DNA
the blueprint for life
The monastery and garden in which Gregor Mendel did his experiments.

Nobel Prize winners, James Watson (left) and Francis Crick (right), with their model of DNA after discovering its structure in the 1950s.
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This educational resource provides answers to the following key questions:

• What is genetics?
• What is DNA?
  - What does DNA stand for?
• Who has DNA?
• Where is my DNA?
• What does DNA look like?
• What is so important about DNA?
• What is the impact of genetics?
• What career opportunities are there if you have a qualification in a subject that uses genetics?

What is Genetics?
This is the scientific study of the principles of heredity (resemblance) and the variation of inherited traits among related organisms. All living things inherit traits from their parents. This knowledge has been used since prehistoric times for plant and animal improvement in the form of selective breeding. Modern genetics science seeks to understand the process of inheritance. The word ‘genetics’ is derived from the Greek root ‘gen’ which means ‘to become or to grow into’. It was coined by William Bateson in 1906 for the study of physiology of heredity and variations. Modern genetics science traces its roots to Gregor Johann Mendel.

Who was Mendel?
Gregor Johann Mendel was a 19th century monk who passionately completed a series of breeding experiments on pea plants. He examined how traits were inherited by the offspring from the crosses of various parent plants. He found that the inheritance of traits always followed certain patterns and he established several “Laws” of inheritance. He was not a geneticist, in that he did not study genes or genetic material, and it was only later - with Darwin’s theory of evolution by natural selection (which relies on there being inherited and variable characters for evolution to act upon) and the study of the actual genetic material of organisms in the 20th century - that the significance of his pea experiments was realised.
What does DNA stand for?
DNA stands for DeoxyriboNucleic Acid. It is a molecule that constitutes the genetic material of the cell. It is found in the chromosomes in the nucleus of the cell, as well as in other cellular organelles like the mitochondria in animals and chloroplasts in plants. DNA is the biological code that contains the genetic instructions used in the development and functioning of all living organisms. This code is divided up into different regions called genes. Products that the genes code for, which most often are proteins, are used to build features and initiate or regulate certain processes in the cell or tissue. So the genes determine how organisms are made, what they look like and all the processes that sustain them.

DNA is often described as the blue-print of an organism. The information that DNA contains is passed from one generation to the next. Half of an organism’s DNA comes from the female parent and the other half from the male parent. Genetic diseases can result if an organism has too much or too little DNA, is missing pieces of DNA or if the DNA pieces have been mixed up.

Where is DNA found?
You are made up of billions of cells. These cells are organised into tissues such as skin, muscle and bone. Inside nearly every cell is a nucleus containing an unique set of chromosomes. Each of these chromosomes consists of a compact coil of an incredibly long molecule of DNA. DNA is tightly coiled so that it is able to fit into the nucleus of a cell. DNA stores all the coded information needed for everyday growth and metabolism. This information is passed down from generation to generation.

Who has DNA?
All living organisms have the capacity for reproduction. Reproduction is the production of a new generation of offspring that resembles the parent. It involves the transfer of biological information of the parent generation to the new organism via the egg and sperm. Thus, the offspring of dogs will always resemble their parents and never cats or elephants. This tendency of individuals to resemble their progenitors is called ‘heredity’.

The structure of DNA
DNA is a molecule composed of a chain of different types of chemical building blocks called ‘nucleotides’. DNA looks like an incredibly long twisted ladder. This shape is called a ‘double helix’. The sides of the ladder are a linked chain of alternating sugar and phosphate molecules. The rungs connect to the sugar molecules and are known as ‘bases’ or ‘nucleotides’. There are four bases - adenine (A), thymine (T),
guanine (G) and cytosine (C). Each rung is made up of two bases that link together. DNA naturally occurs in a double stranded form, with nucleotides that are complementary to each other on each strand. Because of their chemical nature, A will only link with T and G will only link with C. Genes correspond to regions within DNA. So, a particular gene will represent a given stretch of the DNA ladder and will be responsible for building a particular product or regulating a particular process in the cell.

The sequence of these nucleotides is the genetic information that organisms inherit. DNA from all living organisms is made of the same sugar and phosphate molecules and the same four bases. DNA is made of the same chemicals and has the same structure wherever it is found, whether in human cells, those of a cactus, of a worm or a bacterium. The most important difference is the order or the sequence of the bases in the DNA molecule. It is this sequence that is referred to as the genetic code, and why it is sometimes called ‘the code of life’.

**Gene interaction and function is a complex phenomenon**

It seems reasonable that if two genes with the same sequence are in the same cell, they should act the same way. But that is not always true. So-called ‘epigenetic factors’ can alter how a gene works regardless of its DNA sequence.

One well studied example is parental imprinting. Certain genes are marked with chemical tags via a process called ‘methylation’ while they are still in a sperm or egg, meaning that only the maternal or paternal copy is active in the offspring. As a result, certain traits are inherited exclusively from one side of the family.

Genes can also influence each other. Identical sequences in identical cells of different individuals can be expressed or behave differently, because of the influence of other genes which differ between these individuals.

**Genes and the environment**

There is debate about the influence of nature (genes) and nurture (environment) on an organism. Modern genetics suggests that both factors play a role. That is no surprise as genes are a piece of cellular machinery. Dangerous chemicals, such as cigarette smoke, can jam that machinery or interfere with its workings.

On the other hand, a healing environment can compensate for a faulty gene. For example, babies who are born with the disease phenylketonuria (PKU) lack an enzyme that metabolises the amino acid phenylalanine. As a result this amino acid builds up to toxic levels causing mental retardation. Babies are now screened for the defect at birth. There is no cure for the disorder, however, those with two
copies of the defective gene are given special diets low in phenylalanine. As a result, they develop normally.

**Impact of genetics**
As a biological science, genetics has a deep impact on the cultural and social evolution of mankind. It has helped the modern man to improve agricultural and horticultural resources and to cope with various hereditary diseases. In agriculture, domestic animals and crops are improved by selective breeding. In medicine genetics has increased the understanding of diseases like diabetes, blindness and deafness. Various legal problems like paternity disputes are solved by applying genetic testing. Above all genetics has removed faulty beliefs that persist in our cultures concerning inheritance.

**Career opportunities**
Have you thought about what you want to be when you leave school? One option is for you to study genetics. Studying genetics does not mean you need to follow this course of study to the exclusion of everything else. Studying subjects related to genetics such as Molecular Biology, Anthropology, or even Evolutionary Biology can be excellent starting points for a range of exciting and varied careers. Below are listed some of the career paths a person who studies genetics can follow.

**Medicine**
- **Medical Genetics** involves the diagnosing and management of genetic disorders.
- **Pharmacogenetics** examines the impact of genetic variation on the way different people respond to medications

**Biotechnology**
- Biotechnology uses and adapts biological processes to meet human needs

**Agriculture and Wildlife**
- **Genetic Modification**: using genetic engineering techniques to alter genetic material for improved productivity, better resistance to illness or disease
- **Wildlife Management**: using models to estimate genetic variation and social structure
- **Pedigree and breeding**: to determine the pedigree and kinship of stock and to select organisms for breeding
- **Species Identification**: for understanding and managing biodiversity or for identifying invasive organisms and pests

![The inheritance pattern of PKU](image)
**Law and Justice**
- Forensic Science: using DNA as a method to solve crimes

**Academic Science / Research**
- Sequencing of Organisms: using methods to determine the order of nucleotide sequences in a DNA molecule to determine how populations are structured, how organisms are related and how they have evolved
- **Functional Genomics**: the use of genetic data to describe gene function and interactions
- **Microbial Genetics**: involves the genotype study of small organisms

**Genetic Fun Facts**
- DNA was first isolated in 1869 by Friedrich Miescher.
- The DNA helix (ladder) usually twists to the right.
- The average gene is 10,000 to 15,000 bases long.
- It takes about 8 hours for one of your cells to completely copy itself.
- You can fit about 1000 nuclei across the full stop at the end of this sentence.
- Humans are 99.9% identical - only 0.1% makes up for the differences.
- We share 90% of our genes with mice and 85% with zebra fish.

**Activity**
Many traits (characteristics) in humans are controlled by genes. Some of these traits are common features like eye colour and whether you have dry or sticky earwax! A characteristic (gene) that is possessed by one parent which appears in the offspring and hides the corresponding characteristic derived from the other parent is said to be dominant. The gene which is hidden is said to be recessive.

**Example**: In Mendel’s classic experiment (see above), he found that peas having yellow seeds crossed with peas having green seeds gave rise to progeny all having yellow seeds: i.e. yellow as the dominant characteristic and green was the recessive characteristic.

Genes may actually cause disease as they are inherited from one generation to the next.

Human geneticists illustrate the inheritance of a gene within a family by using a pedigree chart.

**Challenge Question**
On the following page is a pedigree chart for the inheritance of achondroplasia (ay-kon druhiplay-zhuh), which is a form of dwarfism. Dark circles or squares indicate individuals with achondroplasia. Examine the pedigree chart and answer the following questions.
1. Is the gene that causes this form of dwarfism a recessive or dominant trait?
2. How do you know?
3. Using (D) to represent the dominant allele and (d) to represent the recessive
allele, write the genotypes of the indicated individuals. For one of the labelled individuals, there are two possible genotypes. Write both of the genotypes and indicate which one is more likely.

Based on the frequency of dwarfs among the people you have seen in your lifetime, do you think that the allele for achondroplasia is common or rare in the population?

**Glossary**
- **Allele**: one of a set of alternative forms of the same gene
- **Base**: a building block of DNA
- **Chromosome**: a structure in the nucleus of a living thing that contains DNA, and is used to pass genes down to any future generations
- **Cloning**: a process of producing cells and living things with identical genes
- **Dominant gene**: Describes a characteristic (or gene) possessed by one parent which appears in the offspring and masks the corresponding characteristic derived from the other parent which is said to be recessive.
- **DNA**: the substance of which genes are made
- **Gene**: A part of a chromosome, consisting of DNA, with the ability to transmit hereditary characteristics from one generation to another.
- **Genetics**: the branch of biology that studies heredity and variation in organisms
- **Genetic engineering**: the set of techniques used to alter the genetic material of a cell or living organism
- **Heredity**: the passing on of traits from the parents to the children
- **Inheritance**: Manner in which genes and traits are passed from parents to offspring
- **Metabolise**: break down to smaller components
- **Nucleotide**: the basic structural unit of nucleic acids

**References**
2. http://learn.genetics.utah.edu
Challenge Answers
1. Dominant
2. The trait is present in all generations.
3. 
   1 = Dd (use Punnett square to verify)
   2 = dd
   3 = dd
   4 = DD/Dd
   5 = Dd

   Punnet square:
   
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4. Individual 4 = DD or Dd. The most likely genotype is DD, this is because all the offspring show the dwarfism trait.
5. The frequency of dwarfs that I have seen in the general population is low. My observations show that the allele is rare in the population.

Gregor Johann Mendel