

Tips for Diagnosing Myotonic Dystrophy

Patients may present with:

Being "tired all the time"

Early onset cataracts

Unexplained arrhythmia (eg. heart block/LBBB)

Carry out your usual investigations, paying particular attention to other aspects of the Past Medical History, such as:

Facial weakness

Myotonia (eg. Difficulty releasing grip)

Frontal Balding

Previous anaesthetic difficulties

If investigations are normal, and there are still concerns, ask more generally about the Family History (FH), paying particular attention to:

FH Sudden death

FH anaesthetic difficulties FH muscle weakness

FH Myotonic Dystrophy FH Early onset cataracts

FH child with neuromuscular symptoms

If continuing concern, examine for Myotonia (eg. Grip myotonia - ask patient to squeeze fingers tightly for 10 seconds, then release)

If a diagnosis of Myotonic Dystrophy seems possible, please refer to your local regional genetics centre for Advice.

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written by General Practitioners for General Practitioners

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If you have a patient with myotonic dystrophy, please pass on our details as we would like to hear from them and offer the hand of support and friendship.

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