The basics

Our bodies are made up of trillions of cells. All the cells in our body (except red blood cells) contain a nucleus, in the centre of the cell.

Inside the nucleus we have tiny thread like structures called chromosomes. Each cell has 46 chromosomes in its nucleus. These are what we inherit from our parents in the egg and sperm. We get 2 sets of chromosomes, 23 matching pairs, one set from Mum and one set from Dad.

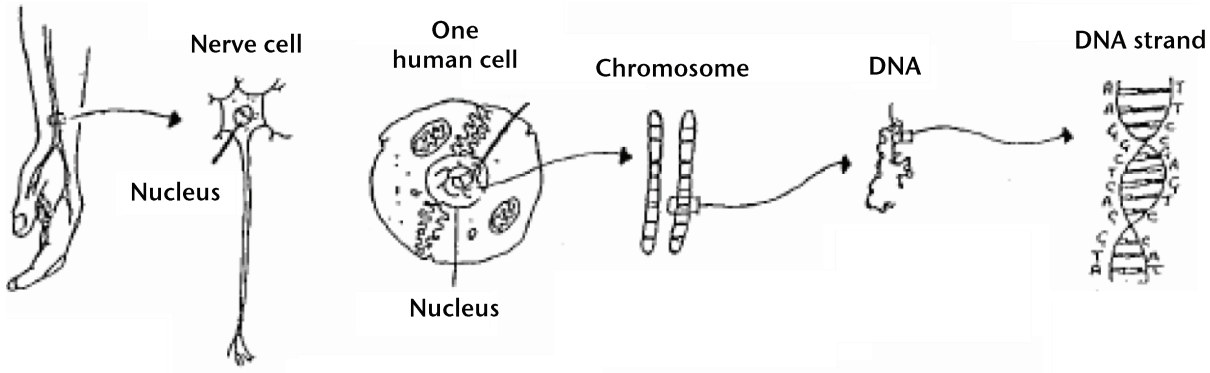
Chromosomes are made up of a substance called DNA (deoxyribonucleic acid) which is a chain of chemicals, mostly fat and sugar molecules and four 'bases' called Adenine, Thymine, Guanine and Cytosine (A, T, G and C for short). These 4 chemicals – A, T, G and C – are the building blocks of our DNA.

A gene is simply a length of DNA made up of thousands of bases arranged in a very specific order. This is our genetic code. The sequence of the bases in each gene are responsible for how we look but more importantly, they act like a set of instructions for our body to function.

The sequence of the bases are actually instructions for our body to make proteins. It is these proteins that control and carry out all the functions of our whole body.

Proteins can be things like hormones, chemical messengers in the brain, components of our skin and hair, signalling molecules in the immune system and lots more.

Mistakes or mutations in the order of the DNA bases, the set of instructions, can cause faulty proteins to be manufactured. If these proteins can't carry out their jobs properly the body malfunctions; when this occurs a person is said to have a genetic disease.



THE HUMAN BODY CONTAINS 100 TRILLION CELLS

THERE IS A NUCLEUS IN EACH HUMAN CELL

IN EACH NUCLEUS THERE ARE 46 CHROMOSOMES – 23 PAIRS

CHROMOSOMES ARE MADE UP OF TIGHTLY COILED STRANDS OF DNA

GENES ARE SEGMENTS OF DNA THAT CONTAIN INSTRUCTIONS TO CREATE PROTEINS

Genetic Disease

There are many examples of genetic diseases that people can be born with. Here are just three:

Cystic fibrosis - a gene mutation causes a defective protein to be made that is vital for our lungs and digestive system to function. People affected with this condition need physiotherapy to dislodge the sticky mucus that fills their airways; they may eventually need a lung transplant. All affected people must take medicines in order to digest food properly.

Beta thalassaemia - caused by a faulty gene that makes part of the haemoglobin protein. Haemoglobin carries oxygen and carbon dioxide around the body. People affected have severe anaemia as oxygen cannot reach their organs. Several blood transfusions can be required in the first years of life.

Muscular dystrophy - gene mutations in muscle proteins cause the muscles to malfunction. This can cause severe muscle weakness and disability. Different types of dystrophies have different symptoms; in severe cases children are severely disabled by age ten and most only survive to their twenties.

Other conditions also often have important genetic components that can increase a person's risk or susceptibility to a disease; for example some types of cancer (breast, bowel, prostate), allergies and asthma, cardiac disorders (incl. high cholesterol and blood pressure), coeliac disease (gluten sensitivity), hearing and eyesight impairments.

Children's Medical Research Institute conducts fundamental scientific research into the genetics of human health and disease. Our research efforts include:

- Understanding and developing more effective treatments for cancer
- Understanding and trialing therapies for rare diseases like OTC-deficiency
- Discovering the genes vital for the development of a healthy baby in order to one day prevent conditions such as cleft palate and eye abnormalities; and
- Unlocking the mysteries of how brain cells communicate and what goes wrong in conditions such as epilepsy and schizophrenia

Resources

- Smith, T (ed). 1995. The Human Body – an illustrated guide to its structure, function and disorders. Dorling Kindersley Ltd. London
  A complete, well-illustrated guide to the structures, functions and disorders of the human body; including medical illustrations with easy-to-follow text showing body cells, tissues, organs and structures.
- Excellent fact sheets on basic genetics and genetic disorders can be obtained from the Centre for Genetics Education. www.genetics.com.au, or phone on 02 9926 7324

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