Summary

- Gene therapy is an experimental form of treatment. It works by replacing a faulty disease-causing gene with a working version, or by introducing a new gene to cure a condition or modify its effects.
- The aim is to eliminate genetic diseases at their source.
- The challenge for nations experimenting with gene therapy is to come up with workable, fair and ethical guidelines for its use.

This type of therapy is called 'therapeutic gene therapy' or 'the use of genes as medicine'. It is an experimental form of treatment that is still being developed, but it has the potential to revolutionise treatment for all kinds of genetic conditions.

Gene therapy targets the faulty genes responsible for genetic diseases. Inheriting a faulty (mutated) gene can directly cause a wide range of disorders such as cystic fibrosis and haemophilia. It can also cause susceptibility to some cancers. Gene therapy can be used to replace a faulty gene with a healthy version or to introduce a new gene that can cure a condition or modify its effects.

Inheriting one or both copies of a faulty gene can cause a wide range of conditions such as haemophilia and cystic fibrosis, and can also result in increased susceptibility to some cancers. Gene therapy targets the faulty genes responsible for a genetic condition. Gene therapy can be used to replace a faulty gene copy with a working version or to introduce a new gene that can cure a condition or modify its effects.

The gene therapy process

The basic steps of gene therapy include:

- The faulty gene that causes a specific condition must be identified.
- The location of the affected cells in the body’s tissues or organs must be pinpointed.
- A working version of the gene must be available.
- The working version of the gene has to be delivered to the cell.

A range of delivery techniques

The current problem is to find a way to successfully ‘deliver’ the working version of the gene. To begin with, the affected cells are taken from the person’s body and the working version of the gene is either ‘spliced’ or injected into these cells. They are left to grow in the laboratory and then replaced into the person.

One promising technique is to put the working gene inside a harmless virus, which has had most of its own genes removed – it has been ‘deactivated’. A virus that causes disease (such as the common cold) works by slipping into a cell, taking over its DNA and forcing it to produce more viruses. Similarly, a deactivated virus can enter the specific cell and deliver the working gene.

Other techniques involve using stem cells. These are immature cells that have the potential to develop into cells with different functions. In this technique, stem cells are manipulated in the laboratory to accept new genes that can then change their behaviour. For example, a gene might be inserted into a stem cell that could make it better able to survive chemotherapy. This would be of assistance to those patients who could benefit from further chemotherapy following stem cell transplantation.

Some examples of gene therapy

- **Leber’s congenital amaurosis (LCA):** In February 2007, a gene therapy trial was conducted in the NIHR Biomedical Research Centre in the US with three patients (about 18 years old) with a condition called Leber’s congenital amaurosis (LCA), a rare inherited eye disease. The condition appears at birth or in the first few months of life and causes progressive deterioration and loss of vision. There are currently no effective treatments available. The trial’s purpose was firstly to find out whether gene therapy for retinal disease is safe, and secondly, to find out if it can benefit vision in young adults who already have advanced retinal disease. The cells beneath the retinas of the patients were inserted, using a very fine needle, with the modified virus in a controlled retinal detachment that resolved...
Gene therapy

Things to remember

Where to get help

Ethics, morals and genetic engineering

More research is needed

Body cells versus reproductive cells

The risks of gene therapy

Adenosine deaminase deficiency: A person born with adenosine deaminase (ADA) deficiency lacks an important enzyme of their immune system. This means that infections are likely and can even be fatal. ADA deficiency was the first genetic disorder to undergo experimental gene therapy trials in 1990. It was chosen because a single, relatively uncomplicated gene causes it. The results were promising.

Bolstering the immune system: Current research is focusing on the immune system, which is a collection of special cells and chemicals that fight infection. If the immune system isn’t functioning in the right way, illness can result. One theory on cancer suggests that the immune system is failing to stop the overgrowth of cells that form a tumour. If the immune system could be ‘bolstered’ with gene therapy, perhaps the body would be able to prevent the spread of cancer by itself. One day, gene therapy may also be used as a form of immunisation against particular infections, such as HIV/AIDS and malaria.

X-SCID: Children affected by X-linked severe combined immune deficiency (X-SCID) have a faulty gene that means they have no working immune system, so their bodies cannot fight infections. Only boys are affected due to the pattern of inheritance of the faulty gene. Until recently, boys with X-SCID faced a lifetime living in a sterile bubble, unless they could be given a matched bone marrow transplant.

With gene therapy, bone marrow from the boy is first removed to ‘harvest’ stem cells. The stem cells are then infected with a virus carrying a working copy of the X-SCID gene, before returning the cells to the boy’s body. This treatment was described in 2000. Seven out of 10 infants treated to date have restored immune function, but two of the children treated initially have developed a form of leukaemia. The leukaemia in these two patients was caused when the virus used to deliver the therapeutic gene activated a cancer-causing gene.

After the first boy developed leukaemia in October 2002 and the second in January 2003, clinical trials of the gene therapy being conducted in a number of countries were halted. These have now been resumed, but only for patients with no other treatment options. Work is continuing to make the therapy as safe as possible.

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References
- Gene therapy [online], The Gene Therapy Research Unit, The Children's Hospital, Westmead, Australia. More information here.
- Gene Therapy [online], The Gene Therapy Research Unit, The Children's Hospital, Westmead, Australia. More information here.
- Gene Therapy [online], The Gene School, ThinkQuest International Library. More information here.
More information

Genes and genetics

The following content is displayed as Tabs. Once you have activated a link navigate to the end of the list to view its associated content. The activated link is defined as Active Tab

- A-Z of genetic conditions
- Genes and genetics explained
- Genetic testing

A-Z of genetic conditions

- Ambiguous genitalia
  
  The causes of ambiguous genitalia include genetic variations, hormonal imbalances and malformations of the fetal tissues that are supposed to evolve into genitals.

- Angelman syndrome
  
  The characteristic features of Angelman syndrome are not always obvious at birth, but develop during childhood.

- Ankylosing spondylitis
  
  Ankylosing spondylitis (AS) is a type of inflammatory arthritis that targets the joints of the spine.

- Barrett's oesophagus
  
  Symptoms of Barrett's oesophagus are similar to regular heartburn, which means many people don't seek treatment until their condition is quite advanced.

- Bipolar disorder
  
  Bipolar disorder is a type of psychosis, which means the person's perception of reality is altered. It is characterised by extreme mood swings.

- Central nervous system birth defects
  
  Folic acid taken before conception, and during at least the first four weeks of pregnancy, can prevent around seven out of 10 cases of neural tube defects.

- Charcot-Marie-Tooth disease (CMT)
  
  Charcot-Marie-Tooth disease is the most common inherited disorder affecting the peripheral nervous system.

- Cleft palate and cleft lip
  
  Most cleft palates and cleft lips can be repaired so that appearance and speech develop normally.

- Congenital adrenal hyperplasia (CAH)
  
  CAH is a rare genetic disorder, but it is well understood and treatment is readily available.

- Creutzfeldt-Jakob disease (CJD)
  
  Creutzfeldt-Jakob disease is characterised by physical deterioration of the brain, dementia and walking difficulties.

- Cri du chat syndrome
  
  Most children born with cri du chat syndrome have moderate intellectual disability, with varying degrees of speech delay and some health problems.

- Cystic fibrosis (CF)
  
  When a person has cystic fibrosis, their mucus glands secrete very thick sticky mucus that clogs the tiny air passages in the lungs and traps bacteria.

- Digestive tract birth defects

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Too much amniotic fluid surrounding the baby during pregnancy (polyhydramnios) may indicate the presence of defects of the digestive tract.

- **Down syndrome**
  With the support and opportunities available to them today, most people with Down syndrome are able to achieve and participate as valued members of their community.

- **Dwarfism**
  Dwarfism refers to a group of conditions characterised by shorter than normal skeletal growth.

- **Eczema (atopic dermatitis)**
  Eczema can vary in severity, and symptoms may flare up or subside from day to day.

- **Essential tremor**
  Essential tremor causes involuntary shaking or trembling of particular parts of the body, usually the head and hands, but it is not Parkinson's disease.

- **Fragile X syndrome**
  The facts about fragile X syndrome are complicated, and parents and family members are invited to ask their doctor to refer them to a genetics clinic.

- **Friedreich's ataxia**
  To the casual observer, a person with Friedreich ataxia may seem to be drunk.

- **Genetic factors and cholesterol**
  Familial hypercholesterolaemia is an inherited condition characterised by higher than normal levels of blood cholesterol.

- **Haemochromatosis**
  Haemochromatosis (iron overload disorder) tends to be under-diagnosed, partly because its symptoms are similar to those caused by a range of other illnesses.

- **Haemophilia**
  All children with severe haemophilia are given preventative treatment with infusions of blood products before they have a bleed.

- **Hair**
  Human hair grows one centimetre every month.

- **Hearing problems in children**
  The earlier that hearing loss is identified in children, the better for the child's language, learning and overall development.

- **Huntington's disease**
  The symptoms of Huntington's disease usually, but not always, first appear when the person is approaching middle age.

- **Kabuki syndrome**
  Kabuki syndrome affects males and females equally and there is no cure.

- **Kennedy's disease**
  Kennedy's disease is a rare inherited neuromuscular disorder that causes progressive weakening and wasting of the muscles, particularly the arms and legs.

- **Kidneys - medullary cystic kidney disease**
  Medullary cystic kidney disease causes the growth of abnormal cysts in the kidneys.

- **Kidneys - polycystic kidney disease (PKD)**
  Polycystic kidney disease is a common cause of kidney failure in Australia and equally affects men and women of different ethnic backgrounds.

- **Klinefelter syndrome**
  Klinefelter syndrome is often diagnosed at puberty, when the expected physical changes don't occur.

- **Leukoencephalopathy**
  Leukoencephalopathy refers to a group of inherited disorders that affect the white matter of the brain, which causes loss of normal brain functions.

- **Long QT syndrome**
  You should be investigated for long QT syndrome if you faint for no apparent reason, during or after exercise or emotional excitement.

- **Marfan syndrome**
  Some people may not realise they have Marfan syndrome, because their features are either mild or not obvious.
• McCune-Albright syndrome
  The severity of symptoms or how a child with McCune-Albright syndrome will be affected throughout life is difficult to predict.

• Muscular dystrophy
  People affected by muscular dystrophy have different degrees of independence, mobility and carer needs.

• Neurofibromatosis
  Neurofibromatosis is caused by faulty genes, which may be inherited or have spontaneously mutated at conception.

• Noonan syndrome
  Noonan syndrome is a genetic condition that usually includes heart abnormalities and characteristic facial features.

• Osteoporosis in children
  Osteoporosis in children is rare and usually caused by an underlying medical condition.

• Phenylketonuria (PKU)
  PKU is an inherited disorder that prevents the normal breakdown of a protein found in some foods.

• Prader-Willi syndrome
  A feature of Prader-Willi syndrome is the child's excessive appetite, which often leads to obesity.

• Premature and early menopause
  The symptoms of premature or early menopause are the same as for menopause at any age.

• Rett syndrome
  People with Rett syndrome have a keen desire to communicate.

• Spinal muscular atrophy (SMA)
  A child with spinal muscular atrophy type 1 rarely lives beyond three years of age.

• Tay-Sachs disease
  Tay-Sachs disease is a serious genetic disorder common in Ashkenazi Jews and French-Canadians.

• Thalassaemia
  Thalassaemia is an inherited blood disorder that can cause anaemia or death if not treated.

• Tourette syndrome
  Milder forms of Tourette syndrome can be misdiagnosed, as it often occurs at the same time as attention deficit hyperactivity disorder (ADHD) and other disorders.

• Treacher Collins syndrome
  Treacher Collins syndrome is a genetic disorder that affects growth and development of the head, causing facial defects and hearing loss.

• Tuberous sclerosis
  Tuberous sclerosis is a genetic disorder that affects various parts of the body to varying degrees of severity.

• Turner's syndrome
  Turner's syndrome is a random genetic disorder that affects females, causing short stature and infertility.

• Usher syndrome
  Services aim to help a person with Usher syndrome prepare for the dual loss of sight and hearing.

• Von Willebrand disease
  A person with von Willebrand disease may have frequent nosebleeds, heavy menstruation or excessive bleeding from the mouth.

• Williams syndrome
  Williams syndrome often goes undiagnosed, which means that some people with the disorder fail to get the support and treatment they need until later in life.

• Wilson disease
  In Wilson's disease, a build-up of copper damages organs including the liver, nervous system, brain, kidneys and eyes.

Genes and genetics explained
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Children inherit physical characteristics such as eye colour from their parents through their genes.

Gene therapy
Gene therapy is an experimental form of treatment that targets the faulty genes that cause genetic diseases.

Genetic disorders
Genetic disorder is caused by an altered or faulty gene or set of genes.

Genetic services in Victoria
Genetic services can help people who are affected by, or who are at risk of, inherited conditions or birth defects, to make informed choices about their healthcare.

Genetic testing

DNA profiling
DNA profiling is a way of establishing identity and is used in a variety of ways.

Egg freezing
You can freeze your eggs for medical reasons or for reasons that are more to do with your life circumstances.

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Genetic testing for inherited cancer
A predisposition to certain cancers can be inherited via altered genes.

Newborn bloodspot screening
Every newborn baby in Australia is offered a newborn bloodspot screening test to identify those at risk of rare, but serious, medical conditions.

Pregnancy tests – chorionic villus sampling
Chorionic villus sampling (CVS) is a pregnancy test that checks the baby for some abnormalities.

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Australian Stem Cell Centre.
Gene Therapy for Cancer - Questions and Answers.
Genetic Support Network Victoria.
The protection of human genetic information in Australia.

Support Groups
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