Muscular dystrophy family
Most common of the dystrophies
Myotonia- inability of muscles to relax after contraction
Myotonia then leads to muscle deterioration → versus Duchenne’s muscle swelling
Muscle weakness, irregular heartbeat, cataracts, mental deterioration
Categorized according to severity of symptoms: mild, classic, congenital
No cure for DMI (yet)- treatment is only alleviating symptoms
- Pain relievers, anti-inflammatory drugs
- Possible aerobics treatment may strengthen muscle (Orngreen 2005)
- Assistive devices
- DMI patients should avoid drugs that can cause muscle weakness or pain
DMPK gene, chromosome 19q13.3- codes for protein kinase in muscle.

Protein kinase- regulation of cell activity

DMI defect- increased repetition of CTG (>34)
→ the more repetitions, the more severe the disease

Abnormal length causes irregular hairpin folds in RNA

Gene test checks this repetition

Dominant allele of gene

Differential diagnosis with genetics can tell the difference between DMI and other dystrophies
Genetic Treatment

- Still being researched
- Strong conservation in other animals = high research potential
- 3 potential treatments:
  1. gene therapy to replace DMPK and protein kinase
  2. RNA-binding protein complex control (Mahadevan 2006)
  3. Normalizing the CTG chain by splicing (Timchenko 2006)
References


