

Kennedy's disease

Information for people with or
affected by Kennedy's disease



“My father has been able to reach out for advice about the support available and the changes that will continue to develop with his condition.”



Contents

How can this booklet help me?	4
1: What is Kennedy's disease?	5
2: What are the symptoms?	10
3: What help can I get?	13
4: What do all the words mean?	22
5: How do I find out more?	24



Patient Information Forum

This content has been evidenced, user tested and reviewed by experts

<https://piftick.org.uk>

How can this booklet help me?

If you have been diagnosed with Kennedy's disease, also known as spinal bulbar muscular atrophy (SBMA), this booklet explores 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 offers support to people living with MND, or Kennedy's disease, and their families and carers.



We mention various MND resources in this booklet, where these may be useful for symptoms or care with Kennedy's disease.



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

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

1: What is Kennedy's disease?

Kennedy's disease is also known as spinal bulbar muscular atrophy (SBMA), and causes progressive weakness and wasting of muscles.

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.

Only 1 person in every 40,000 is affected by Kennedy's disease. This means it is rare, and health and social care professionals may not be aware of this condition.



“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 the later heading in this section *How can I be involved in research about Kennedy's disease?*

There is currently no known cure, but symptoms can be managed to improve quality of life. Although Kennedy's disease can impact on the way you live, most people with the condition have a relatively normal lifespan.

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, which means a genetic change can be passed from parent to child. This change can cause damage to the muscles and nerves that control voluntary muscle movement.

Both males and females can pass the affected gene to their children.

Females carrying the changed gene have a 1 in 2 chance of passing it on to their sons and daughters.

Males do not pass Kennedy's disease to their sons, but will pass it to all their daughters, who will become carriers of the changed gene.

See the later heading in this section *Have I inherited the gene?*

How is Kennedy's disease diagnosed?



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

If your doctor suspects you have a neurological condition, you will be referred to a neurologist. 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 changed gene. This will confirm the diagnosis.



For more about how Kennedy's disease can affect you, see Section 2: *What are the symptoms?*



Have I inherited the gene?

If you are a blood relative of someone with Kennedy's disease, you may want to find out if you carry the changed gene that causes the condition. Ask to be referred for genetic counselling, which can help you explore whether or not to go ahead with genetic testing.

Genetic counselling is sensitive to the emotional aspects of the situation, but is not a form of psychotherapy. The counsellor will explore all of the options that are involved.

Choosing to go ahead with gene testing can be difficult as it affects the wider family and may raise questions about options relating to pregnancy.

Genetic testing can only show if you carry the gene. It cannot predict how or when the condition might start.

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 3108 7507**

Email: **uclh.kdregister@nhs.net**

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).



For further details see Section 3: *What help can I get?*



"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.

How does Kennedy's disease affect you if you're male?

The symptoms of Kennedy's disease in males are:

- muscle weakness leading to mobility problems
- muscle cramps
- tremors
- twitching and rippling sensations under the skin (fasciculations)
- fatigue (feeling tired all the time)
- 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 and fatty liver
- difficulty chewing and swallowing
- a nasal sounding and slurred voice leading to speech and communication problems
- anxiety and depression.

Kennedy's disease can cause an abnormal heart rhythm in rare cases. This may need heart monitoring by your specialist doctor or they can contact the Kennedy's disease clinic in London.



See *Useful organisations* in Section 5: *How do I find out more?* for contact details.

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, or after doing a lot of activity.

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 would assess your condition and if needed provide support. There may be equipment that can help with this symptom, however ventilation support is not usually needed with Kennedy's disease.

You may experience hand and muscle tremors and cramps for several years before any weakness develops. Weakness may not develop in exactly the same area where you experience tremors and cramps.



Over time muscles become weaker and begin to waste away. This makes movement tiring and difficult. This can affect your posture, balance, or loss of mobility.

Ask your physiotherapist about exercises and orthotics (devices to help support limbs) that can help you with balance.

How does Kennedy's disease affect you if you're female?

In very rare cases, females may develop symptoms, but these are usually much milder than a male might experience at a later age.

The main symptoms of Kennedy's disease in females 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 females and research is ongoing.

Females with the Kennedy's disease gene are usually unaffected but can still pass it down to their children.



See Section 1: *What is Kennedy's disease* for more about how the disease is inherited.



3: What help can I get?

There is currently no known cure for Kennedy's disease. Instead, care and support is focused 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
- plan a rest day before and after a particularly busy time, such as a day out.
- seek advice from your occupational therapist (OT), physiotherapist and dietitian about ways to manage your fatigue



For ways to manage fatigue see: our booklet, *Personal care*

What can be done about twitching, cramps and spasms?

You may experience:

- twitching or a sense of rippling beneath the skin (known as fasciculations)
- muscle cramps or spasms.

Medication, physiotherapy or complementary therapies may help to ease these symptoms. Ask your GP, consultant or specialist nurse for guidance.



For more see:

Information sheet 6A – *Physiotherapy*

Stretching may help to alleviate painful cramps.

Gentle exercise can also maintain muscles, balance and movement.



For detail on support for pain, see:

- Information sheet 6B – *Complementary therapies*
- Information sheet 6C – *Managing pain*



What can be done about muscle wasting, weakness and mobility?

Muscle wasting and weakness cannot be reversed by exercise. However, regular exercise to maintain muscles and joints can support balance, flexibility and range of movement. Gentle muscle stretching can also ease cramps.



“I was given gentle exercises ... 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, for guidance on exercise to meet your needs. Your physiotherapist, occupational therapist and NHS Wheelchair Services, can also help you explore assistive equipment.



For more see:

- Information sheet 6A – *Physiotherapy*
- Information sheet 11C - *Equipment and wheelchairs*
- *Getting Around with MND* booklet



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

What can be done about numbness?

Kennedy’s disease mainly affects the muscles and nerves controlling movement. However, some people can also feel reduced sensitivity and numbness, usually in the lower legs and feet. Damage to your nerves cannot be undone, so numbness cannot be treated.

Reduced sensitivity and changes in sensation can sometimes make your feet feel very hot or cold with pins and needles, or nerve pain.

If these changes in sensation are affecting your everyday wellbeing, ask your neurologist for guidance as they may be able to prescribe medications or treatments to reduce the impact of these symptoms.



For more about equipment, see:
Information sheet 6C – *Managing Pain*



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 males 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, sexual function is not affected. However, where symptoms are more severe, Kennedy's disease can impact on fertility and your sex drive if you're male. You may find it more difficult to get an erection.

Hormone medications 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 tightening in my throat (laryngospasm)?

Sometimes you may experience 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.

Although this can be distressing it will pass. It can help to try to remain calm and relaxed 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.



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

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. You may also feel a sense of urgency. Wearing clothes with easy to undo fastenings may help. Other aids, such as a raised toilet or toilet seat, with grab rails can help you get on and off more easily.



For more information on managing toilet care see:

- our booklet, *Personal care*
- information sheet 11C - *Equipment and wheelchairs*

There is a slightly higher risk of diabetes with Kennedy's disease. This may 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, chewing and swallowing can become difficult and lead to coughing or a bout of 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.

They will also be able to 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 can help with techniques to support your cough.



For more on swallowing see:

- information sheet 7A - *Swallowing difficulties*
- our guide *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. It's rare for someone with Kennedy's disease to lose their voice completely, but your speech may become unclear and slurred.

Ask your GP for a referral to a speech and language therapist for assessment of your needs. They can advise:

- ways to manage your speech
- equipment that can help
- recording your voice to bank it for future use on communication aids.



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, which can lead to diabetes. Ask your GP to monitor the level of glucose in your blood. If this is not happening, ask them to discuss this with your specialist doctor.

If needed, you maybe prescribed medication and your GP may refer you to a dietitian who specialises in diabetes.

What can be done about high cholesterol and triglycerides?

Some people with Kennedy's disease may be at risk of high cholesterol and triglyceride levels. These affect the amount of fat content in the blood, which can increase risk of other conditions, such as fatty liver disease.

Statins are not commonly used with Kennedy's disease so ask your GP or specialist doctor to monitor these 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 neurological services where Kennedy's disease is a specialism.



See Section 5: *How do I find out more?* for contact details of the Kennedy's disease clinic based in London.

4: What do all 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. The androgen receptor is also a gene, which carries the mutation typical of Kennedy's Disease.
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.



5: 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 booklet 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**

Kennedy's disease clinic (based in London)

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

Telephone: 020 3108 7507 (General Enquiries)

Email: uclh.kdregister@nhs.net

Website: **www.uclh.nhs.uk/our-services/find-service/neurology-and-neurosurgery/motor-neuron-diseases**

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: **<https://kd-uk.com>**

Kennedy's Disease Association (KDA)

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

Telephone: (734) 288-5580

Email: through the website 'Contact us' form

Website: <https://kennedysdisease.org>

Kennedy's disease raising awareness (Facebook group)

Online support from others affected by Kennedy's disease.
The MND Association is not responsible for this group.

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

NHS and UK healthcare

Information about NHS services and healthcare across the UK

Website: www.nhs.uk

For England

Telephone: 111 (for urgent medical advice, available 24/7)

Website: <https://111.nhs.uk>

For Wales

Telephone: 111 (for urgent medical advice, available 24/7)

Website: <https://111.wales.nhs.uk>

For Scotland

Telephone: 111 (for urgent medical advice, available 24/7)

Website: www.nhs24.scot

For Northern Ireland

Telephone: Available via individual trusts 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

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 by email: infofeedback@mndassociation.org

Or write to us at:

Information feedback, Motor Neurone Disease Association,
Francis Crick House, 6 Summerhouse Road,
Northampton, NN3 6BJ

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Further information

If you have Kennedy's disease, you may find our other information resources helpful. Many of these refer to motor neurone disease (MND), but the symptoms and available support are very similar.

We provide the following information sheets that may be helpful:

- 6A – *Physiotherapy*
- 6C – *Managing pain*
- 7A – *Swallowing difficulties*
- 7C – *Speech and communication support*
- 7D – *Voice banking*
- 10A to 10G – *Our range of information sheets on social care and financial support*
- 11C – *Equipment and wheelchairs*

We also produce the following guides and booklets:

- ***Personal care*** – Suggestions of ways to manage everyday personal care tasks, such as washing, dressing and intimate care.
- ***Eating and drinking with MND*** – includes information to help you adapt how you eat and drink, if needed. It includes information, tips and easy-swallow recipes.
- ***Sex and relationships*** – Guidance on intimacy for anyone affected by MND or Kennedy's disease, whether in a relationship or not and regardless of sexuality.
- ***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.

- ***Living alone with MND or Kennedy's disease*** - explores the challenges you are likely to face and the relevant resources available to help.
- ***Caring and MND: support for you*** – guide for providing support for a partner, friend or relative, whether living with MND or Kennedy's disease.

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 website and online forum

Website: **www.mndassociation.org**

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

Our *Care information finder* helps you search by need, grouping together all resources on selected topics. Access the finder at: **www.mndassociation.org/careinfinder**

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 access our online form at:

<https://www.smartsurvey.co.uk/s/KDbooklet/>

Email your comments to: **infofeedback@mndassociation.org**

Or write to:

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“I have really come to terms with how it has changed my overall outlook upon life, as the impact of the condition on my father becomes more apparent.”



