

When a gene code is altered

Some human diseases are caused because a small part of the code for one of the proteins essential for normal body functioning has changed (mutated). Mutations can result in proteins with incorrect form and shape, or no protein production at all. As a result, some body functions can be affected.

Some examples of such diseases are phenylketonuria (PKU), sickle cell anaemia, cystic fibrosis, thalassaemia and haemophilia.

In this research task, you can investigate one of these diseases, or another one that you might find for yourself.

You will need:

- access to the internet
- access to library resources

What to do:

1. Carry out a web search using the name of the disease you have chosen, or using a general query such as 'genetic disorders'. Also look in the library for relevant references.
2. Prepare a presentation to the class or a small group that outlines some or all of the following features of the disease:
 - Where is the gene located?
 - What is the alteration to the genetic code that causes the disease?
 - How is protein production affected?
 - What is the nature of the disease?
 - How is the disease treated?
 - How effective is the treatment and how does it contribute to the quality of life of the patient?
 - Are there any social, economic or ethical issues associated with the diagnosis, treatment and management of the disease?
 - Is there current research into new ways to treat the disease with biotechnology techniques?

Try this web site as a starting point: <http://www.genetics.com.au>

For more information about causes, diagnosis, incidence, symptoms, inheritance and treatment of a number of genetic diseases, as well as what it is like to live with a genetic disorder, go to: <http://www.ygyh.org/>