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Genetics in a Nutshell

This is the first instalment of the 'Science in a Nutshell' series, produced by the Science Media Centre. It is designed to be useful for busy news desks, especially those that lack ready access to a science correspondent. 'Genetics in a Nutshell' provides you with a clear explanation of some of the terms that are now used in stories about **medical genetics**.

It is intended as a handy guide, to be kept on your desk for use when genetics hits the headlines. It is not meant to teach you genetics from scratch.

If you're still not sure where to get the answers you're looking for, or need to talk to a scientist about genetics, contact the Science Media Centre:
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"Imagine that the **genome** is a book.
There are 23 chapters, called **chromosomes**.
Each chapter contains several thousand stories, called **genes** … Each paragraph is made up of words, called **codons**. Each word is written in letters, called **bases**."
Matt Ridley, from *Genome: the autobiography of a species in 23 chapters*.

celebrating life
DNA DOUBLE HELIX 1953-2003
www.DNA50.org.uk

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Glossary

Bases are the parts of DNA that carry the information to make and maintain a living thing. Most genes contain thousands of bases. There are four different types of base, called A, G, T and C (the initials of their chemical names). A group of three bases (a '**codon**') can carry the code for one building block of a protein.

BRCA1 and 2 <pronounced 'bracca', short for 'Breast Cancer'> are genes whose normal task is repairing DNA. When they go wrong, they are linked with some cases of breast cancer. Only 5 to 10% of breast cancers are associated with these genes, but women with damaged versions of either of them have a 3 to 7 times greater risk of developing breast cancer.

Cell Every body is made of trillions of tiny cells. There are hundreds of types, each with a specialised job. Most cells have a nucleus that contains a copy of the entire genome. Cells are miniature chemical factories, built and regulated by genes.

Chromosome DNA is coiled into structures called chromosomes, found in the nucleus of each cell. Humans have 46 chromosomes - 44 are paired, and the other two are either XX (female) or XY (male) **sex chromosomes**. Each human chromosome can contain between 5 - 20 million bases.

Clone has several meanings: 1) An exact copy of a DNA molecule, as in 'to clone a gene' 2) More commonly, clone refers to an entire living thing that is genetically identical to another. Identical twins are natural clones, as are all the bananas in a bunch. There are several different ways of **cloning an animal**. Dolly the sheep was created by taking all the chromosomes from an adult sheep's cell, and putting them into an empty (sheep) egg cell. In the UK it is illegal to transfer cloned human embryos into the womb to enable them to develop further. It is probably unsafe to do so at the moment because these **reproductive cloning** experiments in animals produce many embryos that develop abnormalities.

Therapeutic Cloning DNA is taken from a human patient to make cloned **embryonic stem cells**, which many scientists believe could be useful for treating 'cell-loss' diseases like Parkinson's (this has, as yet, only been attempted in animals). The stem cells are turned into specialised cells for transplant, which are unlikely to be rejected because they are an exact genetic match with the patient. Therapeutic cloning is permitted by UK legislation, but research is strictly regulated by the HFEA.

Code The order of the bases in the working parts of DNA is a code, which can be translated into instructions to make proteins. Hence we say that a gene 'codes for' a protein.

Designer Babies A non-scientific term for children whose genetic make-up has been altered or specially chosen e.g. by selecting a particular embryo during IVF. This may help parents to avoid having a child with a genetic disease. If parents have a sick child that could be cured by a special cell transplant, they may choose to have another child with the correct genetic match by IVF (e.g. the Hashmi case), although this is still a matter of legal dispute (Jan 2003). Manipulating genes to produce children with specific traits such as eye colour, physical prowess or musical ability is currently impossible.

DNA Deoxyribonucleic acid <pronounced 'dee-ox-ee-rye-bow new-clay-ick acid'> is the long chemical chain that genes are made from. DNA contains two strands, twisted together into a **double helix** (a shape that looks rather like a spiral staircase). Pairs of chemical units called bases stick these strands together – their precise order, the **DNA Sequence**, determines which proteins are made, and when. All the DNA in a living thing is known collectively as its genome.

Expression A gene is **expressed** when the information it stores is used to make proteins, or to switch other genes on.

Functional genomics studies the relationship between proteins and the genes that make them, in order to understand the role of every single gene (also see proteome).

Genes carry the information to create everything from organs to offspring. They determine your hair colour, your blood type, susceptibility to certain diseases and so on. Genes are made from the chemical DNA. A gene can ultimately produce a particular protein, or act as the trigger to start other processes in the body. Although we share many of our genes with other animals, the actual **DNA sequence** of that same gene often varies between species - they are telling the same story with different words.

Gene Therapy is a potential way of treating disease where damaged or abnormal genes are replaced with normal ones. Although still quite experimental, there have been a few successful cases. For example, Rhys Evans was born with a defective immune system, forcing him to live inside a sterile 'bubble'. Gene therapy on his blood cells repaired the fault in their DNA, allowing him to live a normal life. However, recent gene therapy trials in France seem to have caused leukaemia in two patients, casting some doubt on this technique. **Germline gene therapy** applies the method to eggs or sperm, so that *all* the DNA in a resulting embryo is altered. This is not done in humans for ethical, practical and safety reasons.

Genetic Fingerprinting uses an individual's unique genetic code in criminal investigations or paternity cases. Most scientists believe that it is very reliable - the odds of two people having the same genetic fingerprint are about one in a billion.

Genetic Testing Kits can detect individual differences in DNA, some of which may have medical importance. In the US, many simple test kits are available from pharmacists, but their accuracy is limited. Some claim that these tests may help people to tailor their diet or medical treatment to their personal genetic profile. The UK situation is under review by the Human Genetics Commission (Jan 2003).

Genome The sum total of all your DNA. There are about 3 billion base pairs in the human genome, which are organised into

30,000 to 40,000 genes. A map of the human genome allows scientists to understand more about our body chemistry, and potentially helps them to tackle disease. The mouse genome has already helped scientists to improve their experiments on mice, relating effects seen in this 'animal model' to human diseases.

IVF or **in vitro** <"in glass"> **fertilisation** brings a sperm and an egg together outside the womb. Children conceived using this technique are sometimes called ‘test tube babies’.

Junk DNA is that part of DNA that appears to serve no *known* function. Although 97% of human DNA never actually makes a protein, some of this can still be very important because it turns nearby DNA on and off.

Mutation Changes in a DNA sequence that may have a profound effect e.g. mutations that cause cystic fibrosis. However, many mutations will have no detectable effect.

Nucleus The information hub of a cell that contains the chromosomes.

Phenotype Any physical effect (e.g. growing an arm, or developing breast cancer) that is influenced by a gene.

Preimplantation Genetic Diagnosis/Screening When a couple plan to have children, they can take genetic tests for inherited diseases. If they BOTH carry one copy of the gene for, say, sickle cell anaemia, their child will have a 1 in 4 chance of having the disease. If only one parent carries the offending gene, their child is safe. This can help parents in planning a family. It may also prompt them to screen an embryo in the womb – if it has a genetic disorder, parents may choose to abort the child. Screening can present huge ethical dilemmas e.g. tests for diseases with no cure; screening for disabilities.

Proteins are chemicals that are essential for almost all life processes. Genes carry the information needed to make proteins. Some proteins are **enzymes** that speed up chemical reactions in our bodies. Others can: carry messages between cells; control the activity of other genes or proteins; build muscle, hair etc.

Proteome The sum total of all the proteins in your body. Studies to explore what proteins do and how they are made, known as **proteomics**, will be the focus of much research for decades to come and will help scientists to tackle disease.

RNA (Ribonucleic acid) is a chemical cousin of DNA. Messenger RNA (mRNA) carries the genetic information stored in DNA to the part of the cell that makes proteins.

RNAi (**RNA Interference**) uses short sections of RNA to turn the effects of DNA on and off. This is a natural process in cells that scientists can use to understand how DNA works, and will probably be the subject of many breakthroughs in the next few years. *Science* chose RNAi as their 'Breakthrough of 2002'.

Sex Selection During IVF, it is possible to select only male (or only female) embryos for implantation into the womb. This is currently illegal in the UK – however, sperm sorting is still allowed, where sperm that will create a male child are separated from those that will give rise to a female. The HFEA are (Jan 2003) consulting the public about whether / how to regulate sex selection.

Shotgun Sequencing is the method most often used to read a genome. It involves chopping the DNA into little pieces, working out what they all are and then getting a computer to fit all the jigsaw pieces back together in the right order.

Stem Cells have the potential to turn into many different types of cell (described by scientists as ‘pluripotent’). **Embryonic stem cells** can turn into any type of tissue – they are taken from very early embryos that are about five days old, and comprise about 100 cells. These embryos are not suitable to implant into a womb. **Adult stem cells**, found principally in bone marrow, appear to be more limited in the range of cells they can turn into. However, there is still debate about whether embryonic or adult stem cells will ultimately be more useful. Stem cells could be collected to replace damaged tissue e.g. to treat Parkinson's disease, in cancer patients, to treat blood disorders etc.

Transgenic Any living thing with artificially added or altered DNA *from another species* is 'transgenic' e.g. the transgenic mouse with the cystic fibrosis gene. Such creatures may help scientists to understand, and perhaps treat, the disease. Other genes can be changed so that an animal's organs are coated with human antigens (chemical markers), allowing **xenotransplantation** without rejection.

Vector A virus used to carry a section of DNA into a cell e.g. during **gene therapy**.

Xenotransplantation Tissue transplant between two different species. Animal-to-human organ transplants have not yet been carried out.

Issues

Disease A few rare diseases are caused by a single mutation in the DNA – just one faulty base out of about three billion e.g. **cystic fibrosis** or **Huntingdon's disease**. Others, such as **breast cancer**, are caused by a variety of factors including diet, exercise, age *and* genetic influence. Genetic tests might say if someone is *more likely* to get a particular disease. In the future, this information might lead to better treatments for disease.

Insurance In 2001, the government and the Association of British Insurers established a five-year moratorium on the use of genetic test results in insurance. The Human Genetics Commission is currently (Jan 2003) reviewing how insurers use 'family histories'. Insurance companies cannot ask individuals to take a genetic test. Insurance can only be affected by a genetic test is if you have a family history of Huntington’s disease; a negative test result counts in your favour.

Gene Patenting A patent usually lasts for 20 years. If other people want to use the invention commercially, they need to obtain permission from the inventor. A patent stakes a claim on something useful, novel, and inventive. Human genes, as they exist in the cells of our bodies, cannot be patented because they are not inventions. However, copies of genes manipulated in the lab can and do form parts of patents. In July 2002, the Nuffield Council on Bioethics concluded that genes have sometimes been patented too easily and that patents involving DNA sequences should be the exception rather than the rule. Although patents give companies an incentive to invest money in research, some fear that non-commercial research may suffer as a result.

Genes For … We often hear about a 'gene for' something. But if scientists discover a gene that is *associated* with a certain condition, it doesn't necessarily follow that the gene is *responsible* for that condition. For example, most criminals are male. They are male because of a small gene on their Y chromosome. But that does NOT mean that this is the gene for criminality, even though there is a high degree of association between the two.

Contacts and Links

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Human Fertilisation and Embryology Authority
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