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Myotonic Dystrophy

Patient education guide · Nerve & Muscle · Madison Neurology LLC

What is it?

Myotonic dystrophy is the most common adult-onset muscular dystrophy — an inherited condition causing myotonia (muscles that grip and can't relax quickly — trouble releasing a handshake) plus progressive muscle weakness. It is a multisystem disease: it also affects the heart (rhythm problems), eyes (cataracts), hormones, gut, and sleep, so it requires coordinated care beyond muscles alone.

How does it happen?

It is caused by expanded DNA repeats — in the DMPK gene (type 1) or CNBP gene (type 2) — passed in an autosomal dominant pattern (50% transmission). In type 1, the repeat can expand between generations, causing earlier, more severe disease in children ('anticipation').

Common symptoms

- Myotonia: delayed muscle relaxation — slow to release a grip, stiffness after rest
- Progressive weakness of face, neck, hands (grip), and feet (foot drop)
- Excessive daytime sleepiness and fatigue
- Early cataracts; frontal balding; swallowing and gut problems (constipation)
- Heart rhythm abnormalities — potentially serious, requiring periodic ECG/monitoring

How is it diagnosed?

The pattern (myotonia plus characteristic weakness and family history) is distinctive; genetic testing confirms the diagnosis definitively and distinguishes type 1 from type 2. EMG shows the classic 'dive-bomber' myotonic discharges. Family members should be offered counseling and testing.

How is it treated?

No disease-modifying drug exists yet (several are in trials), but systematic care transforms outcomes: annual cardiac screening (ECG, Holter; pacemakers when indicated) prevents the condition's main danger; mexiletine can reduce bothersome myotonia; modafinil helps daytime sleepiness; physical therapy, braces (ankle-foot orthoses for foot drop), and fall prevention maintain mobility. Cataract surgery, hormone management, sleep apnea treatment, and anesthetic precautions (important for surgery) round out care.

What does the future hold?

The course is slowly progressive over decades; many people with adult-onset type 1 and type 2 live long lives with good management. The heart rhythm risk is the main threat — and it is precisely the one surveillance handles best. Genetic counseling supports family planning; research into genetic treatments is active and promising.

Questions patients commonly ask

Q: What is myotonia exactly?

A: A delay in muscle relaxation after contraction — a handshake that's slow to release, or stiffness that eases as you 'warm up.'

Q: Why do I need heart checks for a muscle disease?

A: The gene affects heart conduction too; rhythm problems can be silent but are preventable with monitoring and, when needed, a pacemaker.

Q: Will my children be affected earlier or worse?

A: In type 1, repeats can expand between generations (anticipation); genetic counseling explains your family's specific situation and reproductive options.

National support groups & resources

- **Muscular Dystrophy Association** — mda.org
- **National Organization for Rare Disorders** — rarediseases.org — 1-800-999-6673
- **American Academy of Neurology — Brain & Life patient resources** — brainandlife.org

(Organization contact details were current at the time of compilation; please verify online.)

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