WHAT IS A GENETIC DISORDER?

A CONDITION CAUSED BY A CHANGE IN THE DNA SEQUENCE

Your body is made up of trillions of cells. Every cell contains **DNA**, deoxyribonucleic acid, which is the fundamental building block of every person.

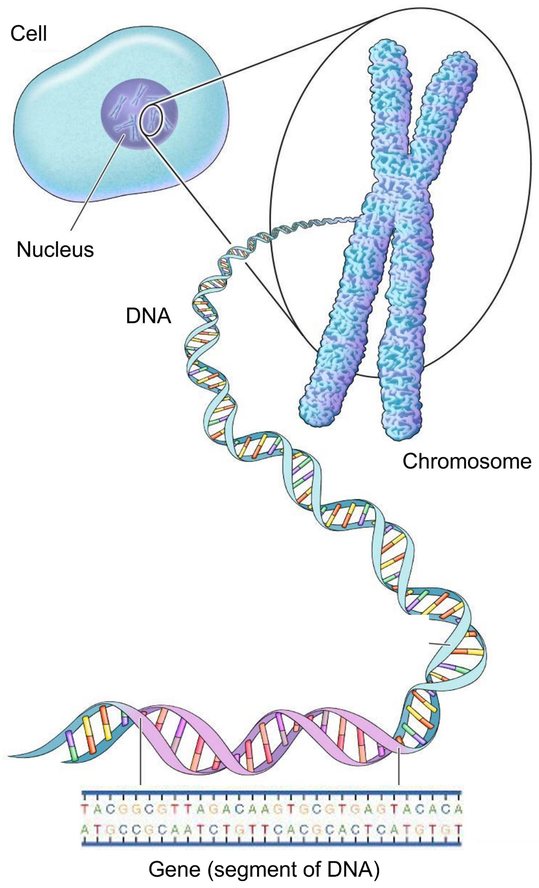
Your DNA is made of 3.1 billion letters and contains all the instructions for you to grow throughout your lifetime.

In each cell of your body, DNA is tightly packaged into structures called **CHROMOSOMES**.

GENES are sections of the DNA, each containing a message (specific sequences of letters).

Most genes contain the instructions to produce **PROTEINS** in our bodies which carry out specific jobs.

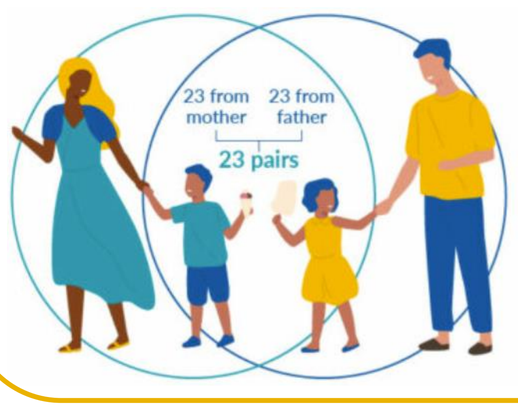
Like determining your hair and eye colour



Gene (segment of DNA)

DID YOU KNOW?

You inherit one copy of your genes from each parent.



Most cells contain 23 pairs of chromosomes (46 total).

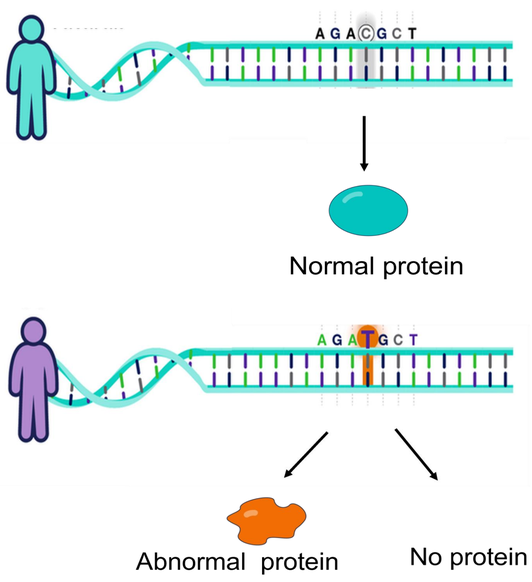
You receive one set of chromosomes from each parent.

WHAT HAPPENS IF THERE IS A CHANGE IN THE DNA SEQUENCE?

Think of it like reading, every letter counts so changing even a single letter can alter the meaning of an entire word or sentence.

WHAT IS THE COLOUR OF YOUR HAT?

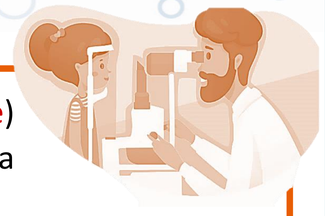
WHAT IS THE COLOUR OF YOUR RAT?



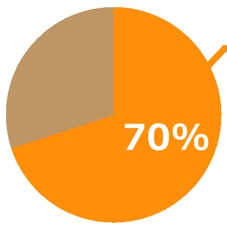
- To function correctly, each cell depends on thousands of proteins to do their jobs in the right places at the right times.
- A change in the DNA sequence can alter a gene's instructions for making a protein.
- This can cause the protein to not work properly or to not be produced at all.
- When this happens, genetic disorders may occur.

WHAT DO WE DO?

SOLVE THE MYSTERY BEHIND EYE DISORDERS



Anophthalmia (missing eye), Microphthalmia (small eye) and Coloboma ('gap' in the eye), known as **AMC**, are a group of developmental eye disorders.



UNSOLVED CASES

- **Every year in the UK, ~100 babies** are born with AMC
- Many of these children have additional needs or other health challenges

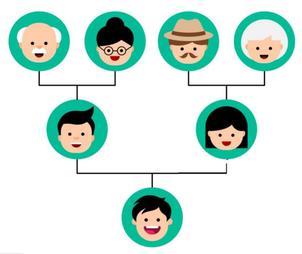
OXFORD BROOKES UNIVERSITY

RAGGE GROUP: Molecular Genetics of Human Eye Development
We are a group of scientists whose job is to try and solve these cases!

MEDICAL RECORDS

Symptoms and other details about the patient

FAMILY HISTORY



The family tree can hold important clues about the condition

DNA

DNA is extracted from the blood of the individual and close family relatives

Information is analysed

The genome will be read letter by letter and analysed with medical records and family history to give a full picture

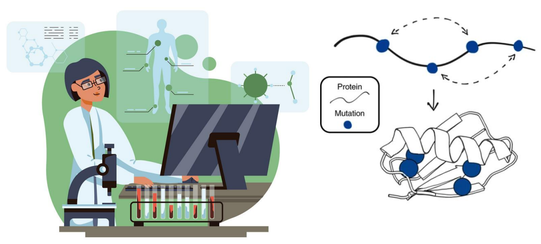
We compare the sequence of the patient with other genomes to find DNA changes that are rare in the general population

Once we have found all the rare DNA changes, we then look for more clues

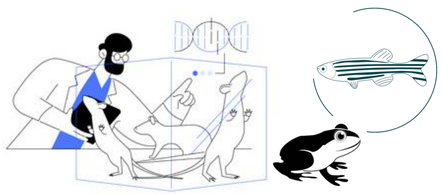
RESEARCH

We use computer programs to understand the effect of DNA changes

- How does the DNA change affect the message coded by the gene?
- Do the new instructions result in a protein with a different shape?



We look at animals to help us understand the role of the genes



Is the gene important for eye development in other animals?

If so, the gene might have an important role in the formation of the human eye too!

By publishing our research, we help doctors diagnose patients and understand these disorders better

GENETIC DIAGNOSIS AND COUNSELLING

- Characteristics of the condition
- Risk of passing on the condition
- Treatment options
- Support and information
- MACS is a charity that supports children and adults born with these eye conditions