Early research into genetics

By 1900, a German scientist, Mendel, had theorised that genes come in pairs, and one is inherited from each parent. These were known as the **fundamental laws of inheritance**. Unfortunately, he did not have scientific proof that his laws were correct. Microscopes were not yet powerful enough to be able to identify gene pairs.

Timeline

Early work on genetics

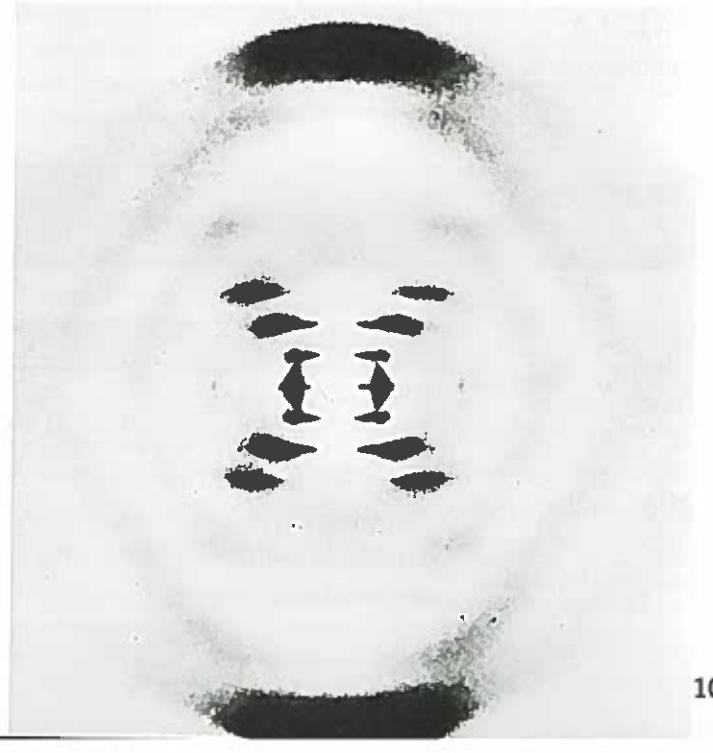
1902 Archibald Garrod, an English doctor, theorises that hereditary diseases are caused by missing information in the body's chemical pathways.

1941 US scientists George Beadle and Edward Tatum prove Garrod's theory.

1951 At King's College in London, Rosalind Franklin and Maurice Wilkins create images of DNA using x-rays (see Source A).

Source A

Rosalind Franklin took this x-ray photograph of DNA in 1951 whilst working alongside Maurice Wilkins.



By 1951, scientists knew that characteristics were passed down from parents to children, as children often look like their parents. They theorised that a substance in human cells passed on this information from one person to the next. This substance also passed on a variety of hereditary diseases. However, it was not until 1953 that technology finally made it possible for scientists to find the missing piece of the puzzle: DNA*.

Key term

DNA*

Short for deoxyribonucleic acid, DNA carries genetic information from one living thing to another. DNA information determines characteristics like hair and eye colour.

Watson, Crick and the discovery of the human gene

James Watson was an American biologist. Francis Crick was an English physicist. In 1953, they were both working at Cambridge University, where they shared an office. Even though neither men were investigating DNA, they both had a strong interest in researching and finding out more about human biology.

Crick and Watson saw the x-rays provided by Franklin and Wilkins (see Source A). They built their model of DNA and shared it with Franklin, who made a correction based on her x-rays. Wilkins also shared clearer photographs that they had managed to take of the DNA. Due to this additional input, Crick and Watson were able to solve the puzzle of the structure of DNA. They discovered that it was shaped as a double helix, which could 'unzip' itself to make copies.

Watson and Crick published their paper in April 1953. Crick suggested that they had discovered the secret of life. Understanding the shape of DNA meant that they could now begin to look at its structure and identify the parts that caused hereditary diseases.

*The mapping of the human genome**

Once the structure of DNA was understood, teams of scientists began to break it apart to understand how it worked. All the information that builds a person is stored in their DNA.

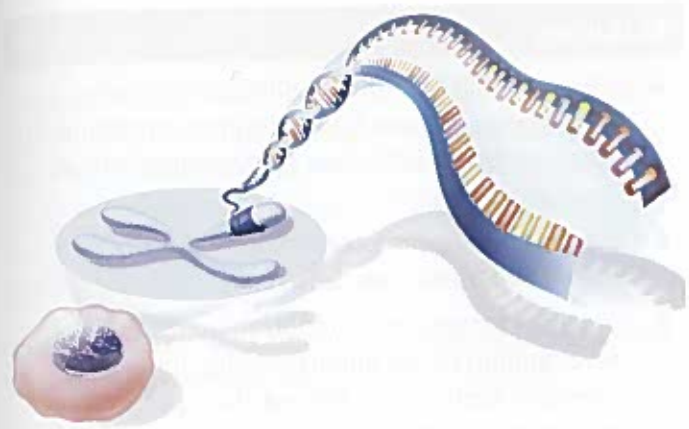


Figure 4.1 An image of the double helix formation of DNA. DNA is stored in every human cell.

Understanding that information – mapping the DNA's code – was vital to helping scientists understand the cause of genetic diseases, such as haemophilia*.

The Human Genome Project was launched in 1990. It was originally led by James Watson himself. For a decade, 18 teams of scientists all over the world worked together to decode and map the human genome. Even though hundreds of scientists were working towards this goal, they did not complete the first draft until 2000.

Once the human genome was mapped, it then became possible for scientists to use this blueprint of human DNA to look for mistakes or mismatches in the DNA of people suffering from hereditary diseases.

For example, scientists have now been able to identify a gene that is sometimes present in women who suffer from breast cancer. Although they cannot use this knowledge to **treat** breast cancer, women now have the opportunity to **prevent** this disease by identifying their risk of developing the disease and then having a mastectomy*. A famous example of this is the actress Angelina Jolie, who had herself tested for the gene because her mother had died of breast cancer.

Key terms

Genome*

The complete set of DNA containing all the information needed to build a particular organism. In humans, this is more than three billion DNA pairs. It is unique for every human being, except identical twins.

Haemophilia*

A genetic disease passed from parent to child that stops blood clotting. Sufferers from haemophilia must be careful, as an open wound will not heal correctly. Famously, many of Queen Victoria's ancestors suffered from haemophilia.

Mastectomy*

Surgery during which a person has one or both of their breasts removed.

Source B

In this 2013 news article, Angelina Jolie explains why she chose to have a mastectomy.

Angelina Jolie bravely reveals she has had a preventive double mastectomy after tests showed an 87% chance of contracting breast cancer.

The Hollywood actress is healthy and made the decision to undergo the procedure after discovering she carries the BRCA1 cancer gene.

Angelina said: "My doctors estimated that I had an 87% risk of breast cancer and a 50% risk of ovarian cancer. I made a decision to have a preventive double mastectomy."

The surgery was successful and doctors believe Angelina's chances of developing breast cancer have reduced to less than 5%.