COMMON QUESTIONS AND ANSWERS ABOUT DUCHENNE MUSCULAR DYSTROPHY (DMD)

We Speak DUCHENNE®

DUCHENNE & DYSTROPHIN

Q When you have Duchenne, what is happening in the body?

A People with Duchenne have a genetic mutation that prevents their body from making usable dystrophin, an essential protein for muscle function. As a result, their muscles—including the heart muscle—become weaker over time.

Q What role does dystrophin play in Duchenne?

A Dystrophin is part of a group of proteins that work together to stabilize and protect muscles as they work. Because people with Duchenne can't make enough dystrophin, their muscles become damaged. This leads to muscle weakness and loss of function.

Q Are there ways to increase dystrophin in people with Duchenne?

A For eligible people with Duchenne, dystrophin can be increased using two different types of therapy: exon-skipping therapy and gene therapy. Each therapy, through different methods, allows the person's body to create a shorter, functional dystrophin protein.

CARDIOMYOPATHY IN DUCHENNE

Q What is cardiomyopathy?

A Cardiomyopathy is a general term for any disease of the heart muscle. In Duchenne, symptoms of cardiomyopathy may include excessive fatigue, irregular heartbeat, weight loss, abdominal pain, vomiting, and respiratory changes like shortness of breath. It's important to know these symptoms because almost everyone with DMD will experience cardiomyopathy at some point.

Q How is cardiomyopathy managed?

A Currently, there are various cardiac medications that are prescribed to manage cardiomyopathy. Symptoms can be managed through a combination of medications and monitoring. Talk to your medical team and cardiologist to determine the best combination of care.

Q When should I talk to my child's doctor about cardiomyopathy?

A For people with Duchenne, it's best to start monitoring the heart as early as possible. In fact, delaying therapies could negatively impact outcomes. Talk to your child's doctor about consulting a cardiologist for additional support.

TREATMENTS

Q What is the difference between exon-skipping therapy and gene therapy?

A **Exon-skipping therapy** helps the person's body bypass their existing genetic mutation. It results in a protein that's typically 84% to 97% as long as the dystrophin protein made by someone without DMD. **Gene therapy** inserts a replacement micro-dystrophin gene to make a protein that's 32% to 40% as long.

Q Should I talk to my doctor about treatments that could increase dystrophin?

A Yes, your doctor is always the best source of guidance when making treatment decisions. They can help you decide whether a treatment that could increase dystrophin may be an option, and when would be the best time to begin. While these therapies can be started at different ages, it's typically best to start as early as is appropriate. This is because therapies that increase dystrophin may be most effective before muscles weaken.

Q How can I find out about currently available therapies for Duchenne?

A Being eligible for, or "amenable to," a therapy may depend on your child's specific gene mutation. A simple genetic test may help determine your treatment options. Ask your child's doctor to learn more about all available therapies for Duchenne.

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