



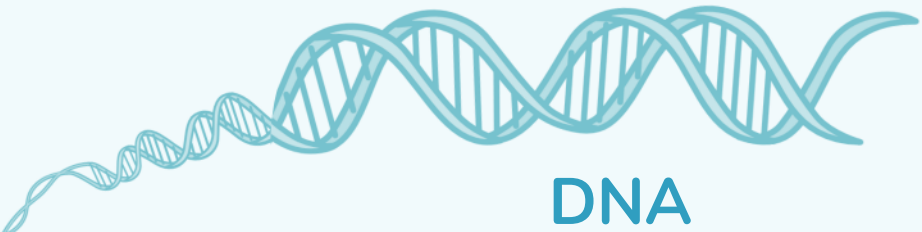
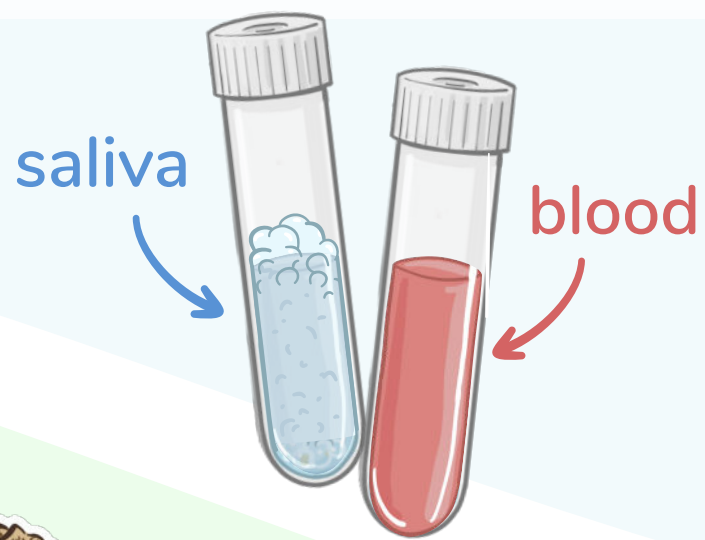
GENETIC TESTING for RARE DISEASES



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▶ WHAT is GENETIC TESTING?

Genetic testing is medical testing on a sample of **blood or saliva** that doctors use to **look for changes or variants** in a person's genes or DNA that can cause diseases.



▶ WHAT are GENES and DNA?

DNA is the genetic information in cells that is **passed from parents to their children**.

DNA contains thousands of genes with **instructions the body needs to develop and function**.

▶ WHY SHOULD I CONSIDER GENETIC TESTING for MYSELF or my CHILD?

Many rare and undiagnosed diseases are caused by gene changes. Genetic testing **can provide a diagnosis**, and this can help doctors **determine the best treatment and care team**.

A diagnosis may also be **helpful for family planning**, and it can help you find a **patient community** for information and support.



▶ WHAT GENETIC TESTS are USED to DIAGNOSE RARE DISEASES?

SINGLE GENE TESTS

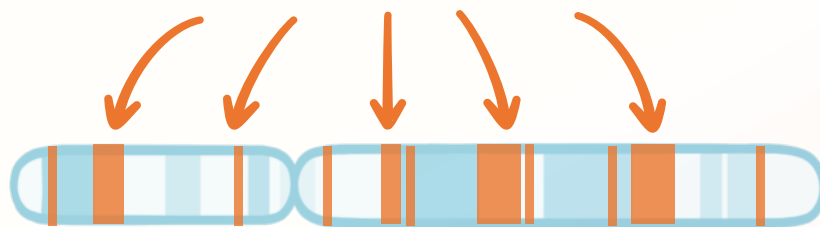
Looks for changes in a **single gene** that cause a specific disease.



For someone who has symptoms that match a specific rare disease

GENE PANEL TESTS

Looks for changes in **many genes** that cause different diseases.



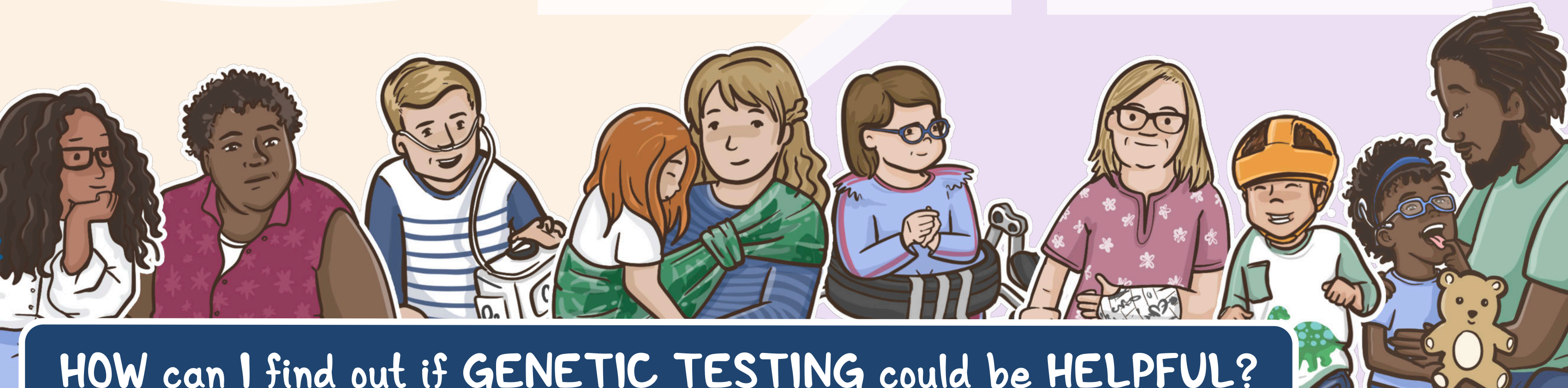
For a child who is developing slowly or has seizures because these problems can match many different rare diseases

WHOLE EXOME TESTS

Look for changes in **all of a person's genes**.



For someone who has many different symptoms that can match many different rare diseases



HOW can I find out if GENETIC TESTING could be HELPFUL?

Ask your doctor if genetic testing could provide information about the cause of your disease. You can also ask for a referral to a genetic counselor or look for a genetic counselor on the National Society of Genetic Counselors website: findageneticcounselor.nsgc.org