



2B

Kennedy's Disease


Information for people with or affected by Kennedy's Disease

If you have been diagnosed with Kennedy's disease, you may be looking for information to help you manage the condition. This information sheet explains how the disease may affect you and how to find support.

Due to similar symptoms, people with Kennedy's disease are sometimes misdiagnosed as having motor neurone disease (MND). Although it is not classed as a type of MND, the MND Association provides support if you are living with this condition.

This information includes the following sections:

- 1: **What is Kennedy's disease?**
- 2: **What are the symptoms?**
- 3: **What help can I get?**
- 4: **How do I find out more?**

 This symbol is used to highlight **our other publications**. To find out how to access these, see *Further information* at the end of this sheet.

 This symbol is used to highlight **quotes** from other people with or affected by MND.



The MND Association has been certified as a producer of reliable health and social care information.

www.theinformationstandard.org

What do the words mean?

Your health and social care team may use medical terms. Here are some words you may hear when you discuss Kennedy's disease.

androgen:	a hormone that stimulates the development of male and female characteristics.
androgen receptor:	a protein that the androgen hormones fit into, part of a chain of events that trigger the development of sexual characteristics.
atrophy:	wasting of the tissues or organs of the body.
bulbar:	generally used to describe symptoms that affect the mouth and throat – the word refers to the bulb-shaped part of the brain stem that contains motor neurones needed for swallowing, speaking and chewing.
chromosome:	the part of any cell that contains our genes.
dysarthria:	problems with speech and language.
dysphagia:	difficulty with swallowing.
fasciculation:	involuntary twitching or spasms in the muscles.
genes:	these make up the chromosome in each cell to carry the information that determines your physical traits.
genetic mutation:	a change in the information that makes up our genes.
gynaecomastia:	excessive breast development in men.
testosterone:	an androgen hormone.
X-linked spinal bulbar muscular atrophy (SBMA):	another name for Kennedy's disease.
X-linked bulbospinal neuropathy:	another name for Kennedy's disease.

1: What is Kennedy's disease?

Kennedy's disease causes progressive weakening, wasting of the muscles and hormonal changes. It is a rare condition, with an estimated 1 in 40,000 people affected. As it is rare, health and social care professionals may be unaware of it. Research into Kennedy's disease is ongoing. See later heading in this section *How can I be involved in research about Kennedy's disease?*



"I have been diagnosed for 10+ years and find knowledge of Kennedy's to be very limited in the medical profession, so I personally have had no information on managing symptoms, available help or information on how to find out more".

Most people with Kennedy's disease start to show symptoms when they are 30-60 years old, but it can appear in older or younger people. There is currently no known cure, but symptoms can be managed to improve quality of life. Most people with Kennedy's disease live an average life span.

What causes Kennedy's disease?

Kennedy's disease is hereditary, passed on from parent to child in the genes. It is caused by a genetic change (mutation) that damages the nerves that control voluntary muscle movement. See later heading in this section *Have I inherited the gene?*

How does it affect women?

Most women who inherit the gene will not develop any symptoms, but will be carriers of it and may pass it on to their sons or daughters. See later heading *Have I inherited the gene?*

In rare cases, women may develop symptoms, but these are usually milder than those experienced by men. The main symptoms of Kennedy's disease in women are mild muscle cramps and fatigue. Little is known about Kennedy's disease in women and research is currently underway.

How is Kennedy's disease diagnosed?

You will be seen by a neurologist, who should be able to exclude other conditions. As Kennedy's disease is rare, many neurologists will have little experience of the disease and you may be referred to a regional centre. A range of tests may be offered, including:

- measuring electrical activity in your muscles to check for nerve damage
- blood tests to look for a rise in a substance that is produced when muscle breaks down
- genetic tests to check whether you have the gene for Kennedy's disease.

You may also be asked about your family's medical history.

Have I inherited the gene?

Both men and women can pass the gene to their children. Women carrying the gene have a 1 in 2 chance of passing it on to their sons and daughters. Men do not pass it to their sons, but will pass it to all their daughters, who will become carriers of the disease. If you are concerned about a family history of Kennedy's disease and what this could mean for those close to you, you may wish to seek genetic counselling.

A genetic counsellor will explain all the facts to you as clearly as possible, and give you accurate information on the possible implications for your family. This will include information about options such as genetic testing, to help you decide whether or not this is something you wish to do.

You may find this decision difficult, as it affects the wider family and may raise questions about options relating to pregnancy. Genetic testing can show if you carry the gene, but cannot predict how you or your children might be affected.

How can I be involved in research about Kennedy's disease?

There is a national register of people with Kennedy's disease.

Joining the register will mean that you can be contacted by the National Hospital for Neurology and Neurosurgery and kept up to date with the latest information about the disease. They will also be able to contact you about taking part in new studies, drug trials and other initiatives for people with Kennedy's disease.

You can register by calling the National Hospital for Neurology and Neurosurgery by calling 020 3448 3899.

2. What are the symptoms?

Kennedy's disease affects people in different ways. Not everyone will develop all of the symptoms listed here and some people may only experience very mild symptoms. The most common symptoms in women are mild muscle cramps and fatigue.

The symptoms of Kennedy's disease in men are:

- fatigue
- twitching and rippling sensations under the skin
- tremors and muscle cramps

- muscle weakness
- growth of breast tissue
- reduced sex drive and difficulty getting an erection
- reduced fertility
- difficulty swallowing
- slurred speech.

You may experience hand and muscle tremors for several years before any weakness develops.

Over time muscles become weaker and begin to waste away. This makes movement tiring and difficult and can affect your posture and balance, increasing the risk of falls and injuries.

3. What help can I get?

There is currently no known cure for Kennedy's disease. The focus is on relieving your symptoms and maintaining the best quality of life possible.

How do I manage fatigue?

It might be helpful to take on the idea that you have a 'bank account' of energy to spend each day. If you overspend on energy one day, you may feel extremely tired the next.

Plan to do valued and essential tasks first. Delay or get assistance for other tasks as needed. Try to:


- listen to your body and pace yourself
- prioritise tasks, manage your time and be flexible with routines
- use equipment to help you do things more easily
- talk to your physiotherapist, occupational therapist and dietitian about ways to manage your fatigue
- plan a rest day before and after a particularly busy time, such as a family event.

What can be done about twitching and rippling sensations?

Talk to your GP, consultant or specialist nurse, as medication may help to relieve this symptom.

What can be done about muscle cramps and tremors?

Your GP, consultant or specialist nurse may be able to offer you medication to ease cramps and muscle spasms. Physiotherapy may also provide some relief.


 For information about physiotherapy with MND see:
Information sheet 6A – Physiotherapy

Getting plenty of rest will help to alleviate painful cramps. It is also helpful if you have any trip or fall injuries, as these are made worse by tired muscles and exhaustion.

What can be done about muscle wasting and weakness?

Muscle wasting and weakness cannot be reversed by exercise. Gentle regular exercise may assist with flexibility and range of movement in your joints, help maintain unaffected muscles, and support posture and balance. Gentle muscle stretching can also relieve cramps.

Ask your GP or health and social care team for a referral to a relevant physiotherapist who can recommend a suitable exercise programme to meet your needs.

 For information about physiotherapy with MND see:
Information sheet 6A – Physiotherapy

What can be done about numbness?

Damage to your nerves cannot be undone, so the numbness cannot be treated. As this nerve damage reduces your sensitivity, it is important to ensure that the skin is kept clean, dry and undamaged to reduce the risk of pressure sores. Talk to your district nurse and occupational therapist for advice about taking care of your skin and for information about any equipment that may help to reduce the risk of pressure sores.

What can I do about male breast development?

Surgery to remove the enlarged breast glands may be an option. Ask your GP or health and social care team for a referral to an appropriate surgeon to discuss your options.

What can be done about sexual problems?

If you are a man with Kennedy's disease, you may have a reduced sex drive and difficulty getting an erection. You may also develop fertility problems. In mild cases of Kennedy's disease, your fertility and sexual function will not be affected.

Medications that replace hormones are not suitable as there is a risk that these can make some of your symptoms worse. Ask your GP for advice as other medications may be available to help. They will also be able to advise you about your options if you are planning a family.

What can I do about swallowing difficulties?

With Kennedy's disease, swallowing can become difficult and lead to coughing and choking. This can lead to an increased risk of inhaling small pieces of food or saliva into your lungs, which can cause chest infections and pneumonia. Swallowing difficulties can also lead to weight loss, dehydration and lack of energy.

Ask your GP for a referral to a speech and language therapist and a dietitian who will be able to assess your ability to swallow safely and give you information about the types of diet, supplements or aids that might support you to maintain a healthy diet.



For more information about swallowing see:
Information sheet 7A – Swallowing difficulties

What can I do about problems with my speech?

Kennedy's disease may affect the muscles in your tongue and lips, making it difficult to speak. Your speech may become unclear and slurred.

Ask your GP for a referral to a speech and language therapist for an assessment and advice about techniques or equipment to help with your speech problems.

What can I do about diabetes?

With Kennedy's disease, you may develop type 2 diabetes. The main symptoms to look out for are:

- feeling thirsty
- urinating more than usual
- blurred vision
- slow healing of cuts and grazes
- recurring thrush.

If you experience these symptoms, ask your GP for advice. They may prescribe medications or refer you to a dietitian for advice on controlling your blood sugar levels.

Where can I get further support?

You can ask your GP to refer you to a neurologist who is familiar with Kennedy's disease. Contact MND Connect who can help direct you to centres where Kennedy's disease is a specialism. See section 4. *How do I find out more?* for contact details.

4. How do I find out more?

Useful organisations

We do not necessarily endorse the external organisations listed here. These have been provided to help you search for further information if necessary.

Details are correct at the time of print, but may change between revisions. If you need help to find an organisation, contact our MND Connect helpline (see Further information at the end of this sheet for details about our helpline and how they can support you).

GOV.UK

Online government advice on a variety of welfare topics, including support for people with disabilities.

Email: email addresses are provided on the website, related to each enquiry
Website: **www.gov.uk**
www.nidirect.gov.uk (Northern Ireland)

Health in Wales

Information on NHS services in Wales, including a directory of the Welsh health boards. This is an online information service only (see also NHS Direct Wales).

Email: through the website contact page
Website: **www.wales.nhs.uk**

MND Scotland

MND Scotland provides care, information and research funding for people affected by motor neurone disease in Scotland.

Address: 2nd Floor, City View, 6 Eagle Street, Glasgow G4 9XA
Tel: 0141 332 3903
Email: info@mndscotland.org.uk
Website: **www.mndscotland.org.uk**

Kennedy's Disease Association (KDA)

American organisation supporting people with Kennedy's disease. Their website includes a chat room and forum.

Address: PO Box 1105, Coursegold, CA 93614, USA
Telephone: (559) 962-5950
Email: info@kennedysdisease.org
Website: www.kennedysdisease.org

Kennedy's Disease UK

UK based charity run by volunteers and people with Kennedy's disease. Working to raise awareness of the disease.

Email: via the website
Website: <http://kennedysdiseaseuk.com>

NHS Choices

The main online reference for the NHS, including information on continuing healthcare.

Address: Customer Service Centre, The Department of Health, Richmond House,
79 Whitehall, London SW1A 2NL
Telephone: 0207 210 4850
Email: through the website contact page
Website: www.nhs.uk

NHS 111

The NHS online/telephone service if you need urgent, but not life-threatening medical help or advice. Available 24-hours a day, 365 days a year.

Telephone: 111 (England)
or: 0845 4647 (for Cornwall, Scilly Isles, Luton and Bedfordshire)

NHS Direct Wales

Information on NHS services in Wales.

Telephone: 0845 4647
Website: www.nhsdirect.wales.nhs.uk

NHS Northern Ireland

Information on NHS services in Northern Ireland. This is an online service only.

Email: through the website contact page
Website: www.hscni.net

NI Direct

Providing government information on a variety of welfare subjects, including health services and support for disability for Northern Ireland.

Email: through the website contact page

Website: **www.nidirect.gov.uk**

References

References used to support this document are available on request from:

Email: infofeedback@mndassociation.org

Or write to:

Information feedback
MND Association
PO Box 246
Northampton
NN1 2PR

Acknowledgements

Many thanks to:

Pietro Fratta MD, PhD. MRC/MNDA LEW Clinician Scientist and Honorary Consultant Neurologist at UCL Institute of Neurology and National Hospital for Neurology and Neurosurgery

P Nigel Leigh, Professor of Neurology at Brighton and Sussex Medical School, Trafford Centre for Biomedical Research and University of Sussex and Professor Emeritus, King's College London

Professor Adrian Wills, Consultant Neurologist, Nottingham University Hospitals NHS Trust

Further information

We provide other sheets related to this information.

Although they are aimed at people with MND, much of the advice and information given may be helpful with Kennedy's disease too.

6A – Physiotherapy

7A – Swallowing difficulties

We also produce the following guides:

Living with motor neurone disease – our main guide to MND and how to manage its impact
Caring and MND: support for you – comprehensive information for unpaid and family carers, who are supporting someone living with MND
MND Association Recipe Collection: easy to swallow meals

You can download most of our publications from our website at:
www.mndassociation.org/publications or order in print from the MND Connect team, who can provide additional information and support:



MND Connect
MND Association, PO Box 246, Northampton NN1 2PR
Telephone: 0808 802 6262
Email: mndconnect@mndassociation.org

MND Association website and online forum
Website: **www.mndassociation.org**
Online forum: **<http://forum.mndassociation.org>** or through the website

We welcome your views

Your feedback is really important to us, as it helps improve our information for the benefit of people living with MND and those who care for them.

If you would like to provide feedback on any of our information sheets, you can access an online form at: **www.surveymonkey.com/s/infosheets_1-25**

You can request a paper version of the form or provide direct feedback by email:
infofeedback@mndassociation.org

Or write to:
Information feedback, MND Association, PO Box 246, Northampton NN1 2PR

MND Association website and online forum
Website: **www.mndassociation.org**
Online forum: **<http://forum.mndassociation.org/>** or through the website

Last revised: 06/15
Next review: 06/18
Version: 3