The Information Standard

Factsheet

Muscular Dystrophy Campaign

Version: 3
Date published: March 2002
Original author: Dr Reghan Foley, Dr Stephanie Robb and Prof Francesco Muntoni, Dubowitz Neuromuscular Centre
Updated: May 2012
Updated by: Dr Reghan Foley
Date of review: May 2012
Available online: www.muscular-dystrophy.org/factsheets

Bethlem myopathy

What is Bethlem myopathy?

Bethlem myopathy falls under the category of congenital muscular dystrophies, within a sub-type known as the 'collagen VI-related myopathies'. Bethlem myopathy has the following specific features:

- the joints of the hands develop 'contractures' or 'tightness' at the level of the fingers, and the feet develop 'contractures' or 'tightness' of the Achilles tendons
- a variable degree of muscle weakness, especially affecting the legs and to a lesser extent the arms, develops and slowly progresses over time
- respiratory muscles are more mildly affected, and problems related to respiratory insufficiency are very rare and limited to the late adult years.

What causes Bethlem myopathy?

Mutations in at least three genes are responsible for Bethlem myopathy. These genes are called *COL6A1, COL6A2* and *COL6A3,* and are responsible for the production of a protein called collagen VI which is required to maintain the structure of the muscles.

How is Bethlem myopathy diagnosed?

The diagnosis of Bethlem myopathy is usually suspected from an individual's medical history and examination. The specific diagnosis, however, is generally made by looking at a piece of muscle or sometimes skin (muscle and skin biopsy). Before doing a muscle biopsy a few other tests may be done.

A **blood test** can measure the level of a muscle protein (called creatine kinase or CK). In patients with Bethlem myopathy, the CK level is either normal or mildly elevated.

Muscle biopsy involves taking out a small piece of muscle, usually from the thigh. This is then studied under the microscope, looking for signs which might indicate the type of abnormality which characterises the condition. It is also possible to look at the expression of collagen VI in the muscle under the microscope. There are specific 'markers' or 'tags' which can detect whether collagen VI is normally present or reduced.

Genetic tests looking for abnormalities in one of the three genes responsible for Bethlem myopathy (*COL6A1, COL6A2* and COL6A3) are now available in the UK in a specific nationally designated central laboratory (NSCT centre for CMD and congenital myopathies, London) and can provide a definitive diagnosis.

Is there a treatment or cure?

At present there is no cure for Bethlem myopathy but there are ways, described below, of helping to alleviate the effects of the condition. Research into the congenital muscular dystrophies is nevertheless developing, and it is likely that clinical trials will start in the not-too-distant future.

Physiotherapy is one of the main forms of help. An initial physiotherapy assessment at the time of the diagnosis should be followed by an exercise programme and regular check-ups. The main aim of physiotherapy is to keep the muscles as active as possible and to prevent or slow the progression of joint contractures (tightness). Individuals with Bethlem myopathy are encouraged to remain as active as possible. Swimming is a particular good form of exercise. Physiotherapists can also help provide advice on orthoses, walking aids and a wheelchair, when necessary.

Occasionally surgery to release the Achilles tendons can help individuals with Bethlem myopathy to stand and walk more easily.

Children and adults with Bethlem myopathy should ideally regularly visit a specialist neuromuscular clinic, with access to physiotherapy, orthotic, respiratory, orthopaedic and genetic specialists as needed.

What is the prognosis?

The first symptoms of Bethlem myopathy can present at any time from birth through to adulthood and are variable. In childhood these symptoms can be hypotonia (floppiness) and joint laxity (double jointness), muscle weakness, delayed achievement of motor milestones such as sitting unaided or walking. Joint contractures are common in Bethlem myopathy, typically affecting the wrists, fingers, elbows and Achilles tendons of the feet. Some stiffness of the spine can also develop over the years.

Other symptoms such as poor stamina/exercise intolerance and difficulty rising from a seated position and climbing stairs are related to the proximal muscle weakness evident in Bethlem myopathy.

As collagen VI is found in the skin as well as the muscles, individuals with Bethlem myopathy have a tendency for scars which heal slowly or become thickened and elevated (keloid formation). The surfaces of the arms and legs can feel ‘rough’ or ‘bumpy’ due to ‘keratosis pilaris’ which commonly occurs in the skin of patients with Bethlem myopathy.

The main complications in Bethlem myopathy are the progression of joint contractures (tightness), increased proximal limb (hip and shoulder girdle) weakness and, to a much lesser extent, a decline in lung function. Physiotherapy and a regimen of stretching can help to prevent or slow the progression of joint contractures. A proportion of adults will develop a dependence on walking aids (cane, crutches or wheelchair) for walking outdoors.

Lung function in individuals with Bethlem myopathy should be monitored. If indicated (depending on the respiratory function values), overnight sleep studies could be required. Rarely, adults with Bethlem myopathy develop sleep hypoventilation for which night-time non-invasive ventilation (NIV) will be necessary.

Other related publications

- Congenital muscular dystrophies
  - MDC1A (merosin-deficient congenital muscular dystrophy)
  - SEPN1-related myopathy
  - Ullrich congenital muscular dystrophy
- Carrier detection tests and prenatal diagnosis of inherited neuromuscular conditions
- Inheritance and the muscular dystrophies
- Muscle biopsies
- Surgical correction of spinal deformity in muscular dystrophy and other neuromuscular disorders

The friendly staff in the care and support team at the Muscular Dystrophy Campaign's London office are available from 8.30am to 6pm Monday to Friday to offer free information and emotional support. If they can't help you, they are more than happy to signpost you to specialist services close to you, or to people who can help.

Contact our Freephone helpline on 0800 652 6352 or info@muscular-dystrophy.org

Disclaimer