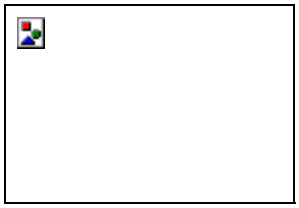

Kennedy's Disease

What is Kennedy's Disease?
A disorder of the motor neurones. A rare form of spinal muscular atrophy also known as X-linked recessive bulbospinal neuropathy or X-linked spinal and bulbar atrophy. (Spinal muscular atrophies are inherited neurological disorders in which only the lower motor neurones are affected.)

How is it caused?
A mutation on the androgen receptor gene on the X chromosome seems to affect regulatory activity in motor neurones, leading to degeneration of these cells.

Who gets it?
Since the mutation is on the X chromosome, the disease affects males only as they have only one X chromosome. Women may have the mutation, but as they have two X chromosomes, they do not show the effects of the mutation. For female carriers, however, 50% of their sons may get the disease, and 50% of their daughters may become carriers. Sons of affected males do not get the disease since the male does not pass on his X chromosome.

How is it diagnosed?
Since the disease is compatible with normal life span and patients can be mis-diagnosed with motor neurone disease, accurate diagnosis, using the gene mutation is critical.

When does it **show itself?**
Usually in the 40s or 50s, (though also known in teenagers, 60s or 70s).

Symptoms
Slowly progressive muscle weakness and atrophy; some facial weakness and fasciculation, tremor of the hands, atrophy of the tongue, of ten dysarthria and dysphagia (later in the course of the disease). There may be gynaecomastia (breast development in the male), testicular atrophy and reduced fertility.

Prognosis
This is usually good, and the disease is compatible with a normal life span. The degree of weakness, however, may slowly worsen. The overall prognosis is much better than classical MND.

What treatments exist?
There is currently no treatment to alter the progression of the disease, but symptomatic treatments can ensure quality of life is maintained.

Is there an argument for genetic screening?
If a treatment became available, then it may be important to detect men at the pre-symptom stage. It is possible to detect female carriers. However, this is usually a mild disorder, so pre-natal screening for affected foetuses may not be necessary. Nevertheless, there are some severely affected individuals and it is not possible to predict how an individual carrying the gene will be affected. Detailed counselling is therefore necessary, before a mother can make a fully informed decision.

{Source: **Motor Neurone Disease,** Leigh & Swash (eds), Springer-Verlag, 1995}