

## DNA, genes and genomes - General

### Variation

#### Am I unique?

Yes. Although, at the level of our DNA, researchers currently think that any two human beings are more than 99% alike, there are differences between us. These differences can be small changes in a single DNA letter or duplicates and deletions of much larger chunks of DNA.



#### Can genomes change?

Genomes change - between generations or over a lifetime - these changes are called mutations. Mutations can be helpful, harmful or make no difference at all.

You can inherit mutations from your parents. Mutations are also happening in your cells all the time. Environmental factors like smoking and sunlight can increase the rate of DNA mutation in your cells.



Mutations can happen anywhere in the DNA - in the noncoding DNA as well as the coding DNA of our genes. Many mutations will do nothing at all because they occur outside the important coding and noncoding regions of our DNA.

At this stage, we know most about mutations that we can detect. These are the relatively few mutations that directly cause disease or an obvious change in the way our bodies work. In the future, researchers expect to learn more about mutations with small or indirect effects. For example, small changes that switch a gene on at the wrong time, or mean that not quite enough protein is produced, could contribute to complex diseases like diabetes or hypertension.

## DNA, genes and genomes - General

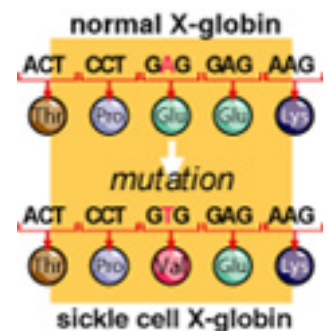
### Variation

#### How can mutations cause disease?

Sometimes, one of the DNA letters is accidentally swapped for another letter. This mutation might have a very serious effect, or none at all.

If the letter T is swapped for an A in the codon GCT then the protein will still be the same, since both the old codon (GCT) and the new codon (GCA) code for the amino acid Alanine.

However, swapping an A for a T in a gene for haemoglobin causes a serious disease called sickle cell anaemia. Haemoglobin does the job of carrying oxygen around the body in the blood. In people with sickle cell anaemia, the sequence of amino acids in haemoglobin is different, and so it doesn't work as well.



There are other types of mutations as well. Sometimes, a bit of the DNA sequence is missed out by mistake, or a new bit added in. Sometimes, parts of the sequence are swapped over, even between different chromosomes.

#### Inheriting mutations

Each of our genes is a copy from either our mum or our dad. If there is a mutation in one of these genes, this can be passed on from parent to child along with the rest of the gene. This is why diseases can run in families.

Small inherited changes can make big differences in our bodies. For example, the most common mutation to cause cystic fibrosis - a disorder where a person's internal organs become clogged with thick mucus - is the loss of three letters in a gene called CFTR.