GLOSSARY

Assisted reproduction The use of clinical and laboratory techniques to increase chances of conceiving a baby. An example is ***in vitro* fertilisation**, or **IVF**.

Asymptomatic Having no signs or symptoms of disease.

Alzheimer's disease A degenerative brain disease of unknown cause that is the most common form of dementia, that usually starts in late middle age or in old age as a memory loss for recent events spreading to memories for more distant events and progressing over the course of five to ten years to a profound intellectual decline characterized by dementia and personal helplessness, and that is marked histologically by the degeneration of brain neurons especially in the cerebral cortex and by the presence of neurofibrillary tangles and plaques containing beta-amyloid.*

Carrier Someone who carries only one copy of a **mutant gene** in question. A carrier usually shows no symptoms or very mild symptoms for the disease gene that he or she carries, as two copies of the disease gene are required for a full-blown manifestation of the disease. A carrier has the risk of transmitting the mutant gene to the next generation.

Chromosome Structure in a cell that contains DNA and proteins. With the exception of sperm and egg cells and red blood cells, each human cell with a nucleus contains two sets of chromosomes, one inherited from the mother and one from the father. Each set consists of 23 chromosomes, 22 autosomes (non-sex chromosomes) and one sex chromosome, either X or Y. These human cells thus contain 46 chromosomes and are termed diploid. A male diploid cell has an X and a Y chromosome, whereas a female diploid cell contains two X chromosomes. Sperm and egg cells are haploid and contain only 23 chromosomes. Each chromosome contains genes arranged linearly, and is made up of proteins and **DNA**.

Clinical validity The accuracy with which a test determines the presence or absence of a clinical condition or which a test predicts a predisposition.

Congenital Existing at or dating from birth.

Cystic fibrosis Cystic fibrosis (CF) is an inherited disease characterized by an abnormality in the body's salt, water- and mucus-making cells. It is chronic, progressive, and is usually fatal. In general, children with

CF live into their 30s. Children with CF have an abnormality in the function of a cell protein called the cystic fibrosis transmembrane regulator (CFTR). CFTR controls the flow of water and certain salts in and out of the body's cells. As the movement of salt and water in and out of cells is altered, mucus becomes thickened. The thickened mucus can affect many organs and body systems including:

- respiratory - sinuses and lungs
- digestive - pancreas, liver, gallbladder, intestines
- reproductive - more so in the male, where sperm-carrying ducts become clogged
- sweat glands *

Diagnostic chain The chain of events or procedures that begins from the collection of sample and ends with a diagnosis based on analyses of the sample.

DNA DNA, or deoxyribonucleic acid, is the hereditary material in humans and almost all other organisms. Each DNA is a linear molecule made up of nucleotides or bases. There are four different types of bases in DNA and the order in which these bases are arranged determines the protein to be formed.

Each individual's body contains an identical set of DNA in nearly all of its cells. A great fraction of cellular DNA is located in the cell nucleus (where it is called nuclear DNA), while the remaining can be found in the mitochondria (where it is called mitochondrial DNA).

Down's syndrome A congenital condition characterized by moderate to severe mental retardation, slanting eyes, a broad short skull, broad hands with short fingers, and by trisomy of the human chromosome numbered 21.*

Duchenne muscular dystrophy A severe progressive form of **muscular dystrophy** of males that appears in early childhood, affects the muscles of the legs before those of the arms and the proximal muscles of the limbs before the distal ones, is inherited as an X-linked recessive trait, is characterized by complete absence of the protein dystrophin, and usually has a fatal outcome by age 20.*

Early-onset The early manifestation or occurrence of a disease normally characterised by delayed development. For example, Alzheimer's disease usually occurs in late middle-age years or old age, but early-onset Alzheimer's disease may occur in early middle-age years.

Familial adenomatous polyposis	A disease of the large intestine that is marked by the formation especially in the colon and rectum of numerous adenomatous polyps which typically become malignant if left untreated, that may be either asymptomatic or accompanied by diarrhoea or bleeding, and that is inherited as an autosomal dominant trait.*
Gene	A gene is the basic physical and functional unit of heredity. It is made up of DNA which carries instructions to make molecules of **RNA** and proteins. Every person has two copies of each gene, one inherited from each parent. Most genes are commonly found in all people, but about one percent of each person's **genome** is slightly different from that of another. The slight difference is what makes people physically unique.
Gene therapy	Treatment of a genetic disorder by inserting functional genes in order to replace, supplement, or manipulate the expression of non-functional or abnormal genes. Gene therapy has thus far only advanced into clinical trials and is not yet an established therapy.
Genetic variant	Genetic variance is the differences in **phenotypes** and **genotypes** in a population.
Genome	The complete set of genetic instructions for making an organism is called its genome. The genome contains the master blueprint for all cellular structures and activities for the lifetime of the cell or organism. Found in every nucleus of a person's many trillions of cells, the human genome consists of tightly coiled threads of DNA and associated protein molecules, organised into structures called chromosomes.
Genotype	A specific set of alleles (variant forms of a gene) at particular position on the chromosome.
Germ cell (Germline)	The cell (or cell line) from which sperm and egg (gametes) are derived.
Glucose-6-phosphate dehydrogenase deficiency	A hereditary metabolic disorder affecting red blood cells that is controlled by a variable gene on the X chromosome, that is characterized by a deficiency of glucose-6-phosphate dehydrogenase conferring marked susceptibility to haemolytic anaemia which may be chronic, episodic, or induced by certain foods (as broad beans) or drugs (as primaquine), and that occurs especially in individuals of Mediterranean or African descent.*
Haemoglobin	The substance inside red blood cells which binds oxygen molecules and transport them from the lungs to other tissues.

Haemophilia	A sex-linked hereditary blood defect that occurs almost exclusively in males and is characterized by delayed clotting of the blood and consequent difficulty in controlling haemorrhage even after minor injuries.*
Huntington's disease	A progressive chorea that is inherited as an autosomal dominant trait, that usually begins in middle age, that is characterized by choreiform movements and mental deterioration leading to dementia, and that is accompanied by atrophy of the caudate nucleus and the loss of certain brain cells with a decrease in the level of several neurotransmitters.*
Hypothyroidism	Deficient activity of the thyroid gland; *also*: a resultant bodily condition characterized by lowered metabolic rate and general loss of vigour.*
Immunogenetic status	The genetic makeup of the immune system of an individual.
Institutional Review Board (IRB)	A committee appointed by an institution to review the ethical standards of biomedical research proposals.
***In vitro* fertilisation (IVF)**	A clinical and laboratory procedure whereby the eggs and sperms from a couple are extracted and fertilised outside their bodies. Such a procedure is a kind of assisted reproduction aimed at increasing the chances of a couple conceiving a baby.
Jaundice	A yellowish pigmentation of the skin, tissues, and certain body fluids caused by the deposition of bile pigments that follows interference with normal production and discharge of bile (as in certain liver diseases) or excessive breakdown of red blood cells (as after internal haemorrhage or in various haemolytic states).*
Karyotype	The chromosomes of a cell can be stained by a dye to become observable under the microscope and to display characteristic banding patterns. The analysis of a set of chromosomes arranged in corresponding sizes and banding patterns is called a karyotype.
Late-onset	The development of a hereditary disorder beginning only in late childhood or adulthood.
Metabolite	A product of biochemical processes in a cell or organism.
Muscular dystrophy	Any of a group of hereditary diseases characterized by progressive wasting of muscles.*

Mutation A gene mutation is a permanent change in the DNA sequence that makes up a gene. It ranges in size from one DNA base to a large segment of a chromosome.

Gene mutations can be inherited from a parent or acquired during a person's lifetime. If a mutation occurs in an egg or sperm cell during a person's life, there is a chance that the person's children will inherit the mutation.

Most mutations do not cause genetic disorders. For example, some mutations alter a gene's DNA base sequence but don't change the function of the protein made by the gene.

Neonatal Of, relating to, or affecting the newborn and especially the human infant during the first month after birth.

Phenotype The observable characteristics of the expression of a gene.

Preimplantation genetic diagnosis (PGD) A procedure whereby early embryos created by IVF are evaluated to determine the presence of one or more genetic conditions. It is then followed by the selection and implantation of unaffected embryos into the uterus.

Preimplantation tissue typing A procedure whereby early embryos created by IVF are tested for tissue compatibility with an existing sibling. This is then followed by the selection and implantation of tissue compatible embryos into the uterus with the aim of bringing about the birth of a child who can provide a matched tissue donation. It can be used as the sole clinical objective or in combination with PGD to avoid a serious genetic condition in the resulting child.

Prenatal genetic diagnosis Tests performed during pregnancy to determine if a foetus is affected with a particular genetic disorder.

Presymptomatic testing Testing of an asymptomatic individual to determine if the individual has inherited a defect in a specific gene for a late-onset disease which confers on him or her an almost 100% risk of developing the disease at a later stage in life.

Protein Large and complex molecules that play many critical roles in the body. They do most of the work in cells and are required for the structure, function and regulation of the body's tissues and organs.

RNA RNA, or ribonucleic acid, is mainly involved in the translation of genetic information coded in DNA to make protein molecules in the cell.

Scientific validity	The reliability of a test performed in the laboratory. A validated test should consistently detect the presence of its gene substrate and should consistently show negative results in the absence of its gene substrate.
Sex-linked	A disease gene that is situated on either the X or Y chromosome is said to be sex-linked. An X-linked disease, for example, is caused by a genetic defect in the X chromosome.
Sickle-cell anaemia	A chronic anaemia that occurs primarily in individuals of African descent who are homozygous for the gene controlling haemoglobin S and that is characterized by destruction of red blood cells and by episodic blocking of blood vessels by the adherence of sickle cells to the vascular endothelium which causes the serious complications of the disease (as organ failure).*
Somatic cell	All the body cells except the reproductive (germ) cells.
Susceptibility (Predisposition) testing	Testing of an asymptomatic individual to determine if the individual has inherited a genetic variant or variants, which may increase his or her risk of developing a multi-factorial disease such as Alzheimer's disease, diabetes and certain cancers, some time in the future.
Thalassaemia	Any of a group of inherited hypochromic anaemias and especially Cooley's anaemia controlled by a series of allelic genes that cause reduction in or failure of synthesis of one of the globin chains making up haemoglobin and that tend to occur especially in individuals of Mediterranean, African, or southeastern Asian ancestry – sometimes used with a prefix (as alpha-, beta-, or delta-) to indicate the haemoglobin chain affected; called also *Mediterranean anaemia*.*
X-linked	See *sex-linked*.

** From Merriam-Webster Medical Dictionary*