Muscular Dystrophy New Zealand

Updated: September 2016

Scapuloperoneal Myopathy

Scapuloperoneal Myopathy or Myopathic Type Scapuloperoneal Syndrome.

About the Condition:

Scapuloperoneal myopathy causes weakness and wasting of particular muscles. Areas first affected are the shoulder blade area (scapula) and the small leg muscle groups below the knee (peroneal). In some cases facial muscles can also be affected. Stiffness in the affected muscles, muscular cramps and shrinkage may occur in individuals with Scapuloperoneal Myopathy. This condition can begin in childhood or adulthood with varying progression rates.

Inheritance:

This is a rare genetic disorder that is inherited as an autosomal dominant trait therefore, a positive family history of the condition is a risk factor for developing Scapuloperoneal Myopathy. It can also be the result of new spontaneous mutations. It equally affects males and females

It has been suggested that this condition could be a variant of other neuromuscular conditions however genetic analysis demonstrated linkage particularly to the MYH7 gene on chromosome 14.

Diagnosis:

Testing required to achieve a diagnosis may include: obtaining a detailed personal and family medical history, a complete physical exam, blood tests to detect abnormal levels of proteins, such as pyruvate kinase, creatine kinase, and lactic dehydrogenase (LDH) and genetic testing to determine the mutation in the defective gene.

Treatment and Management:

Therapeutic exercise and physical therapy could benefit individuals with this condition, as well and the use of symptomatic and supportive medication such as corticosteroids. As the disorder progresses, a multitude of complications may result because of muscles weakening, particularly respiratory complications. Spirometry exercises have been shown to help and special respiratory care may be needed.

For vaccination recommendations and respiratory care, please refer to the MDA website.

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Genetic counseling may also be beneficial to the affected individuals and their families.

For further information on this condition you can also refer to the following websites/organizations:

- Genetic and Rare Diseases (GARD) Information Center
  - Website: http://rarediseases.info.nih.gov/GARD/
- Muscular Dystrophy Association
  - Website: http://www.mda.org/
- NIH/National Institute of Arthritis and Musculoskeletal and Skin Diseases Website: http://www.niams.nih.gov/
- OMIM (Online Mendelian Inheritance in Man) Website: http://www.omim.org/entry/181430