occurring in pregnancy. It is attached to the wall of the womb and connected to the fetus by the umbilical cord. The fetus receives its nourishment through the placenta and the vessels of the umbilical cord.

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PREDISPOSITION (GENETIC)

That the individual concerned is intrinsically more prone to develop a particular disorder.

SEX CHROMOSOME

One of the two chromosomes that specify an organism's genetic sex. Humans have two kinds of sex chromosomes, one called X and the other Y. Normal females possess two X chromosomes and normal males one X and one Y.

SOMATIC CELL

Any cell in the body except the gametes.

SPERM (abbreviation of spermatozoon)

The male reproductive cell carrying 23 chromosomes.

TRAIT

Any inherited (gene determined) characteristic.

TRANSLOCATION

Occurs where there is a rearrangement in which a piece of one chromosome is transferred to another with a different number.

TWINS

May be genetically identical (monozygous) when they arise from a single fertilised egg or non-identical (dizygous) when they arise from two separate eggs.

X-LINKED RECESSIVE

The form of inheritance where the altered gene is carried on the X chromosome. With this form of inheritance, girls are usually carriers and boys are affected. This is because the male Y chromosome does not carry the compensatory normal gene.

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