UNDERSTANDING THE BENEFITS OF **GENETIC TESTING**

Answers to frequently asked questions about genes and genetic testing

"Finally finding the answer for Gabe's symptoms after all this time has been a relief. Now, we can focus on getting him the right care."

—Mario, Gabe's grandfather

BIOMARIN®

What can genes tell your doctor?

Everyone is born with a unique set of genes. They are part of what makes you different from others. You inherited your genes from your parents, and you carry traits specific to your family.[1]

Genes are the blueprint for health and development. Sometimes, genes may be abnormal or damaged, which means they may not work properly. These abnormal or damaged genes may lead to disease. Even though parents may be healthy, they may be carriers of an abnormal or damaged gene. When an abnormal or damaged gene is passed on, a child may inherit a genetic disease.[1]

Genetic research is rapidly advancing and a number of genetic diseases that were previously unknown have been identified. Each varies in types of symptoms, severity, and age of onset. Sometimes when a physician suspects a patient may have a genetic disease, he or she will refer them to either a geneticist (a doctor who specializes in genes and genetic disorders) or a genetic counsellor (a healthcare professional trained in genetics and counselling). A geneticist or genetic counselor will review health records, perform genetic testing, evaluate test results, and discuss the implications
of results for a family.[1]

What is genetic testing and what will it reveal?

Genetic testing is a medical evaluation to identify abnormalities or damage in chromosomes, genes, or proteins. Your doctor can use the test results to confirm or rule out a suspected genetic condition or help determine a person's chance of developing or passing on a genetic disorder. More than 1,000 genetic tests are currently in use, and more are being developed all the time.[1]

There are several methods that geneticists use to test for genetic conditions. Depending on the specific test, your doctor will request a sample of blood, urine, or saliva. Your doctor may be able to collect this sample in his or her office, or you may need to visit a laboratory.[1]

Although genetic testing provides important information to your doctor, no single test is likely to reveal everything that he or she wants to know. Your genetic code is like a complex puzzle that has multiple pieces and the results of one test may lead your doctor to order another test. Different tests help your doctor understand key clues and confirm or rule out certain diseases until they arrive at the right diagnosis.[2]

Why might a doctor recommend genetic testing for a child with skeletal symptoms?

There are many reasons that a doctor may recommend genetic testing for a child including, but not limited to:

- **Medical problems that are recognised as a specific genetic syndrome**[3]
  Genetic testing is performed to confirm a diagnosis. For certain conditions, testing may help identify the specific type or severity of a genetic illness, which can help your doctor prescribe the most appropriate treatment.
- **Growth at a very different rate than his or her peers**[4]
  Genetic testing may be able to identify the cause and make a diagnosis.
- **Development of skeletal symptoms at an earlier age or more severe than would be expected**[5,6]
  Children may develop skeletal symptoms, like knock knees, as they grow. While not all skeletal symptoms are reason for concern, some may be caused by genetic disease. If these symptoms are caused by genetic disease, they may appear earlier in life and be more severe than typical cases.
- **Loss of the ability to perform certain activities they used to be able to do**[5,7,8]
  If a child begins to weaken and lose the ability to do certain things (for example, becoming too weak to climb stairs, walk around the house/yard, or keep up with friends at play), a genetic illness may be responsible. Genetic testing may help to identify the cause and help your doctor make a diagnosis.
- **Reduced exercise capacity**[7]
  As a child gets older, their bones and organs typically develop at a rate that allows them to move faster and/or exhibit greater endurance. When children fall behind on developmental milestones like exercise capacity, genetic testing may identify the cause and help doctors make a diagnosis.

What are the benefits of genetic testing?

There are a number of important benefits. When considering genetic testing, you are taking an active step in[1,9,10]:

Confirming or ruling out a diagnosis

The diagnostic journey can be time-consuming, costly, and emotionally draining. Shortening the time to diagnosis can speed up access to care.

Receiving care specific to a diagnosis

Determining the genetic basis for your disease can help your doctor provide optimal care, including possible medications or surgical planning.

Gaining insurance approval for equipment or devices

With a confirmed diagnosis, patients may become eligible for items that can help make day-to-day life easier, like wheelchairs, special car seats, or an adapted car.

Identifying potential clinical trials

There are numerous clinical trials for investigational drugs, and results of genetic testing may uncover one that is relevant to a patient's diagnosis and determine eligibility. Early and prompt diagnosis may increase opportunities to enroll.

Identifying risk of disease for family members

Genetic counsellors can educate your family about inherited diseases and family planning.

Connecting with disease-specific resources and advocacy organisations

Patients and families can access valuable support and resources that are available for the patient's specific diagnosis.

Talk to your doctor about how genetic testing can help confirm or rule out a diagnosis.

References: 1. Lister Hill National Center for Biomedical Communications, US National Library of Medicine. Genetics Home Reference: *Help Me Understand Genetics*. Published November 8, 2016. Available at: https://ghr.nlm.nih.gov/primer. Accessed November 10, 2016. **2.** Frequently asked questions about genetic testing. NIH—National Human Genome Research Institute. Available at: https://www.genome.gov/19516567/faq-about-genetic-testing/. Accessed November 10, 2016. **3.** Genetic testing. KidsHealth®. Available at: http://kidshealth.org/en/parents/genetics.html. Accessed November 9, 2016. **4.** Hendriksz CJ, Harmatz P, Beck M, et al. Review of clinical presentation and diagnosis of mucopolysaccharidosis IVA. *Mol Genet Metab*. 2013;110:54-64. **5.** Tomatsu S, Alméciga-Díaz CJ, Barbosa H, et al. Therapies of mucopolysaccharidosis IVA (Morquio A syndrome). *Expert Opin Orphan Drugs*. 2013;1(10):805-818. **6.** Montaño AM, Tomatsu S, Gottesman GS, Smith M, Orii T. International Morquio A registry: clinical manifestation and natural course of Morquio A disease. *J Inherit Metab Dis*. 2007;30:165-174. **7.** Wood TC, Harvey K, Beck M, et al. Diagnosing mucopolysaccharidosis IVA. *J Inherit Metab Dis*. 2013;36:293-307. **8.** Hendriksz CJ, Berger KI, Giugliani R, et al. International guidelines for the management and treatment of Morquio A syndrome. *Am J Med Genet A*. 2015;167A:11-25. **9.** Institute of Medicine of the National Academies. *The Future of Disability in America*. Washington, DC: The National Academies Press; 2007. doi: 10.17226/11898. **10.** US National Institutes of Health (NIH). NIH Clinical Research Trials and You: Why Should I Participate in a Clinical Trial? NIH website. Reviewed October 27, 2015. Available at: https://www.nih.gov/health-information/nih-clinical-research-trials-you/why-should-i-participate-clinical-trial. Accessed November 10, 2016.

BioMarin Pharmaceutical Australia Pty Ltd
119 Willoughby Rd, Crows Nest, NSW 2065, Ph +61 2 9967 8662,
Medical Information 1800 387 876, medinfoasia@bmrn.com

BIOMARIN®