Abnormal: Any change from the 'correct' or 'usual'. It may not necessarily mean harmful or undesirable; it can equally mean atypical, unusual or uncommon. When used in reference to genes, an abnormal gene may result in a specific genetic condition.

Acrocentric: Acrocentric chromosomes are those with the centromere very close to the outer tip, giving the chromosome a 'V' shaped appearance. The acrocentric chromosomes are numbers 13, 14, 15, 21 and 22. The short p arms are very short and usually have small round appendages on stalks, known as 'satellites'.

Adult stem cell: An undifferentiated cell found in a differentiated tissue that can renew itself and (with certain limitations) differentiate to yield all the specialised cell types of the tissue from which it originated.

Allele: There are usually two copies of a gene. These two copies are called alleles. In some cases, one or both, alleles will be mutated or altered in some way.

Alphafetoprotein (AFP): A protein, which is made by the fetus, that can be found in the mother's blood circulation. The amount of this protein, both in the mother's blood and in the amniotic fluid, at particular periods during the pregnancy, may be associated with the presence of neural tube defects or chromosomal problems in the baby.

Antibody: A protein, produced in response to a foreign substance in the immune system.

Anticipation: The situation where a genetic condition appears to become more severe and/or arise at an earlier age as it is passed through subsequent generations (seen in some trinucleotide repeat mutations).

Amino acids: Small chemical building blocks that join together to form proteins: there are 20 common amino acids which join in different combinations to make up proteins.

Amniocentesis: A procedure for obtaining amniotic fluid for prenatal diagnosis. Using a sterile needle, a sample of amniotic fluid is removed from the uterus; the amniotic fluid contains cells from the fetus that can be analysed to determine if the fetus has a specific condition. The test is usually carried out in the 14th - 18th week of pregnancy (see Genetics Fact Sheet 17C: Prenatal Testing – CVS and Amniocentesis).

Amniotic fluid: Fluid in which the fetus floats in the uterus; fetal cells are found suspended in this fluid.

Assisted reproductive technology (ART): A term describing a variety of measures employed to increase the possibility of pregnancy. Includes IVF (*In Vitro* Fertilisation).

ATP: Stands for adenosine triphosphate. ATP is a chemical that is used in cells to drive chemical reactions in which energy is produced.

Autosomal gene: Any gene which is located on an autosome.

Autosomal dominant mutation: A dominant mutation in a gene which is carried on an autosome.

Autosomal recessive mutation: A recessive mutation in a gene which is carried on an autosome.

Autosome: Any chromosome that is not a sex chromosome (that is not an X or Y chromosome). In humans, the autosomes are the numbered chromosomes and are given the numbers 1 - 22. Chromosome 1 is the largest and 22 is the smallest.

Balanced translocation (reciprocal translocation): A rearrangement of the chromosomes with no apparent loss or gain of chromosomal material. A person with this rearrangement is not affected in any way. When a translocation chromosome results in the gain or loss of genetic material, it is said to be unbalanced and may cause a problem in health, growth or development.

Banding: A series of dark and light stripes across a chromosome which are produced by treating (staining) the chromosomes with different chemicals in a laboratory. There are a number of different staining techniques which produce different patterns eg. G-banding, R-banding etc.

Bases: Also known as nucleotides, they are the basic components of DNA. They are denoted by the letters **A** (ade**n**ine), **G** (Guanine), **C** (cytosine) and **T** (thymine). The sequence of these bases forms the genetic code.

Blastocyst: A 5-6 day old embryo, ready for implantation to occur. The blastocyst consists of a sphere made up of an outer layer of cells (the trophectoderm), a fluid-filled cavity (the blastocoel), and a cluster of cells on the interior (the inner cell mass).

Bone marrow stem cells: Stem cells found in bone marrow that generate bone, cartilage, fat, and fibrous connective tissue.

BRCA 1 and BRCA 2: 'Cancer protection' (tumour suppressor) genes. Mutations in these genes can be inherited and confer a predisposition (increased risk) to breast and ovarian cancer.

CAG/CTG REPEATS: Abbreviation for cytosine-adenine-guanine triplet nucleotide repeat and cytosine-thymine-guanine triplet nucleotide repeats; they are associated with unstable mutations and sometimes show anticipation.

Cancer protection genes: Genes which act as a 'brake' on cell division and prevent uncontrolled cell division/proliferation.

Carcinogen: A physical or chemical agent that causes cancer. It may or may not be a mutagen.

Carrier of a chromosomal rearrangement: This definition applies to an individual who has a rearrangement of his/her chromosomes so that the normal genetic information is present (that is, it is 'balanced') but it is not in the usual 46 chromosome pattern.

Carrier of a mutated gene: Every cell contains two copies of each gene. One gene copy may be mutated and the other may be 'correct'. If the mutated gene is not expressed in the cells (resulting in a particular characteristic or a genetic condition), the mutated gene is said to be recessive to the other 'working' copy of the gene. An individual who has one correct gene copy and one faulty (recessive) gene copy is said to be a 'carrier' for the mutation leading to a specific condition. The carriers of a recessive mutation in a gene are usually not affected but they are at risk for passing on the mutant gene to their offspring.

Carrier screening: Screening populations of people to determine if individuals are genetic carriers of a mutated or faulty gene for a particular genetic condition.

Carrier testing: Testing an individual who is at risk due to a family history to determine if he or she is a carrier of a mutated or faulty gene for a particular genetic condition.

Cell: The basic structural unit of all living organisms. While some organisms are made up of only one or several cells, humans are composed of millions of cells. Each cell is enclosed by a membrane and has a nucleus which contains the genetic material (DNA) in the form of chromosomes. Mitochondria are also found randomly scattered throughout the cell.

Cell culture: A method of encouraging cells to divide and multiply in the laboratory. Culture medium is used to achieve this.

Cell division: The mechanism by which cells multiply during the growth of tissues or organs. The type of cell division involved in the growth of the body is known as mitosis. The cell division which produces sperm or ova in the testis or ovary is known as meiosis.

Centromere: The constricted part of the chromosome that separates it into its two arms. The short arm is called the 'p' arm (for 'petit'); the long arm is called the 'q' arm (because q follows p in the alphabet).

Chimera: A special kind of mosaicism in which an individual or tissue contains a mix of cells derived from two genetically different individuals. The blood of a patient who had a transfusion from another individual would be chimeric for example.

Chorion: The chorion develops into the placenta. Chorionic cells have the same genetic composition as cells of the fetus. Cells of the chorion are sampled during a prenatal diagnostic test called CVS (chorionic villus sampling).

Chorionic villus sampling (CVS) : A procedure for obtaining cells of the chorion to enable testing of the fetus for specific abnormalities. Samples of the cells may be taken through the vagina or through the abdomen of the pregnant mother: it is usually carried out in the 10th - 12th week of pregnancy (see Genetics Fact Sheet 17C: Prenatal Testing – CVS and Amniocentesis).

Chromosome: A threadlike structure found in the nucleus of all the body cells (except red blood cells) consisting of DNA and proteins. Each chromosome can be thought of as a string of beads where every bead represents a gene. (See Genetics Fact Sheet 1: Genes and Chromosomes)

Chromosome translocation: See translocation

Clinical genetics: A specialty of medicine concerned with the diagnosis and discussion of risks of developing a genetic condition in individuals and families.

Clinical geneticist: Doctors with sub-specialty training in clinical genetics. Their role is in diagnosis, management and the provision of genetic counselling and appropriate genetic testing.

Clinical heterogeneity: Refers to the occurrence of clinically different types of genetic conditions due to mutations in the same gene.

Clone: A clone is a cell or group of cells that are identical and are derived from an original source of genetic material.

Cochlea: Small snail shaped organ in the inner ear that transmits sound signals via tiny hairs to the auditory nerve.

Codominance: The equal expression of both copies of a gene in an individual.

Complementary DNA (cDNA) : DNA made in a laboratory from the messenger RNA (mRNA) expressed by a gene. The cDNA is made using an enzyme.

Complex inheritance: Patterns of inheritance that differ from the 'traditional' (Mendelian) patterns of inheritance in that they require multiple factors (either genetic or environmental) for the condition to develop.

Conception: The fusing (joining) of the sperm with the ovum, which leads to the development of an embryo.

Concordance: Presence of the same characteristic in both members of a pair of twins (or set of individuals). The opposite of discordant.

Confined placental mosaicism: Mosaicism that is seen only in the placenta but not in the fetus.

Congenital: Present at birth, not necessarily inherited.

Connective tissue: A general term for all tissues of the body which support and connect various organs and other structures such as the skeleton. Certain types of connective tissue act as a glue, some as scaffolding and others permit expansion and contraction.

Consanguinity: Relationship between two individuals with a common ancestor, for example, first cousins (see Genetics Fact Sheet 16: When Parents are Relatives: Consanguinity).

Consultand: The person seeking, or referred for, genetic counselling.

Cordocentesis: The sampling of fetal blood from the umbilical cord under ultrasound guidance for the purposes of prenatal diagnosis. The procedure is similar to amniocentesis or trans-abdominal CVS (see Genetics Fact Sheet 17C: Prenatal Testing – CVS and Amniocentesis).

Crossing over: When chromosome pairs join together during meiosis, the two chromosomes may exchange material: part of one chromosome 'crosses over' and exchanges places with the corresponding part on its partner chromosome.

Culture medium: The broth that covers cells in a laboratory environment. The culture medium contains nutrients to feed the cells and may encourage them to multiply. Other factors may be added to culture in order to direct desired changes in the cells.

CVS: See Chorionic villus sampling.

Cytogenetics: The microscopic study of chromosomes and how changes in chromosome structure and number affect individuals.

Deletion: The loss of some genetic material. If the deletion is large, it may be observed in the karyotype as missing chromosomal material; if it is small, it may only be detected by analysing the composition of the DNA.

Deoxyribonucleic acid: See DNA.

Diagnostic and statistical manual of mental disorders: A handbook used by mental health professionals to diagnose mental disorders.

Diagnostic test: A term used to describe particular tests that are able to identify (diagnose) a recognised disorder. Prenatal diagnostic tests include; CVS, amniocentesis and some ultrasound imaging.

Differentiate: The process cells undergo whereby they mature and develop the features of a specialised cell with a specific function such as a heart, liver, or muscle cell.

Digestive tract: Also called the gastrointestinal (GI) tract, is the multi-organ system that takes in food, extracts nutrients and expels waste.

Diploid number: This is the number of chromosomes in the somatic (body) cells. There are two copies of each chromosome. In humans, the diploid number is 46.

Discordant: The situation where both members of a pair of twins do not exhibit the same characteristics. Opposite to concordant.

Disomy: Meaning 'two bodies'. Where there are two copies of each chromosome. This is the normal chromosome complement.

Dizygotic twins: Nonidentical twins, arising from two different eggs fertilised by two different sperm; such twins are also referred to as fraternal twins.

DNA (deoxyribonucleic acid): The chemical compound that makes up genes within chromosomes and is the basic material of heredity. It is made up of chemicals called nucleotide bases, linked together in a chain. Two chains of nucleotides twist around each other to form a double helix.

DNA sequencing: Determining the pattern or order in which the nucleotide bases occur in a piece of DNA. This sequence is the genetic code.

Dominant: Every cell contains two copies of each gene. Where only one of the gene copies or allele is mutated, and the other allele is 'correct', but the person is affected by a genetic condition due to that mutation, the mutation is described as dominant. The mutated gene is said to be dominant over the other 'correct' copy of the gene. A condition or characteristic caused by a dominant gene mutation only requires one of the genes to be mutated for the person to be affected.

Duplication: A part of the chromosome is present in two or more copies. If the duplication is large it may be observed under the microscope as a change in a chromosome; a small duplication may only be observed by examining the DNA structure of the chromosome or a gene.

Duodenal atresia: Refers to a blockage in the hollow tube that connects the stomach to the rest of the intestine.

Dysmorphology: Comes from the Greek DYS - meaning abnormal, disease, faulty, impaired and MORPHOLOGY - meaning structure or form. Refers to changes in the usual organisation of a persons cells.

Dysplasia: Abnormal development or growth of tissues or cells.

Egg: The female sex cell (ovum) which carries half the mother's chromosomes (and therefore, half the genes). In humans, this number is normally 23. The egg joins with the sperm at conception to produce an embryo. This process is called fertilisation.

Embryo: The product of the fusion of an egg and a sperm at conception. This term is reserved for the first eight weeks of development.

Embryonic stem cells: Primitive (undifferentiated) cells from the embryo that have the potential to mature and develop into a wide variety of specialised cell types.

Empiric risk: A risk estimate that is given for the chance of occurrence or recurrence of a particular condition in an individual based on the observation of other families with that condition.

Environmental factors: Factors in the environment that may have an effect on our development or growth eg. diet, atmospheric pollutants, cigarette smoke, preservatives, X-rays etc.

Enzyme: A protein molecule which promotes or enables a chemical reaction in the cells (a biochemical reaction) to take place. These biochemical reactions include breaking down food into the essential chemicals required by the body and breaking down toxic by-products of our bodies. Enzymes are essential for the correct function of the body's metabolism.

Enzyme replacement therapy: A method of treating genetic conditions that are due to a deficiency of a particular enzyme. Overcoming the deficiency by providing the body with the enzyme enables the cells to function correctly and the symptoms of the condition may be reduced or eliminated.

Ethics/ethical behaviour: Code of behaviour considered correct; especially that of a particular group, profession or individual.

Eugenics: The practice of trying to influence human heredity by encouraging the transmission of 'desirable' characteristics and discouraging the transmission of 'undesirable' ones.

Exon: The part of the DNA message that is translated into a protein.

Expressed gene: When the coded information contained in the gene is understood by the cells to produce a product such as a protein.

Expressivity: The degree to which an inherited characteristic is expressed in a person. 'Variable expressivity' refers to the variation in expression and severity of particular characteristics of genetic conditions.

Familial: A characteristic or condition that tends to run in families.

Fertilisation: The joining of an egg and sperm at conception to create an embryo.

Fetal blood sampling: A prenatal diagnosis technique where a blood sample is obtained from the fetus.

Fetoscopy: A prenatal diagnosis technique where the fetus and the inside of the uterus (womb) can be directly visualised.

Fetus: In humans, the name given to a product of conception after the end of the eighth week of pregnancy to the moment of birth.

Fibrin: A protein involved in blood clotting.

Fibroblasts: A type of cell found in developing or repairing tissues eg. the skin.

Fragile site: A small break or a constriction of a chromosome that can be visualised after special treatment of the chromosomes. In individuals affected with Fragile X Syndrome, a fragile site can often be seen on their X chromosome (see Genetics Fact Sheet 42: Fragile X Syndrome).

Fraternal twins: See dizygotic twins.

Gamete: Refers to the sperm cells in males and the egg cells in females.

G bands: Banding patterns on chromosomes make it easier to examine the chromosomes under the microscope for abnormalities in structure and/or number. G bands are one type of banding pattern induced to appear on chromosomes by staining them with a special chemical called Giemsa.

Gene: The basic unit of heredity; a segment of DNA that contains the information for a specific characteristic or function.

Gene cloning: Isolating a gene and then making multiple copies of it by inserting it into a bacterial cell or another organism.

Gene mapping: Determining the relative locations of different genes on chromosomes.

Gene therapy: A method of treating genetic conditions by inserting a correct copy of the gene in question into the cells of individuals who have the mutated gene (see Genetics Fact Sheet 27: Gene Therapy).

Genetic carrier: See Carrier of a mutated gene

Genetic code: The information contained in the DNA which is 'interpreted' by the cells to produce proteins. The chemicals (nucleotides) which make up the DNA can be described by the letters **A** (Adenine), **T** (Thymine), **C** (Cytosine) and **G** (Guanine). Thus the genetic code can be written as a series of letters (for example **AAA CGT TTC**).

Genetic condition: A genetic condition is caused by a change in the genetic information. Genetic conditions may be caused by a mutation in a single gene or may be caused by a change in chromosome structure or number (see Genetics Fact Sheet 2: Genetic Conditions - Overview).

Genetic counselling: The provision of diagnosis, information and support by health professionals with specialised training in genetics and counselling (see Genetics Fact Sheet 3: Genetic Counselling).

Genetic counsellor: A health professional with specialised training in genetics and counselling who can provide information and support to individuals or families with concerns about a genetic condition which may run in their family.

Genetic engineering: Laboratory techniques used to alter or manipulate the genetic makeup of cells by deliberately removing, changing or inserting individual genes.

Genetic heterogeneity: Different mutations in a gene causing the same genetic condition.

Genetic mapping: Determination of the relative positions of genes on a chromosome and a measure of the distance between them.

Genetic predisposition: Having some genetic factor(s) that may make an individual more likely to develop a particular condition than the general population.

Genome: The complete set of genes carried by an individual or a cell.

Germ cells: The cells of the body that are used in reproduction (egg and sperm).

Germline: The family of cells that divide to produce new germ cells.

Germline mosaicism: When the germ cells (sperm or egg cells) have a different genetic makeup to the cells in the rest of the body.

Gonadal mosaicism: See germline mosaicism

Haemostasis: Refers to the process of stopping bleeding. There are several different steps in this process including coagulation.

Haploid number: This is the number of chromosomes in the sex cells (sperm or egg). There is one copy of each chromosome. In humans, the haploid number is 23.

Hereditary: The transfer of a gene from parent to child. In mothers, the gene is transferred via the DNA in the egg and in fathers the gene is transferred via the DNA of the sperm.

Homozygote: Refers to an individual in whom the two alleles or gene copies contain identical information. An individual can be homozygous for the correct copies of the gene or can be homozygous for the mutated copies of the gene.

Hormone: A chemical product of the body that has a specific regulatory effect upon the cells.

Identical twins: Twins that arise from a single egg fertilised by a single sperm. These twins are therefore genetically identical. They are also referred to as monozygotic twins.

Immune system: The body's defence system that destroys potentially harmful foreign substances or organisms that enter the body. It is made up of specialised cells.

Implantation: The process whereby a fertilised egg (embryo) is embedded in the lining of a woman's uterus.

Imprinting: The 'memory' held by a chromosome as to whether it was inherited from the mother or the father. The memory is chemically 'stamped' into the DNA and can result in chromosomes or the genes located on the chromosomes behaving differently, depending on the parent of origin.

Inborn error of metabolism: A congenital condition which results from a change in a gene which causes a deficiency in the presence or activity of particular enzymes important for the functioning of the body's metabolism (see Genetics Fact Sheet 20: Newborn Screening for Genetic Conditions).

Incidence: The number of new cases of a condition detected annually, per unit of the population. For genetic conditions, the incidence is quoted as the number of affected individuals per 1,000 births whether detected at birth or not.

Incomplete penetrance: See penetrance

Inherited: The transmission of genetic information from a parent to a child.

Insertion: The addition of a piece of chromosomal material into a chromosome in a place where it is not normally found. This may result in a genetic condition, because the genetic code may then be read or translated incorrectly.

Intron: The part of the genetic sequence that is not translated into the final gene product or message.

Inversion: Where there are two breaks in a chromosome, the segment may flip over and rejoin, that is, become inverted. This results in the genes being in the reverse order along the chromosome. This may cause the genetic code to be read or translated incorrectly.

***In vitro* fertilisation (IVF)**: The process whereby an egg is fertilised with sperm outside of the body and the resulting embryo is transplanted into a woman's uterus.

Isochromosome: A chromosome in which the arms ('p' and 'q') are of equal length and the information in each of the two arms is genetically identical.

IVF: See *In vitro* Fertilisation.

Karyotype: The term used to describe an individual's chromosomes that have been photographed through a microscope and then arranged according to a standard classification based on their group and size. This is done by a specialised scientist trained in cytogenetics.

Kb: A segment of DNA, which is 1,000 base (nucleotide) pairs in length.

Linkage: The tendency for genes or segments of DNA that are located close together on the same chromosome to be inherited together.

Locus: The position on a chromosome of a segment of a gene.

Lysosome: A small body that is found in most cells, but is particularly frequent in the cells of the liver and kidney. They contain important enzymes.

Marker chromosome: A chromosome, or part of a chromosome, usually small, of unknown origin.

Maternal serum testing: A test which assesses the risk of fetal abnormalities such as neural tube defects and Down syndrome by analysing a number of hormones in the mother's blood during pregnancy (see Genetics Fact Sheet 17B: Prenatal Testing – 1st and 2nd Trimester Screening).

Meiosis: The special cell division which only takes place in the sex cells of females and males, resulting in egg and sperm cells that contain 23 chromosomes (the haploid number).

Mendelian inheritance: This refers to the inheritance of single genes and follows specific patterns: autosomal recessive, autosomal dominant and X-linked inheritance (see Genetics Fact Sheet 8: Autosomal Recessive Inheritance, Genetics Fact Sheet 9: Autosomal Dominant Inheritance and Genetics Fact Sheet 10: X-Linked Inheritance).

Metabolism: The physical and chemical processes by which energy is made available for essential body functioning, growth and development.

Metacentric: Refers to a chromosome which has its centromere in the middle and the short (p) and long (q) arms are of equal length.

Miscarriage: Loss of a baby before the twentieth week of pregnancy.

Mitochondria: These structures or organelles in the cell are the main energy source: they are often called the powerhouse of the cell. The mitochondria also contain their own DNA and therefore genes; mitochondrial genes follow maternal inheritance (see Genetics Fact Sheet 12: Mitochondrial Inheritance – Complex Patterns of Inheritance 2).

Mitochondrial DNA: The genetic material contained in the circular genome found in mitochondria.

Mitosis: The process of cell division in somatic cells, not the sex cells. Mitosis results in 'daughter' cells which are genetically identical to the parent cells.

Molecular genetics: The branch of genetics that studies the function and structure of genes at the molecular level.

Monoclonal: A group of cells that are all identical copies of an original cell.

Monogenic: A characteristic which is due to the information contained in a single gene.

Monosomy: Where a particular chromosome appears only once instead of twice in cells. For example, girls with Turner syndrome have only one X chromosome instead of the usual two copies (Monosomy X).

Monozygotic twins: See identical twins.

Mosaic: A situation where some cells have an abnormal or unusual genetic or chromosomal makeup while the rest of the cells in the body have the usual genetic or chromosomal constitution. For example, a person who is mosaic for trisomy 21 would have some cells which have 47 chromosomes with an extra chromosome number 21 and other body cells which have the usual 46 chromosome complement. The number of cells with abnormal genetic or chromosomal content will determine the level of severity of the condition.

mRNA: An abbreviation for messenger RNA which is the chemical that transfers the genetic DNA message to the ribosomes where it is translated into proteins.

Mucopolysaccharide: Important substances in the body which are composed of proteins and complex sugars called polysaccharides.

Multifactorial inheritance: A pattern of inheritance which results from the interaction of one or more genes with environmental factor(s) (see Genetics Fact Sheet 11: Environmental and Genetic Interactions – Complex Patterns of Inheritance 1).

Mutagen: A physical or chemical agent that causes a permanent change (mutation) in a gene. It may or may not be a carcinogen.

Mutation: A permanent change in a gene. If the mutation occurs in the egg or sperm (sex cells), it can then be inherited. Mutations in somatic cells cannot be inherited. Mutations can occur naturally and spontaneously or they may be due to exposure to mutagens.

Neural tube: The embryonic structure which forms into nervous system including the spinal cord and brain.

Neural tube defect (NTD): An abnormality which results when the neural tube in the fetus fails to close. Spina bifida and anencephaly are forms of NTD (see Genetics Fact Sheet 59: Neural Tube Defects - Spina Bifida and Anencephaly).

Neuro-developmental: The process of development of the brain and other parts of the central nervous system.

Neurones: Nerve cells, the structural and functional unit of the nervous system. A neuron transmits electrical signals around the body through a network of nerve cells.

Nondisjunction: Where the chromosome pairs fail to separate correctly in meiosis, resulting in sperm or egg cells which have missing or extra chromosomes eg. if chromosome number 21 fails to separate in the formation of an egg (or sperm), one egg (or sperm) will contain an extra copy of chromosome 21 (24 chromosomes instead of 23) while the other egg (or sperm) will contain only 22 chromosomes (see Genetics Fact Sheet 6: Changes to Chromosomes – Number, Size and Structure).

Non-syndromic: A condition not related to a recognised pattern of characteristics and/or symptoms (syndrome)

Nucleotides: Also referred to as bases, they are the basic components of DNA. The nucleotides are denoted by the letters **A** (adenine), **G** (Guanine), **C** (cytosine) and **T** (thymine).

The sequence of these nucleotides forms the genetic code.

Nucleus: The structure in a cell which contains the chromosomes.

Obligate mutation carrier: A family member who has no recognisable symptoms of a genetic condition but on the basis of their pedigree (family health tree), must be a carrier of a mutated gene for that condition.

Oncogene: A gene which, when triggered, can lead to cancer (see proto-oncogene).

Organelle: Structures within cells such as the nucleus, mitochondria and lysosomes which have specific functions.

Ovum: The female reproductive cell or egg which contains 23 chromosomes.

'p' arm: Each chromosome is divided into two parts, joined by the centromere. The 'p' arm is the shorter of the two segments and is at the top of the chromosome. The longer segment is called the 'q' arm.

Pedigree: A diagrammatic representation of a family health history or family health tree.

Penetrance: The probability of detecting the presence or clinical expression of a gene or combination of genes when they are present. If the penetrance of a particular gene is less than 100%, not all individuals who carry a mutation in the gene will develop symptoms of the condition it causes. Such a genetic condition is said to have reduced or incomplete penetrance.

Phenotype: The physical and/or biochemical characteristics of a person, an animal or other organism which are determined by their genetic makeup and/or environment.

Placenta: The structure that provides the fetus with nourishment during its development prior to birth. It is attached to the wall of the uterus and connects to the fetus through the umbilical cord.

Pluripotent: Cells that may still differentiate into various types of specialised tissue in the body.

Polygenic: A condition or characteristic that is caused by many different genes acting together.

Polymorphisms: Changed genes, DNA sequences or chromosome structures which occur naturally in the population and do not cause any harm to the individual.

Post-lingual: After the development of language (speech).

Predictive testing: A form of genetic testing performed on a person with a family history of a particular genetic condition, but who does not have any symptoms of the condition at the time of testing. This testing determines whether that person has inherited the mutation (present in their family). If testing for this mutation reveals that it is present in the person, then they have an increased risk to developing the condition that was tested for. The detection of a specific mutation does not necessarily mean the individual will definitely develop the condition. Familial breast cancer is an example of a condition where predictive testing is used (see Genetics Fact Sheet 48: Breast and Ovarian Cancer and Inherited Predisposition – Cancer Genetics 2).

Predisposition: A situation in which a person, due to their inherited genetic makeup, may have a particular susceptibility to a condition if exposed to the correct environmental triggers.

Preimplantation genetic diagnosis (PGD): An adjunct to the IVF process where the embryo undergoes genetic testing before it is transferred (implanted) into the uterus.

Pre-lingual: Before the development of language (speech).

Prenatal diagnosis: The detection of fetal abnormalities during pregnancy (see Genetics Fact Sheet 17: Prenatal Testing – An Overview).

Prenatal screening: Tests during pregnancy to assess the possibility that a fetus is affected with a particular condition. These tests are not 100% accurate.

Presymptomatic testing: A form of genetic testing performed on a person with a family history of a particular genetic condition, but who does not have any symptoms of the condition at the time of testing. This testing determines whether that person has inherited the mutation (present in their family). If testing for this mutation reveals that it is present in the person, then they will most likely develop symptoms of the genetic condition it causes at some stage of their life. Huntington disease is an example of a genetic condition where presymptomatic testing is used (see Genetics Fact Sheet 44: Huntington Disease – Neurological Conditions 1).

Prevalence: The proportion of a whole population affected by a certain condition.

Probe: A small segment of DNA of known origin, manufactured in the laboratory, which is designed to recognise the DNA on specific parts of chromosomes. A coloured stain can be attached to the probe and used to confirm the presence or absence of a particular gene or mutation.

Prophylactic surgery: A surgical procedure designed to prevent the development of a disease. For example, prophylactic mastectomy is the removal of one or both breasts in an effort to prevent breast cancer in individuals who carry a mutation in a breast cancer gene and are therefore at high risk of developing breast cancer.

Prostate: A small gland of the male reproductive system that secretes a fluid that makes up part of the ejaculate (seminal fluid).

Protein: Substances which are major components of the body structure, essential to body function. They are made up of smaller units called amino acids.

Proto-oncogene: Genes that are part of a person's usual genetic makeup. They have a role in various aspects of cell division. If these genes are changed in some way, they may give rise to oncogenes that can lead to cancer.

'q' arm: Each chromosome has two segments joined by the centromere. The 'q' arm is the longer of these two segments. The shorter segment is called the 'p' arm.

Recessive: Every somatic cell in our body contains two copies of each gene. Each gene contains the information for a particular gene product, such as a protein. If a gene is mutated, the gene no longer codes for the gene product. Where an individual has one gene copy or allele mutated and the other copy 'working', the cell will only be producing half the amount of gene product. If this does not result in a genetic condition in the individual, the mutation is described as being hidden or 'recessive' to the correct copy of the gene. An individual with this genetic constitution is said to be a 'carrier' of a recessive gene mutation. For a recessive gene mutation to result in a particular characteristic or a genetic condition, both copies of the genes must be mutated (see Genetics Fact Sheet 8: Autosomal Recessive Inheritance – Traditional Patterns of Inheritance 1).

Recurrence risk: The risk that a genetic condition will occur again in a family.

Regulatory gene: A gene containing information for the regulation (switching on or off) of other genes.

Replication: The identical duplication of DNA or a cell.

Restriction enzyme: Enzymes that can cut DNA into strands at specific places along its length.

Ribosomes: Small components in a cell that are composed of ribosomal RNA. They are important in the reading of the DNA messages in a cell. See also mRNA.

RFLPs (restriction fragment length polymorphisms): The fragments of DNA that are formed when DNA is cut by special enzymes called restriction enzymes. The patterns of these fragment lengths are used to indicate the presence or absence of mutations in particular genes.

Ribonucleic acid: See RNA.

Ring chromosome: This occurs as a result of the fusion of the two ends of the same chromosome; there is a consequent loss of genetic material.

RNA (ribonucleic acid): An abbreviation for ribonucleic acid, a chemical similar to DNA which has an important role in protein manufacture. There are several types of RNA (see mRNA).

Robertsonian translocation: A type of translocation exclusive to the acrocentric chromosomes (13, 14, 15, 21 and 22) in which two of these chromosomes join at or near their centromeres. This is effectively a fusion between two whole chromosomes.

Satellites: Small, round appendages attached by fine stalks to the ends of the short 'p' arms of the acrocentric chromosomes. They do not always stain darkly and may be difficult to see in a karyotype.

Sensitivity: The ability of a test to detect the presence of a genetic condition or a mutation when it is truly present.

Sex cells: The reproductive cells (sperm or egg) which are the result of meiosis.

Sex chromosome: An X or a Y chromosome which are different from the 22 autosomes.

Sex influenced: A genetic condition or characteristic whose expression or severity differs between the sexes. That is, it occurs more frequently in either males or females.

Sex limited: A genetic condition or characteristic that is expressed in only one sex. That is, it occurs only in males or in females.

Sex linked: A genetic condition or characteristic which is determined by genes carried on the X or Y chromosomes.

Somatic cells: All the cells of the body except the reproductive cells (sex cells).

Somatic gene therapy: Correcting or transplanting genes that are present in somatic cells, not the sex cells.

Somatic mutation: A change or fault in a gene which is found in the cells of the body but not in the sex cells. Somatic mutations cannot therefore be passed on to future generations.

Somatic stem cells: Another name for adult stem cells.

Specificity: The ability of a test to determine whether a genetic condition or mutation is absent when it is truly absent.

Sperm (abbreviation of spermatozoan): The male sex cell which contains 23 chromosomes. It fuses with the egg during fertilisation.

Sporadic: A mutation that results in a genetic condition and which appears for the first time in a family. The mutation takes place in either the egg or the sperm or at conception.

Stem cells: Undifferentiated cells with the ability to divide for indefinite periods in cell culture and to give rise to specialised, functional cells.

Stereotyped behaviour: Repetitive routines or movements that have no function or use.

Storage disease: Any disease or condition which is characterised by the abnormal accumulation and storage of material within the cells. The stored material will vary depending upon the type of condition.

Syndrome: A group of characteristics and/or symptoms that occur together in a recognisable pattern.

Synthesis: The process of building a complex compound from a number of simple chemical elements.

Telomere: The terminal or end segment of each chromosome arm.

Teratogen: A substance that produces or increases the incidence of birth defects or congenital abnormalities by interfering with development of the fetus during pregnancy.

Termination of pregnancy: Intervention to ensure a pregnancy does not continue.

Tetraploidy: Four copies of every chromosome resulting in 92 chromosomes in a cell instead of the usual 46.

Tetrasomy: Four copies of a particular chromosome present in a cell, resulting in 48 chromosomes in the cell instead of the usual 46.

Thromboembolism: A clot in a blood vessel obstructing the flow of blood.

Thrombophilia: An inherited or acquired predisposition to excessive blood clotting.

Thrombus: A blood clot.

Transgenic mouse: A mouse which has been genetically altered by injecting human or other foreign DNA from another animal into fertilised mouse eggs. This DNA becomes incorporated into the mouse DNA and the mouse will translate the information contained in the foreign gene. This has become a useful model for the study of various genetic conditions.

Translocation: This occurs when a piece of one chromosome breaks off and attaches to another different chromosome. When no material is lost or gained the translocation is said to be 'balanced' and the individual may or may not be affected by it. An 'unbalanced' translocation results in the loss or gain of genetic material which may result in a genetic condition.

Trinucleotide repeat: See Triplet Repeat.

Triplet: A sequence of three nucleotides in the DNA sequence. Each triplet represents the code for a particular amino acid.

Triplet repeat (trinucleotide repeat): A form of genetic mutation which consists of a series of repeated identical sequences of DNA triplets which may be found either inside or outside a gene. An increase in the number of such repeats in a particular gene can lead to instability of the gene and manifestation of the corresponding genetic condition. This form of genetic mutation has been associated with a number of genetic conditions including Huntington disease, fragile X syndrome and myotonic dystrophy.

Triploidy: Having three copies of every chromosome resulting in 69 chromosomes in a cell instead of the usual 46.

Trisomy: Three copies of a particular chromosome are present in a cell resulting in 47 chromosomes instead of the usual 46.

Tumour suppressor gene: A gene which contains the information for proteins whose role in cells is to suppress the formation of tumours by controlling cell division and growth. Loss of this control leads to development of malignancy.

Ultrasound: The use of sound waves for visualising body tissues and structures. In pregnancy, structural abnormalities in the fetus can be detected (see Genetics Fact Sheet 17A: Prenatal Testing - Ultrasound).

Undifferentiated: A cell that has not matured into a functional, specialised cell type.

Uniparental disomy: Where both members of a chromosome pair are contributed by one parent rather than one from each parent. Uniparental disomy may be maternal or paternal in origin.

Uterus (womb): The female reproductive organ in which the fetus develops into a baby over a period of nine months.

X-linked gene: Any gene that is located on the X chromosome.

X-linked recessive mutation: A recessive mutation in a gene carried on the X chromosome.

X-linked dominant mutation: A dominant mutation in a gene carried on the X chromosome.

Zygote: The single cell with 46 chromosomes resulting from the fertilisation of an egg (23 chromosomes) by a sperm (23 chromosomes). Through cell division (mitosis), the zygote develops into a multicellular embryo and then into a fetus.