Tips for Diagnosing Myotonic Dystrophy

Patients may present with:

- Being “tired all the time”
- Early onset cataracts
- Unexplained arrhythmia (e.g. heart block/LBBB)

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

- Facial weakness
- Myotonia (e.g. 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 neuro-muscular symptoms

If continuing concern, examine for Myotonia (e.g. 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.
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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