is recommended. Furthermore, annual ECG and if possible echocardiogram or cardiac MRI or 24-hour Holter monitoring to detect/monitor cardiac conduction defects and cardiomyopathy is endorsed. An annual measurement of fasting serum glucose concentration, glycosylated haemoglobin level, thyroid hormone levels, and serum testosterone is recommended.

For further details, please see and go to:

2019 Consensus-based care recommendations for adults with myotonic dystrophy type 2:
https://cp.neurology.org/content/neurclinpract/early/2019/04/24/CPJ.0000000000000645.full.pdf
or **https://bit.ly/38vnmZ5**

Other publications available from the Myotonic Dystrophy Support Group:

- Anaesthesia and Sedation for patients with Myotonic Dystrophy
- Basic Information for Midwives
- Bowel Problems in Myotonic Dystrophy
- Congenital Myotonic Dystrophy
- Excessive Daytime Sleepiness and Myotonic Dystrophy
- Facts for patients, family members and professionals
- Just Diagnosed
- Myotonic Dystrophy and the Brain
- Myotonic Dystrophy and the Eye
- Myotonic Dystrophy Support Group
- Relatives Information
- Swallowing Difficulties in Myotonic Dystrophy
- The Heart and Myotonic Dystrophy
- Why do we get new families with Myotonic Dystrophy?

Myotonic Dystrophy
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Myotonic Dystrophy
SUPPORT GROUP

Myotonic Dystrophy Type 2

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Myotonic Dystrophy

What is Myotonic Dystrophy Type 2?

Myotonic dystrophy type 2 (DM2) is an inherited neuromuscular condition caused by an abnormality in the CNBP gene, which is located on chromosome 3. The condition is caused by a CCTG expansion (enlargement or repeat) in the CNBP gene. The number of CCTG repeats in expanded alleles ranges from approximately 75 to more than 11,000, with a mean of approximately 5,000 repeats. There is no correlation between the repeat numbers and the severity of this disease.

How is the condition inherited?

Myotonic dystrophy type 2 affects both men and women. It is inherited as a dominant gene, which means that an affected adult (male and female) has a 50% chance of passing the condition on to each of their offspring. No severe congenital form occurs.

What are the symptoms of Myotonic Dystrophy Type 2?

Myotonic dystrophy type 2 is characterised by myotonia and muscle dysfunction such as proximal and axial weakness, myalgia, and stiffness, and less commonly by cardiac conduction defects, iridescent posterior subcapsular cataracts, insulin-insensitive type 2 diabetes mellitus, and other endocrine failures.

Although involuntary muscle contraction with delayed relaxation, named myotonia, has been reported during the first decade of life, the symptom onset is typically between the third to fifth decades of life. Frequently reported are fluctuating or episodic muscle pain and proximal and axial weakness of the neck flexors and the hip flexors and finger flexors. During the disease course weakness occurs in the elbow extensors and the hip flexors and extensors and finger flexors. Facial weakness and weakness of the ankle dorsiflexors are more rare issues. Myotonia may cause severe symptoms in some patients. In up to 20% of patients, calf hypertrophy in combination with brisk reflexes and restless legs syndrome is remarkable.

How can the condition be diagnosed?

Many investigations, including nowadays in rare situation a muscle biopsy are often necessary. A blood test to look for abnormality in the CNBP gene will confirm the diagnosis of Myotonic dystrophy type 2. The diagnosis is most easily made by a blood test to look for an expansion in the CNBP gene. This will confirm the diagnosis of Myotonic dystrophy type 2 (DM2). However DM2 is not common, and not well known. In many countries individuals with DM2 may have many investigations, including muscle biopsy, before the diagnosis is reached.

How is the condition treated?

There is no cure for myotonic dystrophy type 2, but much can be done to help the adults and improve the long-term outcome. Routine physical activity appears to help maintain muscle strength and endurance and to control myalgia. Myotonia rarely requires treatment but mexilitene or lamotrigine may be beneficial. Ankle-foot orthoses, wheelchairs, or other assistive devices are helpful as needed for weakness. Medications used with some success in myalgia pain management include mexilitene, gabapentin, pregabalin, nonsteroidal anti-inflammatory drugs, low-dose thyroid replacement, low-dose steroids (e.g., 5 mg prednisone on alternate days), and tricyclic antidepressants.

Removal of cataracts that impair vision is very helpful. Use hearing aids, defibrillator placement for those with arrhythmias; and hormone substitution replacement therapy for endocrine dysfunction. e.g. hypothyroidism or hypogonadism in males. Prompt treatment of hypothyroidism and vitamin-D insufficiency is advised to reduce secondary weakness and myotonia.

Anaesthetic issues

It is important to be aware that anaesthetics can be risky in anyone who has myotonic dystrophy, independent of whether they are mildly or severely affected. Anaesthetic risk may be increased and therefore perioperative surveillance and assessment of cardiac and respiratory function before and after surgery are recommended.

What annual surveillance for myotonic dystrophy type 2 is recommended?

An annual evaluation with neurologist or neuromuscular / rehabilitation physician, occupational therapist, and/or physiotherapist

continued overleaf.....

MD

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