GENETIC TESTING for RARE DISEASES

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