MYOTONIC DYSTROPHY

KEY CLUES:

Distal muscle weakness
Myotonia
Typically teens, 20's, 30's

- inherited disorder
- adolescent/early adult onset
- distal muscle weakness and wasting progresses proximally (but usually maintains a distal predominance)
- distal weakness out of proportion to wasting
- myotonia after voluntary contraction (= Delayed relaxation of muscle after voluntary contraction due to abnormally increased muscle membrane excitability), e.g. may have difficulty releasing grip on door handle or may slur speech

TESTING FOR MYOTONIA:

1. hit thenar eminence - leaves imprint
2. "grip my fingers, let go really quickly" - fingers relax slowly rather than quickly

- multisystem disease:
  - cardiac arrhythmias
  - respiratory compromise/failure
  - lens opacities and subsequent cataracts (in almost all)
  - 40% patients have cognitive impairment
  - many have a degree of frontal lobe dysfunction
  - central and peripheral sleep apnoea
  - smooth muscle involvement (dysphagia, GI dysmotility, biliary tract dysfunction, impaired uterine contraction during labour, seminiferous tubule dysfunction)
  - impaired glucose tolerance and increased insulin response (DM is uncommon)
  - in men, hormone abnormalities can lead to frontal balding and infertility
  - affected females are at risk of having offspring with a severe and sometimes fatal form of congenital myotonic dystrophy

MYOTONIC DYSTROPHY TYPE 1

- lower legs (causing foot drop), hands (weakness of grip), neck (esp. sternocleidomastoid weakness), shoulders, elbows and hips typically affected
- mutation in DMPK gene (autosomal dominant trinucleotide repeat that exhibits anticipation)

MYOTONIC DYSTROPHY TYPE 2

- also known as proximal myotonic myopathy (PROMM)
- similar to, but milder than type 1 and weakness and myotonia is more proximal
- mutation in CNBP gene (autosomal dominant trinucleotide repeat - ?anticipation)