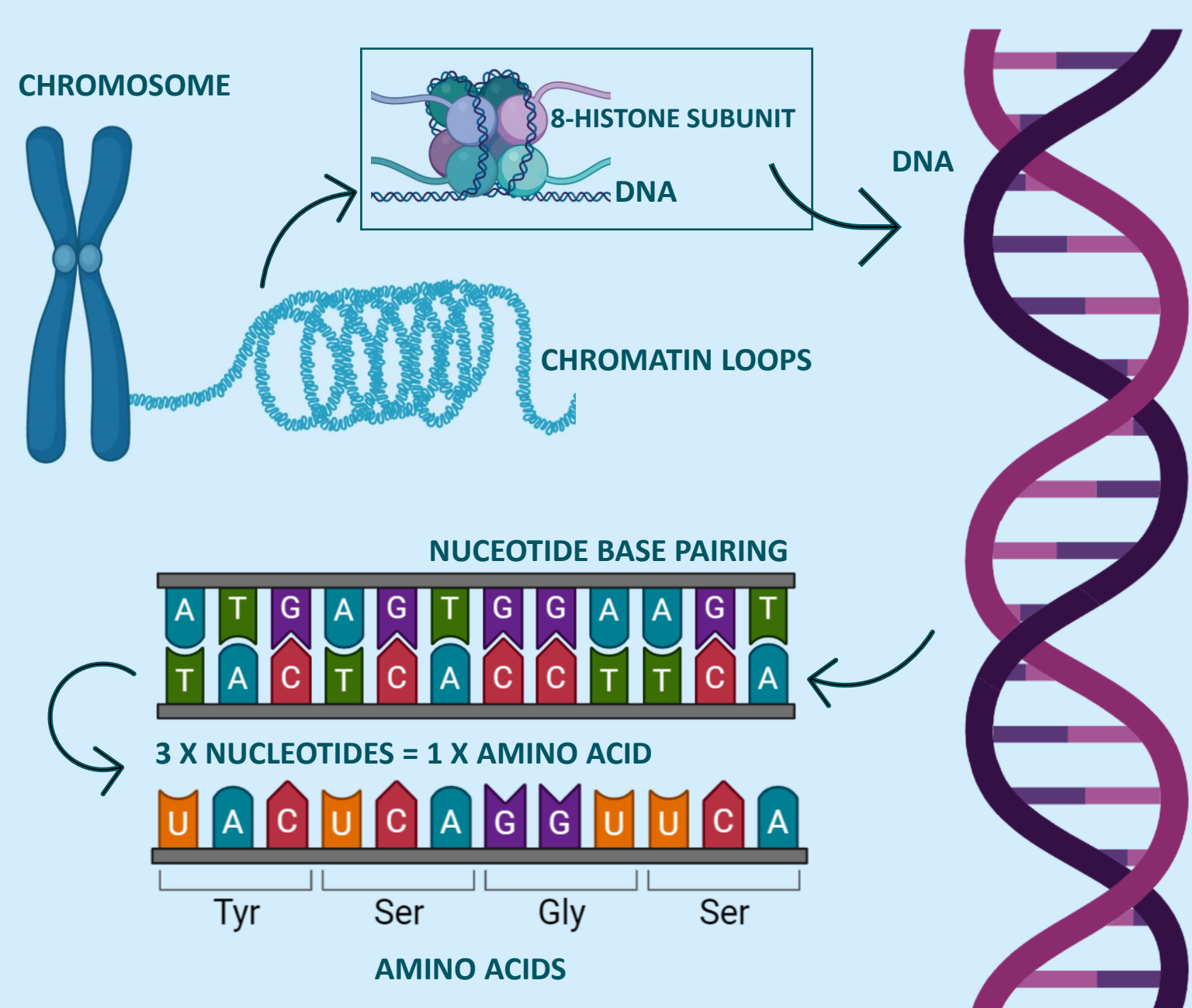


WHERE CAN GENETICS TAKE YOU?



HEALTHCARE SCIENTISTS IN THE NHS

DNA & CHROMOSOMES



CHROMOSOMES are thread-like structures located inside the nuclei of our cells.

Each chromosome is made up of **protein and deoxyribonucleic acid (DNA)**. The DNA is tightly wrapped around proteins called **HISTONES**.

This allows for a large volume of DNA to fit into a single cell.

WHAT IS THE ROLE OF HEALTHCARE SCIENTISTS?

- Healthcare scientists in the NHS play a vital role in the management, treatment and diagnosis of **genetic conditions**.
- Healthcare scientist represent **5%** of the NHS workforce.
- The work of healthcare scientists underpins **80%** of diagnoses.

WHAT CAN DNA TELL US?

Genetic disorders are linked to alterations in specific regions of DNA, called **genes**.

DNA SEQUENCING is a technique used to determine the sequence of nucleotides (As, Ts, Cs and Gs) in a piece of DNA (genes).

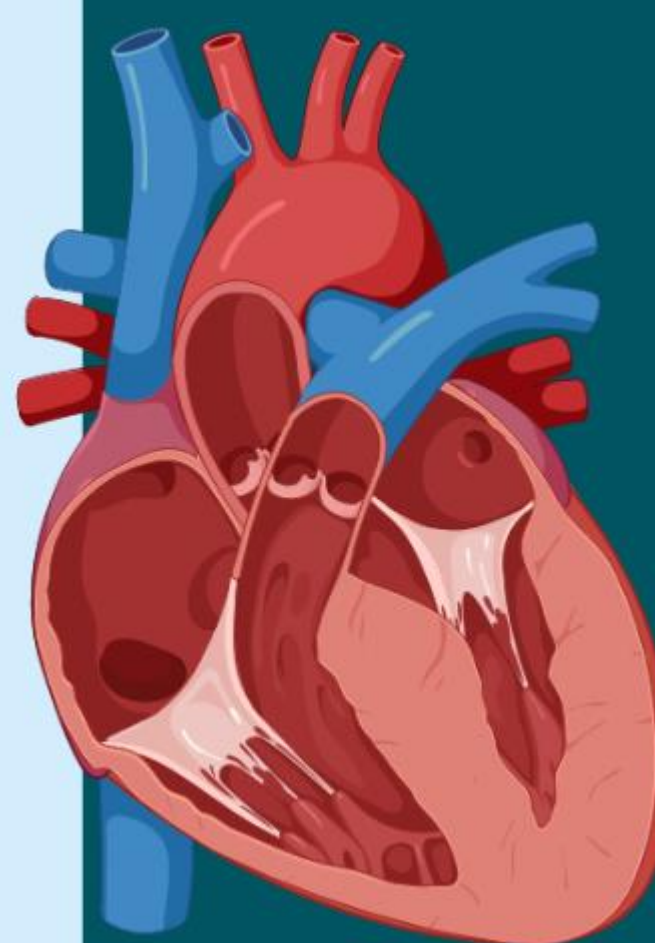


The patient's DNA sequence is **compared** to the known "normal" DNA sequence to allow identification of changes, called **variants/mutations**.

Healthcare scientists **evaluate** these changes to determine if they are **disease-causing** or part of the **natural variation** seen between humans.

CASE EXAMPLE – HYPERTROPHIC CARDIOMYOPATHY

- Hypertrophic cardiomyopathy (HCM) is a **heart condition** which causes the muscles of the heart to become **thickened/stiff**. HCM affects approximately **1 in 200** individuals in Scotland.
- Patient** – evidence of HCM was noted during a scan of the patient's heart. Whilst discussing this finding with the patient they mentioned a history of heart conditions within their family.
- DNA Sequencing** of the genes involved in heart muscle contraction was performed. A nucleotide change from T to C was identified in a gene called **MYH7**.
- Other **family members** were offered genetic testing allowing regular heart monitoring for those who also carried this DNA change.



WANT MORE CAREERS ADVICE?

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