

Kennedy's Disease

X-Linked Spinal and Bulbar Muscular Atrophy

Origin - Australia

What is Kennedy's Disease?

Kennedy's disease is an inherited disorder affecting adult males, which causes slowly progressive weakness and wasting of muscles. It was first described in 1968 by Dr William R Kennedy and his co-workers. The genetic mutation that causes the condition was identified in 1991 by La Spada and others. Although it is a disorder of motor neurones, it is not Motor Neurone Disease, as it is usually much more benign.

What are the symptoms?

Symptoms generally begin to be noticed at about 40 or 50 years of age, although cases from the ages of 15 up to 59 have been described. Symptoms include muscle twitching (fasciculations), cramps, muscle weakness and wasting, fasciculations of the face, hand tremors, twitching and wasting of the tongue, problems with speech and swallowing, and in some cases abnormal sensation. The majority of males also show some breast enlargement, and reduced fertility has been reported in some cases.

Is it hereditary?

Kennedy's disease *is* inherited, and is passed on from mother to son, by an X-linked genetic mutation.

What causes it?

Kennedy's disease is caused by a mutation of the Androgen Receptor (AR) gene, the role of which is to moderate the action of the male sex hormones (androgens). How this mutation causes the symptoms seen in Kennedy's disease is not yet understood.

How is it diagnosed?

Kennedy's disease can be diagnosed by genetic testing using a sample of whole blood. The test is rapid and accurate and could also be used to detect presymptomatic individuals (i.e., those who have the mutated gene but do not show any signs of the disease), carriers, and for undertaking prenatal diagnosis.

Testing is done only after appropriate genetic counselling has been undertaken.

Is there any treatment?

There is as yet no treatment to cure Kennedy's disease or to stop its progression. The severity and course of the disease cannot be predicted, but in most cases, progression is extremely slow, and life expectancy is normal. Support in coping with the effects of the disease is available through the MND Associations in each State, and research is being undertaken which gives hope for the future.

Other fact sheets, including *Genes and Chromosomes*, *Genetic Counselling*, *Prenatal Diagnosis*, and *Drawing up your Family Health Tree*, are available from the [Genetics Education Program of NSW](#), P O Box 317, St Leonards, NSW 2065. Tel: (02) 9926 7324 Fax: (02) 9906 7529.

Reference:

Choi WT, Zajac JD et al. Kennedy's Disease: genetic diagnosis of an inherited form of

motor neurone disease. Aust NZ J Med 1993; 23.

KENNEDY'S DISEASE - PATTERN OF INHERITANCE

When a baby is conceived, each parent passes on one copy of each of their genes to the baby. Therefore the baby is a "mixture" of the genetic information of each of his/her parents. The sex of a child is determined by inheritance from their parents of X and Y chromosomes. Males have an X chromosome (from their mother) and a Y chromosome (from their father). Females have two X chromosomes, one from each parent. To be male, a child must inherit a Y chromosome from his father, (and therefore, in the case of Kennedy's disease, does not inherit the mutated X). **The sons of men with Kennedy's disease do not get the disease, and do not pass on the mutation.**

The daughters of men with Kennedy's disease are called "obligatory carriers". Females have two X-chromosomes, one from their father, and one from their mother. Since males have only one X chromosome to pass on, their daughters always receive the X chromosome carrying the genetic mutation. **Because they also have a normal X chromosome, they do not develop the disease.**

The sons of female carriers of the genetic mutation have a 50 per cent chance of receiving the mutated gene and developing Kennedy's disease. **The daughters of Kennedy's disease female carriers** have a 50 per cent chance of inheriting the mutated gene, but because they also have a normal X chromosome, they do not develop the disease. It is also important to note there is a 50 per cent chance that their female or male children *will not* inherit the mutant gene from their mother and therefore not transmit it to their children. (See Figure 2). ***These risks and possibilities apply to every pregnancy.***

For further information please contact the [Motor Neurone Disease Association in your State.](#)

This information is provided to develop better understanding and knowledge. It is not a substitute for qualified medical advice or genetic counselling. Please contact your Doctor for medical advice and support.

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