Genetics 101

Genes

- Every person has more than 20,000 genes.
- Genes are carried on long strands of **DNA** and are found in almost every cell in the human body. In each cell, the DNA is arranged into **chromosomes**.
- Each gene tells the body to make a different protein.
- Together, genes carry information that tells the body how to grow, develop and function.
- This information is together known as the **genetic code.**

In short...

- **Genes** carry the body's **genetic code** – information that tells the body how to grow, develop and function
- Changes in the genetic code that change the way genes work can cause **genetic conditions**
- Genetic conditions may be inherited (passed down) from a parent, or they may occur for the first time in a person by chance
- Changes in the ***SCN2A* gene** can cause a **genetic condition**

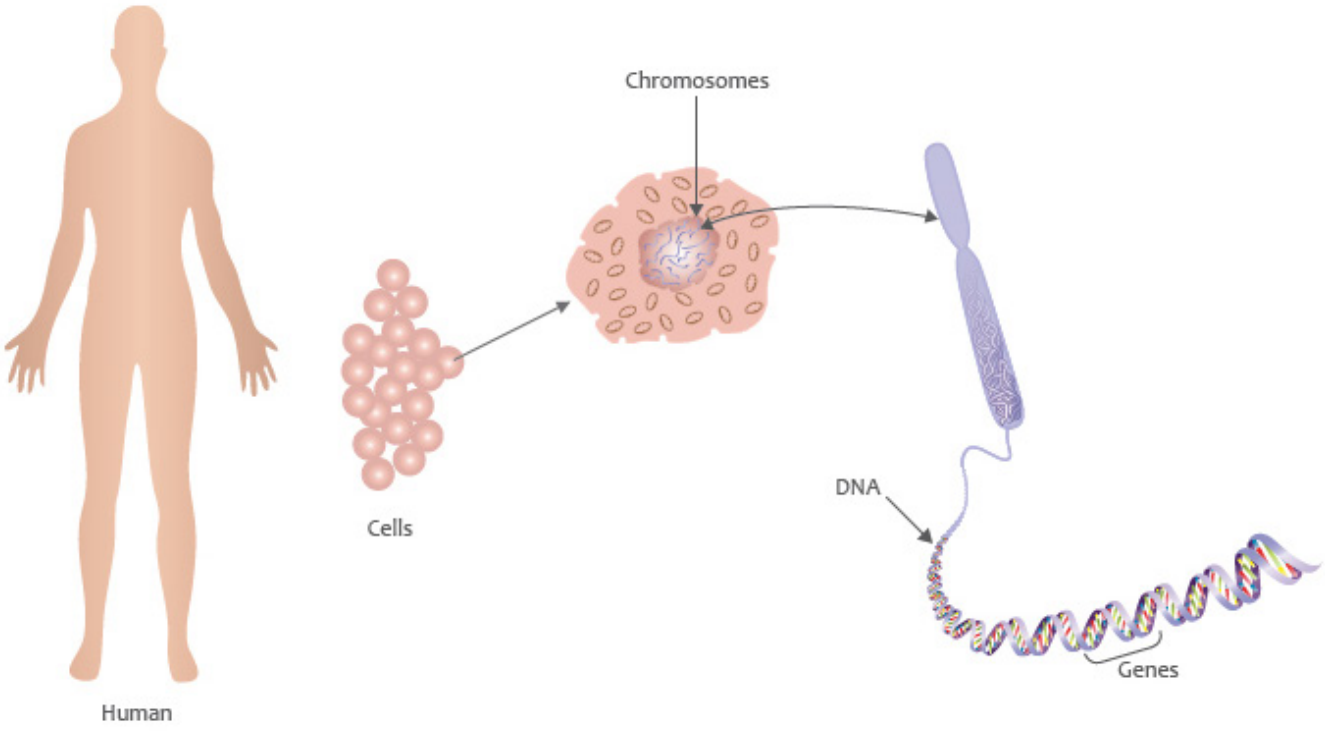

Figure from the Genomics Education Programme: www.genomicseducation.hee.nhs.uk

Inheritance

- Each person's genetic code is inherited (passed down) from their parents.
- There are 23 pairs of chromosomes – 46 in total.
- One of each pair comes from the person's biological mother and one of each pair comes from their biological father.

DNA

- DNA stands for **d**eoxyribo**n**ucleic **a**cid.
- It is a complex molecule made up of many smaller chemicals.
- Four of these chemicals, called bases, carry the genetic code. They are sometimes called the 'letters' of our genetic code.
- The letters in the human genetic code are A, C, G and T.
- These letters are arranged in a different order in each gene. For each gene to work properly, the bases must be arranged in the correct order.

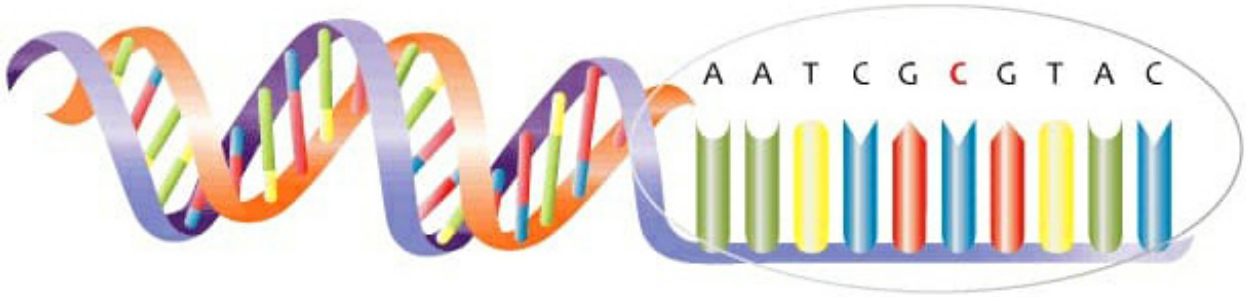

Figure adapted from the Genomics Education Programme: www.genomicseducation.hee.nhs.uk

Variants in genes

- Sometimes there is a small change in the letters of a gene. This is called a **variant**.
- A variant may result in:
  - A change in one or more letters eg 'T' instead of 'A'.
  - Extra letters (called an 'insertion').
  - Fewer letters (called a 'deletion').
- Variants are common and every person has many variants in their genetic code.
- A variant may:
  - Have no effect on the gene or may be in a gene that is not critical. This is known as a **benign variant** as it has no effect on the body.
  - Change the way the gene works, causing a health condition. This is called a **pathogenic variant** (or mutation).

Inherited and *de novo* variants

- Variants in a person's genetic code may either:
  - Be inherited (passed down) from their biological parents.
  - Develop for the first time in that person. These are called *de novo* variants.
- Scientists do not know what causes *de novo* variants – they appear to be random events that happen by chance. They are not caused by anything that a parent did or did not do, they are no one's fault.

Genetic conditions

- **'Genetic condition'** is the name given to a health condition caused by a change in one or more genes.
- Genetic conditions are present in a baby from the time of conception, even if the symptoms only come on at a later age.
- ***SCN2A*-related conditions are genetic conditions caused by a change in the *SCN2A* gene.**
- This means the condition was not caused by anything the parents did before the baby was conceived, during pregnancy or birth, or after the baby was born.

Genetic variants in SCN2A

- Most changes in the *SCN2A* gene are *de novo*.
- Many different variants in the *SCN2A* gene have been reported.
- Some of these variants cause severe developmental and epileptic encephalopathies (DEE). Individuals with DEE have epilepsy and developmental delay / intellectual disability.
- Other variants cause intellectual disability and autism spectrum disorder, but no epilepsy (See Information sheet #1 "What is SCN2A"? for more information about the different signs and symptoms)
- Variants may be broadly categorised as **gain of function** or **loss of function** (see What does gain of function and loss of function mean?), depending on the effect the variant has on the Nav1.2 sodium channel.

Considerations for future pregnancies

- If the change in the *SCN2A* gene was **inherited** from a parent, subsequent children may have the same change in the *SCN2A* gene.
  - It is recommended that individuals who are pregnant or considering more children speak with a Clinical Genetics Service.
  - Genetic testing and reproductive genetic carrier screening are options before or during a pregnancy.
- If the change in the *SCN2A* gene was **not inherited** from a parent, the likelihood of having another child with the same change in the *SCN2A* gene is much lower – about 1% (1 in 100). Again, genetic counselling is recommended if parents would like to understand that chance and screening in pregnancy options in more detail.
- Individuals with a pathogenic variant in the *SCN2A* gene have a 1 in 2 chance of passing that variant on to each pregnancy. The child may be more or less affected than the parent. It is not possible to predict the severity and ways an individual will be affected by SCN2A before they are born.

More information

- **MedlinePlus**: What is a gene?

- **Better Health Victoria**
  - Genes and Genetics
  - Genetic conditions

- **Centre for Genetics Education**
  - DNA, RNA, genes and chromosomes
  - Variations in the genetic code
  - Health conditions caused by a change in the SCN2A gene