Duchenne muscular dystrophy

Support organisations around the world understand the heartache and angst that parents feel with this diagnosis, and the support that is needed thereafter. By Miriam Rodrigues.

Duchenne muscular dystrophy (DMD or Duchenne) is a difficult, complex diagnosis to understand and manage. This is not a world that anyone enters willingly.

Support organisations around the world including Action Duchenne, The US-based Muscular Dystrophy Association, Parent Project Muscular Dystrophy (PPMD), Treat-NMD, World Duchenne Organization (UPPMD) as well as Muscular Dystrophy New Zealand and many others all understand the heartache and angst that parents feel with this diagnosis, and the support that is needed thereafter.

As you journey through this diagnosis, it is important to all of us that you, or your child, receive the very best in care, support and resources.

For this reason, we recommend accessing the *2018 Duchenne Family Guide*. The guide summarises the results of the updates for the medical care of Duchenne muscular dystrophy.

Both the original effort, as well as the updated guidelines, were produced in collaboration with patient advocacy groups and the TREAT-NMD network. The original documents are published in the journal *Lancet Neurology* and are freely available online, as is the current Family Guide.

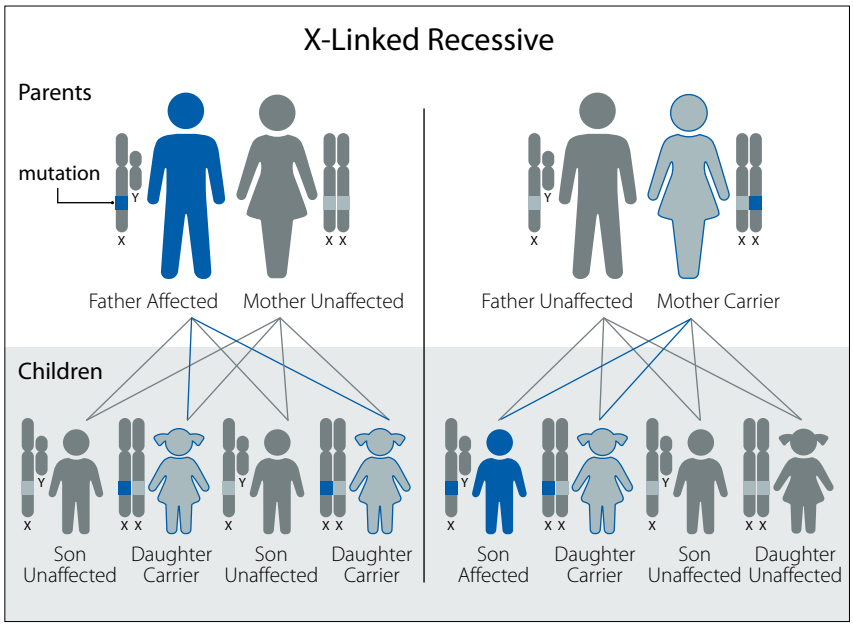
Duchenne is one of a spectrum of muscle diseases known as "dystrophinopathies." Dystrophinopathies result from the absence of the muscle protein "dystrophin" and range from the more severe symptoms that you see in Duchenne muscular dystrophy to the milder, yet more variable, condition Becker muscular dystrophy.

We recommend accessing the 2018 Duchenne Family Guide. The guide summarises the results of the updates for the medical care of DMD.

Dystrophin protein is needed for healthy muscle cell function. A fault in the genetic code for this protein (in the dystrophin gene) means that there is little or no protein manufactured and the muscle cells are easily damaged.

This damage builds up over time and leads to the progressive (getting worse with time) muscle weakness experienced in DMD.

The dystrophin gene is present on the X chromosome. Females have



Mode of inheritance.

two copies of the X chromosome and so are rarely severely affected by dystrophinopathies.

Males have only one X chromosome and so a mistake in the dystrophin gene in a male will have consequences as there isn't a second copy of the gene to provide back up.

In about one third of boys with Duchenne muscular dystrophy the mistake in their dystrophin gene has happened for the first time in them.

In the remaining two thirds of boys with DMD their mothers are carriers of the altered dystrophin gene.

Although carriers are not usually severely affected there are some physiological effects sometimes caused by being a carrier. These effects include increased risk of heart problems and so it's important that carrier women are identified as such and provided with appropriate cardiac monitoring.

A diagnosis of DMD and the identification of mum as a carrier can also have implications for other family members. It's very important that genetic counselling is provided as soon as possible.

Duchenne is a condition that changes very slowly over time. *The Family Guide to DMD* separates key stages of Duchenne to help you anticipate recommendations for care. Although these stages can be somewhat blurred in distinction, you may find it useful to use the stages to identify the kind of care and interventions that are recommended at any particular time and what you should expect of your care team at that time.

Currently, in the absence of new-born screening, most children with Duchenne remain undiagnosed during the pre-symptomatic stage (when children show few, if any, symptoms) unless there is a family history of the condition, or unless blood tests are done for other reasons.

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Symptoms such as delayed walking and crawling or speech are present, but are typically subtle and are often unrecognised at this stage.

Parents are often the first to notice differences in their child's development, the first to ask questions and the ones requesting further testing to explain perceived developmental delays.

Psychosocial and emotional support is extremely important when a new diagnosis of Duchenne is confirmed.

Receiving a diagnosis of Duchenne is tremendously difficult. There are so many unanswered questions, and families often feel alone and overwhelmed, with few places to turn.

Primary care providers, neuromuscular specialists, Muscular Dystrophy New Zealand, and the online Duchenne community can be especially helpful during this time, facilitating connections to appropriate care that can help to provide the resources, information and support parents need to help themselves and their families. Ⓡ

Muscular Dystrophy New Zealand has many resources available to families with Duchenne muscular dystrophy. Kiwi Kids with DMD is a Facebook group for parents of children with Duchenne muscular dystrophy.