Molecular basis of myotonic dystrophy

Myotonic dystrophy is one of the most complex disorders known. In addition to the incredible variability of clinical symptoms, the disease also has unique mechanistic features:

- True autosomal inheritance. The disease phenotype of patients who are homozygous for myotonic dystrophy is essentially the same as those who are heterozygous.

- Variable penetrance. Considerable variability is seen between affected individuals, even within the same family. Somatic mosaicism is common, where the genetic defect can be significantly different in various tissues in a single individual and can change over time.

- Anticipation. The disease symptoms tend to be more severe and occur earlier in successive generations.

- Maternal transmission bias for the congenital form. In the most severe form of myotonic dystrophy (congenital myotonic dystrophy: DM1), transmission is nearly always maternal and does not appear to be related to the severity of the disease in the mother. The mutated gene is only very rarely inherited from the father in newborns with myotonic dystrophy.