

# FEMALES

WITH DYSTROPHINOPATHIES

## FAMILY FACTSHEET

### ABOUT FEMALES WITH DYSTROPHINOPATHIES

#### 1 WHAT IS MEANT WITH FEMALES WITH DYSTROPHINOPATHIES?

- Dystrophinopathies, such as Duchenne muscular dystrophy (DMD) and Becker muscular dystrophy (BMD), primarily affect males due to the inheritance pattern linked to the X chromosome.
- However, females can also be affected by dystrophinopathies, albeit with lower frequency and different manifestations.
- Affected women possibly develop symptoms due to inactivation of an X chromosome.
- However, results presented on this are inconclusive and so, knowing the degree of X chromosome inactivation cannot predict whether or how severely a female will be affected.
- Not only the mothers and sisters of someone with DMD are at risk, but also aunts and cousins on the mother's side of the family.

#### 2 WHAT ARE SIGNS IN FEMALES WITH DYSTROPHINOPATHIES?

- Females who carry a DMD gene mutation can present with a spectrum of features from no to severe symptoms or abnormalities on blood or cardiac testing (the majority).
- Some will have no symptoms but will have a raised blood creatine kinase.
- Some will develop cardiomyopathy and some may develop muscle pain and/or muscle weakness.
- In some women, muscle weakness and cardiac features can have a significant impact upon quality of life.



#### 3 CARE FOR A FEMALES WITH DYSTROPHINOPATHY

- Minimum standards of care should include full genetic testing of all females at risk of being a carrier of a DMD gene mutation.
- All females shown to be carriers of a DMD gene mutation should be referred to a neuromuscular and cardiology specialist for further assessment.
- There is a clear need to increase awareness of the potential for developing symptoms in muscle and/or heart amongst women who carry a DMD gene mutation.

#### 4 IS THERE A CURE FOR FEMALES WITH DYSTROPHINOPATHIES?

- While there is no cure for females with dystrophinopathies, there are interventions and treatments available to manage the symptoms and slow the progression of the disease.
- There is a pressing need to step-up the pace of research and systematic data collection as there are many gaps in research and our current understanding of the condition.



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