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Kennedy's disease

Information for people with or affected by Kennedy's disease

If you have been diagnosed with Kennedy's disease, also known as spinal bulbar muscular atrophy (SBMA), 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). The MND Association provides support if you are living with or affected by Kennedy's disease.

Throughout this sheet, we will refer to other publications to find further information. While these are aimed at people living with MND, their content may also be helpful for Kennedy's disease.

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.

This information has been evidenced, user tested and reviewed by experts.

What do the words mean?

You may hear the following medical terms at appointments:

androgen	A hormone that stimulates the development of mainly male characteristics.
androgen receptor (AR)	A protein that androgen hormones fit into. This is part of a chain of events that trigger the development of sexual characteristics.
atrophy	Wasting of the tissues or organs in the body. This usually applies to muscle.
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	Slurred speech.
dysphagia	Difficulty with swallowing.
fasciculation	Involuntary twitching of the muscles.
genes	These make up the chromosome in each cell to carry the information that determines our physical traits.
genetic mutation	A change in the information that makes up our genes.
gynaecomastia	Excessive breast development in men.
testosterone	A male sex 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 weakness and wasting of the muscles. Kennedy's disease is also known as spinal bulbar muscular atrophy (SBMA). 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.

 *"Until I was diagnosed, I had never heard of Kennedy's disease and neither had my consultant, which shows how little information there has been available."*

Research into Kennedy's disease is ongoing. See later heading in this section *How can I be involved in research about Kennedy's disease?*

Most people with Kennedy's disease start to show symptoms when they are 40-60 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 a relatively normal life span.

What causes Kennedy's disease?

 *"Little seems to be known about Kennedys, even in the medical profession, so any information is essential."*

Kennedy's disease is genetic. This means there has been a change in the genes (a genetic mutation), which is passed on from parent to child. This can cause damage to the nerves that control voluntary muscle movement. See later heading in this section *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.

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 very rare cases, women may develop symptoms, but these are usually much milder than men's and experienced at a later age. The main symptoms of Kennedy's disease in women are mild muscle cramps and fatigue, but this is not usually a sign that they will develop more severe symptoms. Little is known about Kennedy's disease in women and research is ongoing.

How is Kennedy's disease diagnosed?



"Most of us with Kennedy's disease have been misdiagnosed with ALS."

You will be seen by a neurologist. As Kennedy's disease is rare, many neurologists will have little experience of the disease and you may be referred to a regional centre. The following steps are likely to happen:

Assessment: your neurologist will do a basic assessment. You may also be asked about your family's medical history.

Ruling out other conditions: you may be asked to have a range of tests to narrow down the possible causes of your symptoms. These may include:

- measuring electrical activity in your muscles to check for nerve damage
- blood tests to look for a rise in a substance called creatine kinase (CK), which is produced when muscle breaks down.

Genetic testing: once a neurologist suspects it could be Kennedy's disease, a genetic test can check whether you have the gene. This will confirm the diagnosis.

Have I inherited the gene?

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.

Genetic counselling is where you meet with a genetic counsellor to get support, information and advice about a genetic condition. The counsellor will explain all the facts to you as clearly as possible, and give you accurate information on what this might mean 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 or when 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 means that you consent to 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 contacting the National Hospital for Neurology and Neurosurgery on:

Telephone: 020 3448 3899

Email: sbma@ucl.ac.uk

You can also use the above details to find out about the specialist Kennedy's disease clinic hosted at this hospital. At the clinic, you can be supported by a range of professionals working together to provide co-ordinated care and linked services (multidisciplinary team).



"A multidisciplinary team is equally as important for people with Kennedy's Disease as it is for people with MND."

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:

- tiredness (fatigue)
- twitching and rippling sensations under the skin (fasciculations)
- tremors and muscle cramps
- muscle weakness
- growth of breast tissue (gynaecomastia)
- reduced sex drive and difficulty getting an erection
- reduced fertility
- tightening or twitching in the throat (laryngospasm)
- needing to pee more urgently and often
- increased sugar (glucose) and cholesterol levels, meaning you may be at risk of developing diabetes
- difficulty swallowing and slurred speech.

Breathing can also be affected for some men living with Kennedy's disease, but this is usually mild and is most often experienced when lying down. If you do experience breathing difficulties with Kennedy's disease, contact your health and social care team. They may refer you to a respiratory team, who can provide support. There may be equipment that can help with this symptom.

Kennedy's disease can cause an abnormal heart rhythm in rare cases. For more information on heart monitoring, your specialist doctor can contact the Kennedy's disease clinic, based in London (see *Useful organisations* at the end of this sheet for contact details).

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

Over time muscles become weaker and begin to waste away. This makes movement tiring and difficult. This 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. Instead, care and support focus on relieving your symptoms and maintaining the best possible quality of life. If you are not currently supported by any of the professionals mentioned in this section, ask your GP for a referral.

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.



"Listen to your body and perhaps do a task in two steps, rather than all at once. You achieve the task in the end, but without the frustration of tiredness."

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
- seek advice from your occupational therapist (OT), physiotherapist and dietitian about ways to manage your fatigue
- plan a rest day before and after a particularly busy time, such as a family event.



We have an information sheet on fatigue for people living with MND, which you may find useful, see: Information sheet 11D – *Managing fatigue*

What can be done about twitching and rippling sensations?

Talk to your GP, consultant or specialist nurse, as medication may be available to help 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*

 *“Relaxation techniques can be very helpful in decreasing pain somewhat.”*

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.

 For detail on support for pain, see:
Information sheet 6C – *Managing pain*

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.

 *“I was given gentle exercises that could be done lying on a bed, mainly stretching and moving limbs. I now find them essential.”*

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 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 (OT) for advice about taking care of your skin and for information about any equipment that may help to reduce the risk of pressure sores.

 For more information on equipment, see:
Information sheet 11C – *Equipment and wheelchairs*

What can I do about male breast development (gynaecomastia)?

If breast development is impacting on your confidence, it may help to find ways to disguise it such as wearing dark, patterned tops.

Hormone replacement medications are not appropriate to treat breast development in men with Kennedy's disease, as they could make some of your other symptoms worse.

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?

In some cases, men with Kennedy's disease may have a reduced sex drive and difficulty getting an erection. Some men also develop fertility problems. However, in mild cases, fertility and sexual function are not 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 twitching and tightening in my throat (laryngospasm)?

Sometimes you may experience twitching and tightening in your throat, known as a laryngospasm. This is when your vocal folds (also known as vocal cords) move close together, which can make it difficult to breathe and swallow. Laryngospasms usually last for around 60 seconds, but can be longer or shorter. Some people experience more than one laryngospasm at a time.



"I have had several laryngospasms, one after another."

This can be distressing, but it will pass. It can help to try to remain calm and relax during a laryngospasm.

Some people report that 'straw breathing' during a laryngospasm is also helpful. 'Straw breathing' is when you make the shape of your mouth as if you are sucking through a straw and slowly taking breaths in this way. It is thought that this slows down your breathing and allows your vocal folds to relax. Breathing in quickly can make the vocal folds move closer together, making it more difficult to breathe.

If you do experience this symptom, discuss this with your neurologist, as medications are available to control it.

What can I do about needing to pee more urgently and often?

If your mobility is affected by Kennedy's disease, it can make getting to the toilet in time more difficult. Wearing clothes with easy to undo fastenings may help. Other aids, like a raised toilet seat or grab rails can help you get on and off the toilet more easily.



For more information, see:

Information sheet 11A – *Clothing*

Information sheet 11C – *Equipment and wheelchairs*

Sometimes diabetes can lead to needing to pee more often. Ask your GP for support (see later heading *What can I do about diabetes?*).

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 (SLT) 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 help you maintain a healthy diet.

Kennedy's disease may also affect your ability to cough effectively. Ask for a referral to a respiratory physiotherapist, who may be able to teach you techniques to help you cough.



For more information about swallowing see:

Information sheet 7A – *Swallowing difficulties*

Eating and drinking with MND

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 speech problems.



For more detail, see:

Information sheet 7C – *Speech and communication support*

Information sheet 7D – *Voice banking*

What can be done about diabetes?

Kennedy's disease can be linked to glucose intolerance and diabetes. Your GP should be monitoring the level of glucose in your blood. If this is not happening, ask them to discuss this with your specialist doctor.

If you show signs of diabetes, your GP may refer you to a dietitian who specialises in diabetes or be able to prescribe medication to help with this.

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 in England and Wales on a variety of welfare topics, including support for disabled people.

Website: www.gov.uk

Health and social care online (NHS Northern Ireland)

Online information on health and social care services in Northern Ireland.

Website: www.hscni.net

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

Kennedy's Disease Association (KDA)

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

Address: PO Box 1105, Coursegold, CA 93614-1105, USA
Telephone: (734) 228-5580
Email: info@kennedysdisease.org
Website: **www.kennedysdisease.org**

Kennedy's disease clinic (based in London)

The only Kennedy's disease specialist clinic in the UK, providing multidisciplinary care and support.

Address: Motor Neuron Diseases, National Hospital for Neurology and Neurosurgery, Box 125, Queen Square, London WC1N 3BG
Telephone: 020 3448 3899
Email: sbma@ucl.ac.uk
Website: **www.uclh.nhs.uk/OurServices/ServiceA-Z/Neuro/MND/Pages/Home.aspx**

Kennedy's disease raising awareness (Facebook group)

This Facebook group may be helpful for people who want to virtually meet others living with the condition. Please note that the MND Association is not responsible for the group or moderation of its content.

Website: **www.facebook.com/groups/kennedysdisease.raisingawareness**

Kennedy's Disease UK

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

Email: through the website contact page
Website: **<http://kd-uk.com>**

NHS UK

The main online reference for the NHS.

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 and some areas of Wales)

NHS Direct Wales

Health advice and information on NHS services in Wales. Available 24 hours a day, 365 days a year.

Telephone: 0845 4647 or 111 if available in your area
Website: **www.nhsdirect.wales.nhs.uk**

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**

Remember the Girls

American organisation raising awareness and support for women and girls who carry X linked recessive disorders.

Website: **<https://rememberthegirls.org/>**

References

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

Email: infofeedback@mndassociation.org

Or write to:

Information feedback, MND Association, Francis Crick House, 6 Summerhouse Rd, Moulton Park Industrial Estate, Moulton Park, Northampton NN3 6BJ

Acknowledgements

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

Further information

Although our information resources are aimed at people living with and affected by MND, much of the guidance and information provided in them may be helpful for people living with and affected by Kennedy's disease too. We provide the following sheets that may be helpful:

6A – *Physiotherapy*

6C – *Managing pain*

7A – *Swallowing difficulties*

7C – *Speech and communication support*

7D – *Voice banking*

10A-G – Our range of information sheets on social care and financial support

11A – *Clothing*

11C – *Equipment and wheelchairs*

11D – *Managing fatigue*

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

Eating and drinking with MND – including information to help you adapt how you eat and drink, if needed. It includes information, tips and easy-swallow recipes.

Making the most of life with MND – our booklet on quality of life with MND or Kennedy's disease. It focuses on how adapting your approach may help you continue doing the things you want to do.

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

Telephone: 0808 808 6262

Email: mndconnect@mndassociation.org

MND Association, Francis Crick House, 6 Summerhouse Rd,
Moulton Park Industrial Estate, Moulton Park,
Northampton NN3 6BJ

MND Association website and online forum

Website: **www.mndassociation.org**

Online forum: **<https://forum.mndassociation.org>** or through the website

We welcome your views

We'd love to know what you think we're doing well and where we can improve our information for people with or affected by MND, or Kennedy's disease. Your anonymous comments may also be used to help raise awareness and influence within our resources, campaigns and applications for funding.

To feedback on any of our information sheets, access our online form at:

www.smartsurvey.co.uk/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, Francis Crick House, 6 Summerhouse Rd,
Moulton Park Industrial Estate, Moulton Park, Northampton NN3 6BJ

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