MYOTONIC DYSTROPHY TYPE I

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Classical Diagnosis

- 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

Classical Treatment

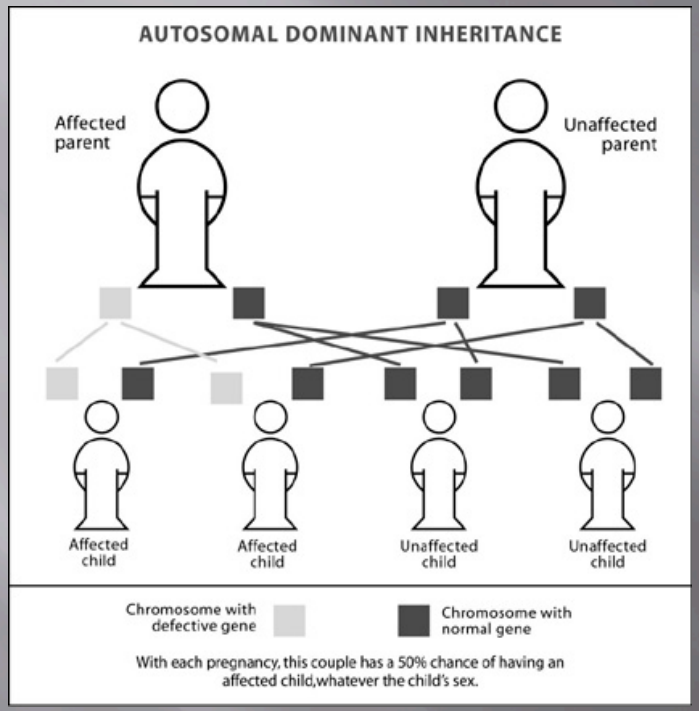
- No cure for DMI (yet)- treatment is only alleviating symptoms
- Pain relievers, anti-inflammatory drugs
- Possible aerobics treatment may strengthen muscle (Orngreen 2005)
- Assistive devices
- DMI patients should avoid drugs that can cause muscle weakness or pain

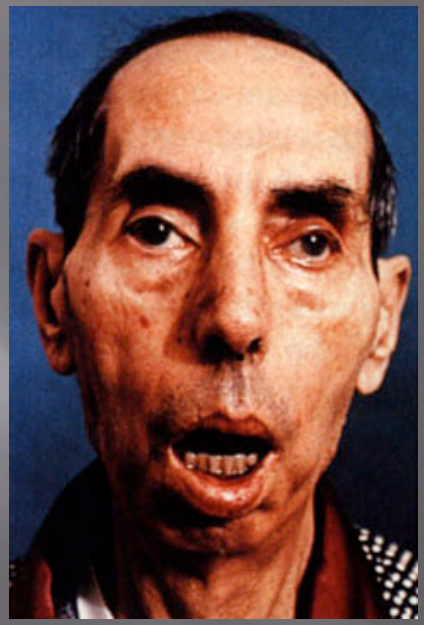
Genetic Diagnosis

- 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 Diagnosis





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

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