ABOUT FEMALES WITH DYSTROPHINOPATHIES

WHAT ARE FEMALES WITH DYSTROPHINOPATHIES?

1. 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.
2. However, females can also be affected by dystrophinopathies, albeit with lower frequency and different manifestations.
3. Affected women possibly develop symptoms due to inactivation of an X chromosome.
4. However, results presented on this are conflicting and so, knowing the degree of X chromosome inactivation cannot predict whether or how severely a female will be affected.
5. 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.

WHAT ARE SIGNS IN FEMALES WITH DYSTROPHINOPATHIES?

1. 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).
2. Some will have no symptoms but will have a raised blood creatine kinase.
3. Some will develop cardiomyopathy and some may develop muscle pain and/or muscle weakness.
4. In some women, muscle weakness and cardiac features can have a significant impact upon quality of life.

CARE FOR A FEMALES WITH DYSTROPHINOPATHY

1. Minimum standards of care should include full genetic testing of all females at risk of being a carrier of a DMD gene mutation.
2. All females shown to be carriers of a DMD gene mutation should be referred to a neuromuscular and cardiology specialist for further assessment.
3. 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.

IS THERE A CURE FOR FEMALES WITH DYSTROPHINOPATHIES?

1. 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.
2. 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.