Myotonic Dystrophy

Description

Myotonic dystrophy, also known as Dystrophia myotonica, is the most commonly diagnosed form of muscular dystrophy in adults, with prevalence estimated to be at least 1 in 8000 world wide. There are 2 forms of myotonic dystrophy (DM1 and DM2) which have subtly different symptoms. Symptoms associated with the more common form, DM1, are often more severe than those seen for DM2. The disease involves multiple body systems and results in variable symptoms which are progressive and can include:

- muscle weakness and wasting
- myotonia (sustained muscle contraction) – this is the characteristic feature which is not found in other forms of muscular dystrophy and is often used for diagnosis.
- cardiac arrhythmia
- breathing problems
- daytime sleepiness
- digestive problems and problems swallowing
- cataracts
- insulin resistant diabetes.
- damage to cortical regions of the brain leading to cognitive impairment occurs in some cases

Generally people will have only a sub-set of these symptoms. People with myotonic dystrophy also have an increased risk of complications with general anaesthesia.

While symptoms typically become apparent in adult life, there is also a congenital form of DM1 generally associated with more severe symptoms which start at birth. In some cases, parents with a milder form of the disease can have children with the congenital form. Congenital forms of DM2 have not been observed.

Myotonic dystrophy is a genetic disease that exhibits dominant symptoms: that is, you only need to inherit the altered gene from one parent in order to develop the disease. The altered gene can be inherited from either parent. One unusual feature of genetic inheritance in DM is that children with the congenital form of DM almost always have a mother with the adult-onset form. The adult onset-form can be inherited from either parent.

DM1 and DM2 are caused by genetic changes in different genes, but the type of change is the same in both cases: it is an increase in the size of a repeating section of the gene. In the case of DM1, this gene is on chromosome 19 while the gene which is altered in DM2 is on chromosome 3. The size of this repeating section can change from generation to generation and it is this property which is responsible for the unusual inheritance pattern of the disease.

Treatment

Despite research providing a clear understanding of the mutation responsible and as a consequence definitive diagnostic tests being made available, there is currently no cure for myotonic dystrophy. Research is now needed to understand the steps leading from the

altered gene to the clinical symptoms so that drugs can be identified to interfere with these steps and in so doing prevent or reduce the impact of the disease symptoms.

In the absence of effective treatment or cure, symptoms can be managed by a range of means which may include:

- medications for digestive problems
- canes and walkers to assist with mobility
- surgery to remove cataracts
- medication to help with myotonica
- medication of pace-maker for heart problems
- nocturnal breathing assistance may be required later in life

Early diagnosis can assist in monitoring symptoms and determining the best treatment course as symptoms arise.

In congenital cases, babies may have significant problems with breathing and swallowing which require special attention: for example artificial ventilation and a feeding tube. In these cases, myotonia will usually not appear until later on.

Prognosis

Myotonic dystrophy is a progressive disease however, since there are frequently no symptoms until adult life, in many cases people are able to be independent for most of their lives. As symptoms worsen, people may require additional medical and personal support.

Many infants with congenital myotonic dystrophy are now able to survive to childhood with specialist treatment, however the progressive nature of the disease means that symptoms tend to worsen into childhood, resulting in health problems which can be potentially fatal. Along with swallowing, breathing and mobility issues, children with congenital DM may have intellectual disabilities and vision, speech and hearing problems. In some cases, muscle symptoms may improve in childhood, however children with congenital DM will develop the adult form later in life.

Support services/further information:

http://www.myotonic.org/
Myotonic Dystrophy Foundation (international)
http://mdaustralia.org.au/
Muscular Dystrophy Foundation Australia

Link to latest Australian research papers (within last 5 years) (PubMed database)

Pubmed references