SUPPORT | RESEARCH | AWARENESS

MPS III Sanfilippo

Information for individuals, parents and families

Society for Mucopolysaccharide Diseases
mpssociety.org.uk

In this booklet

There is huge variability within this condition and some people may experience only some of the symptoms, while the severity of those symptoms can also vary.

This booklet is produced by the **Society for Mucopolysaccharide Diseases (MPS Society)** and is designed to support those affected by MPS III Sanfilippo, their families and support network to understand its causes and effects.

While there is currently no cure for MPS III, this booklet explores the signs and symptoms of MPS III Sanfilippo and how to manage them. It draws on the experiences of patients, carers, families and medical professionals, as well as medical literature.

What is MPS III Sanfilippo?

MPS III, also known as Sanfilippo, was first identified by Dr Sanfilippo in 1963.

MPS III is a mucopolysaccharide disease. Mucopolysaccharides, also called glycosaminoglycans (GAGs), are long chains of sugar molecules used to build bones, cartilage, skin, tendons and other tissues in the body. In MPS III, the specific GAG that cannot be processed is heparan sulphate.

People with MPS III are either type A, B, C or D. Each type is missing, or has lower levels, of a specific enzyme. The enzyme's usual job is to clear used GAGS (waste deposits) in cells. Without these enzymes, heparan sulphate builds up in the cell and impairs its ability to function, until eventually it stops working. Gradually, over time more cells become affected and stop working, meaning that MPS conditions are progressive in nature.

- MPS III A is caused by missing or altered heparan N sulphatase
- MPS III B is caused by missing or low levels of alpha-Nacetylglucosaminidase
- MPS III C is caused by missing or altered acetyl-CoAlpha-glucosaminide acetyltransferase
- MPS III D is caused by missing or low levels of N-acetylglucosamine-6-sulphatase

How is MPS III Sanfilippo diagnosed?

Diagnosing MPS III Sanfilippo can take some time and typically requires looking at the person's medical history and symptoms. A physical exam and laboratory test are used to make a diagnosis.

Diagnosis of MPS III Sanfilippo usually involves three types of laboratory tests:

- A urine analysis will usually show excessive amounts of heparan sulphate and dermatan sulphate present in the urine.
- A blood test will show reduced enzyme activity.
- Finally, a genetic test will identify the IDUA gene alteration (mutation) to confirm diagnosis.

An initial diagnosis is often established during pre-school years when developmental delay becomes noticeable. Many of the early symptoms of MPS III are also commonly seen in many unaffected children of this age (e.g. diarrhoea, respiratory and ear infections and hyperactive behaviours). This overlap, along with the rarity of the condition, makes diagnosis very challenging.

There are around **150** individuals diagnosed in the UK (2023)

Once you have a confirmed diagnosis ...

There are several specialist centres in the UK where you can go to be tested and to see a specialist in MPS III, the most up to date list can be found on the MPS website: **mpssociety.org.uk/our-friends**

How common is MPS III Sanfilippo?

MPS III is considered a rare disease. A rare disease is a condition that occurs in less than 1 in 2000 people.

How is MPS III Sanfilippo inherited?

Genes are the unique set of instructions inside our bodies that make each of us an individual.

A **carrier** will not show symptoms but can pass the altered gene to their child.

Autosomal recessive inheritance is when both parents are carriers of the altered gene. When both parents are carriers of the altered MPS III gene (autosomal recessive) for each pregnancy there is a 25% (1 in 4) chance of having a child with MPS III. The chance of a baby inheriting MPS III is the same for every pregnancy.

Brothers and sisters of a person affected by MPS III might also be carriers of the condition and they may wish to be referred to their local genetic department about the potential risks in future pregnancies.

We have thousands of genes, and they are the blueprint for our growth and development, as well as controlling how our bodies function. If a particular gene is altered, then it may not work efficiently.

Genes are carried on structures called chromosomes. It is usual to have 23 pairs of chromosomes that are numbered in pairs from pair 1 to pair 22, plus one pair of sex chromosomes: XX for a female and XY for a male.

Each gene is a template for making a protein needed for the body to function.

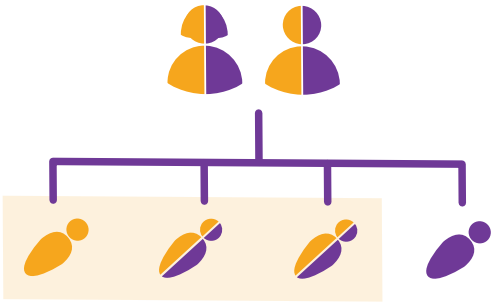
A child will inherit one set of chromosomes from their mother in the egg, and one set from their father in the sperm. Therefore, we each have two copies of each gene, one of which is inherited from each parent.

In a person with MPS III, both copies of the associated gene have genetic alterations (mutations). Their parents each carry one copy of the altered gene, but they do not show signs and symptoms of the disease. This is known as being a carrier.

Genetic counselling

MPS III is a genetically inherited condition and there is a risk of recurrence in future pregnancies for a couple with an affected child. Therefore, parents of children with MPS III should be referred to a genetic counsellor. The counsellor will provide non-directive advice on reproductive choices, the risk to close relatives, and will suggest whether the wider family should be informed.

In order to access genetic counselling, you can request a referral from either your specialist centre or your GP. Genetic services are provided by regional genetic centres.



Autosomal recessive inheritance pattern

- AFFECTED BY THE DISEASE 25%
- CARRIERS OF AFFECTED GENE 50%
- UNAFFECTED BY THE DISEASE 25%

Is there a test for MPS III Sanfilippo in pregnancy?

Amniocentesis involves testing a small sample of amniotic fluid.

Chorionic villus sampling involves testing a small sample of cells from where the placenta attaches to the uterus.

Unless there is a known genetic risk of MPS III, it is unlikely that a test in pregnancy would be offered to you. If you have a child with MPS III it is possible to have tests during any subsequent pregnancy.

It is important to contact your doctor if you are planning to become pregnant, or as soon as you suspect that you may be pregnant, if you wish to discuss your family history and options.

Both amniocentesis and chorionic villus sampling can be used to diagnose MPS III during pregnancy.

Amniocentesis involves using a needle to take a sample of fluid from around your baby. This fluid is then used to test the baby's DNA for genetic alterations in your family. Chorionic villus sampling is similar to amniocentesis but instead involves testing a sample of cells from where the placenta attaches to the uterus.

Pre-implantation genetic diagnosis (PGD) is an assisted fertility treatment. It may be possible to have PGD to avoid passing MPS III to the baby if you and your partner are known to be carriers of the condition. PGD involves checking the chromosomes of embryos for gene alterations before they are implanted in the womb, using IVF techniques. This is a complex process and requires referral from your regional genetics service.

Living with MPS III Sanfilippo

It is important to note that people affected by MPS III may not experience all the potential symptoms, and severity can vary from one person to another. The progression of MPS III is also variable, however, to help understand the condition it is often described in three broad stages:

First Stage

In early childhood (age 1-4 years) parents may notice some delays in their child's development. This may include delayed speech and language (often put down to glue ear), delays in crawling or walking, hyperactive behaviour and sleeplessness. Autistic traits are often reported as well as challenging behaviours.

Other signs can be very subtle and are often associated with common childhood illnesses. For example, frequent cold-like symptoms and diarrhoea.

The combination of these signs and symptoms may lead your child's doctors towards testing for an underlying condition.

Second Stage

As your child gets older the physical features of MPS III may become more noticeable.

Hyperactivity and challenging behaviours typically escalate and become more difficult to manage.

Over time certain skills and abilities are gradually lost. They will begin to slow down, become more unsteady on their feet and tire more quickly. They may lose the ability to use words. Your child may show signs of difficulty when eating and drinking, with increased coughing and risk of choking.

Third Stage

During the third stage there is a further neurological decline, with the loss of skills and abilities; mobility will be lost, with reliance on mobility aids and supportive equipment. The loss of mobility is often accompanied by a deterioration in the ability to eat and drink safely.

Care turns more towards managing health needs and symptom management.

MPS III Sanfilippo is a life-limiting condition; life expectancy is extremely varied.

Kayden lives with MPS III Sanfilippo

Kayden was born in 2014.Throughout his first nine years, he had respiratory issues and he frequently caught head colds and ear infections. I recall him often being prescribed antibiotics.
By 18 months, Kayden began to regress in his speech starting to babble like a baby again.
We also noticed his stomach was quite distended and looked bloated, so we made an appointment with our local GP where we were told that he was healthy and not to be concerned.

At age two, Kayden became very unwell and ran a temperature I couldn't control. Upon taking Kayden to hospital, he was kept in a ward overnight where I discussed my concerns on his regression with speech and past health. Kayden became very unwell and developed pneumonia which worried me greatly. It was a very challenging time for the whole family as Kayden ended up staying in hospital for two weeks. A consultant thankfully had awareness and knowledge of MPS and brought to my attention that Kayden's symptoms were similar to those with the condition. From there, tests were carried out to determine the diagnosis of MPS III Sanfilippo.

Kayden's diagnosis impacted all the family as you are essentially grieving the loss of your child's future and it is difficult to watch your child lose basic motor skills. It took time to process the condition and the severity of it. In Kayden's younger years, he was hyperactive and required supervision 24/7. Daily tasks like shopping could become difficult if Kayden was unsettled. He explores the world with his mouth (sensory related) which leads to any object within arm's reach to go into his mouth. When an extension for our property was completed, which included a bedroom and wet room for Kayden, we were also able to get a safe space bed which gave me peace of mind at night.

It can be isolating and challenging to care for a child with a genetic disorder, as every year brings new symptoms and challenges. Although, I do have support from family, Kayden's school and the MPS Society which make these challenges more manageable. We have hired a fantastic carer, who spends time with Kayden on Saturdays, which enables me to have quality time with my daughter.

The MPS Society has been a great support for us as a family and I feel I can contact our dedicated Support & Advocacy Officer at any time with my concerns or for advice.

Kayden enjoys sunshine and swimming. As a family, we like to go on holidays abroad and recently visited France. It was ideal for us as we could travel by boat and transport Kayden's essential equipment such as his feeding chair, wheelchair and travel safe space bed. We even had enough room in the van to bring a trampoline which allowed him to enjoy a bounce and to stretch his legs. Kayden also enjoys indoor soft play areas. In recent years this has become a blessing due to his poor balance. He loves watching his favourite TV shows, Peppa Pig and Little Einsteins, in his sensory bedroom which has been designed specifically for his ever-changing needs.

The MPS Society has been a great support for us as a family and I feel I can contact our dedicated Support & Advocacy Officer at any time with my concerns or for advice. They offer supportive information and can put us in touch with other families who have been affected by MPS. We follow the MPS Society UK Facebook page which keeps us up to date with any upcoming events and a private support group where families can ask questions and seek help from other families who are going through the same experiences.

Physical appearance

There may be changes in facial features, bone, and skeletal structure which become more noticeable with age.

Facial features

- Thick and coarse hair
- Excess body hair, often on the face and back
- Dark and bushy eyebrows which may meet in the middle
- Noses tend to be upturned and flat on the bridge
- Thick lips, broad gum ridges and enlarged tongue
- Teeth can be widely spaced with fragile enamel

Growth and height

Individuals with MPS III are generally of normal weight and height at birth and in early childhood. However, growth usually slows down and adults with MPS III are generally shorter than expected based on the height of their parents.

Prominent tummy (abdomen)

The liver and spleen are organs within the tummy (abdominal) area of the body. An enlarged liver and spleen can develop from the build-up of mucopolysaccharide deposits (GAGs). Although these organs can continue to function normally, the abdomen may be distended, and the pressure may affect eating and breathing.

Dental

Individuals with MPS III are at an increased risk of dental problems due to weakened enamel. Good dental hygiene is especially important to avoid the need for extractions and other dental treatment.

An individual with MPS III may be unable to communicate that they are in pain. Dental pain should be considered as a potential cause of unexplained behavioural changes and distress. Seeing a dentist regularly is important to ensure any dental problems are identified and treated as soon as possible.

Maintaining dental hygiene can be difficult in individuals with MPS III due to behaviour and sensory issues. It may be helpful to request referral to a specialist dentist who will be able to provide practical advice including the use of specialist toothbrushes.

Developmental milestones

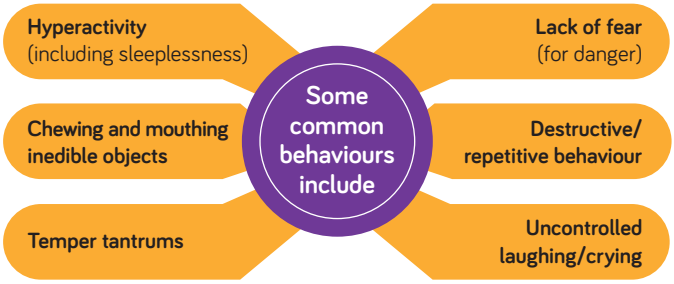
Individuals affected by MPS III will lose their intellectual ability progressively through the course of their life.

Early mental and motor skill development will be delayed. Children show a marked decline (regression) in learning from around 2 years of age, followed by eventual loss of skills. Some children may never learn to speak.

Children with MPS III will be most suited to placement in a school for special educational needs; with access to a sensory based curriculum and various therapies.

Behaviour

Challenging behaviour is a key feature of MPS III. This can be very difficult to manage and has a huge impact on family life. Individuals with MPS III require constant supervision and support.



Parents and carers will often seek support in managing their child's behaviour. It is likely that this will include a combination of medication and behavioural support services.

Continence

Individuals with MPS III are not likely to successfully toilet train. Those that do, will lose this skill as their condition progresses.

You can be referred to your local continence services who will be able to provide continence products. Individuals with MPS III will require more frequent changing due to the accumulation of GAGs in their urine/faeces. This can increase the risk of severe nappy rash and skin breakdown. It is important that continence supplies (such as nappies and continence pads) are provided with this in mind.

Puberty

Some individuals with MPS III may start puberty earlier than expected. If signs of early puberty are noticed, they may be referred to an endocrinologist.

Endocrinologist are doctors who specialise in glands and hormones.

Sleep

Sleeplessness is a very common feature of MPS III and has a huge impact on the individual and their family.

It is possible to support an individual's sleep with a combination of medication and specific routines, but this may have limited impact.

Families can access support from their clinical and social care teams to manage the 24hr needs associated with sleeplessness.

Neurological symptoms

The neurological decline seen in MPS III is often described as being similar to dementia. Individuals with MPS III are likely to display a number of neurological symptoms.

Seizures

Some people who are affected by MPS III may develop seizures or epilepsy.

There are different types of seizures, for example, absence episodes where the person may appear to be staring into space (with or without jerking or twitching movements), or more generalised convulsive (tonic-clonic) seizures. If worried, taking a video of the event to your doctor can be very helpful as MPS III progresses, seizures are more likely to develop.

Did you know?

Laughing and crying out of context can be a sign of seizure activity.

Seizures can be difficult to identify, and diagnose, because of the complex range of other neurological symptoms often present in individuals with MPS III. A Neurologist may request a range of assessments before discussing treatment options. Seizure activity can be well managed with the correct combination of medication.

Did you know?

In the later stages of MPS III, the body's natural temperature control mechanism may become damaged. The individual is not able to regulate their **body temperature**. Patients with reduced mobility often have cold hands and feet. Parents and carers need to be aware of this issue and adjust clothing and the environment as needed.

Movement disorder

Individuals with MPS III are likely to develop movement disorders. Symptoms and severity can be variable and may require treatment.

The terminology surrounding movement disorder can be quite difficult to understand and diagnosis can be difficult due to the complexity of the symptoms displayed.

Fine and gross motor skills

The development of fine motor skills in children with MPS III typically slows down around age 2 or 3 years. At this stage parents and carers may notice that their child does not meet the expected developmental milestones.

Usually in the second half of the first decade, children with MPS III become increasingly unsteady or clumsy when walking. Children will gradually lose their physical ability and will become more reliant on equipment such as specialist seating and mobility aids.

Ataxia describes poor muscle control that causes clumsy voluntary movements, co-ordination, balance and speech can be impacted.

Dyskinesia are involuntary, erratic, writhing movements of the face, arms, legs or trunk.

Dystonia – People with dystonia experience involuntary muscle spasms and contractions which often result in the body getting fixed temporarily in abnormal positions.

Sensory needs

Sensory integration is a broad term that describes an individual's ability to interact with the world around them. We interact with our environment using our senses – touch, sight, taste, smell and sound. As well as body awareness and movement.

We all need to be able to regulate the information we get from our sensory systems, so that we can cope in whatever environment we find ourselves in. We do this by taking in the sensory information that we need, while ignoring the sensory information that we don't need at any given time.

For example, in a shop we might drown out the background noise to prevent distraction and allow us to find the items that we need.

Sensory processing difficulties

Individuals with MPS III have difficulties in processing and organising sensory information. This can lead to individuals feeling overwhelmed and can impact behaviour. This is made more challenging due to communication difficulties.

Sensory processing difficulties can make everyday tasks difficult to tolerate such as showering, wearing certain clothes, eating, or brushing teeth.

With some understanding of how an individual copes in different environments, strategies for management can be tailored to meet their needs. It is not one size fits all.

Individuals may display different types of behaviour depending on the sensory processing difficulties they are experiencing.

Sensory seeking behaviours – When a person struggles to take in and organise sensory information, they may seek out additional sensory experiences to help regulate themselves. This is known as sensory seeking. This can present as chewing, rocking, flapping, hyperactivity, hitting themselves or banging their head on objects.

Sensory avoidance – These types of behaviour are displayed when an individual is unable to ignore background sensory input (this is sometimes called sensory overload). Sensory overload leads to what is known as sensory avoidance, where the child withdraws and struggles to participate. This can present as anxiety and distress.

Ears

Some degree of deafness is common. It may be conductive deafness, nerve deafness or both and can be made worse by frequent ear infections.

Conductive deafness is when sound waves that travel through the ear canal, drum and the middle ear are impaired.

Glue ear is where the middle ear fills with glue-like fluid, blocking the transmission of sound waves. Chronic glue ear can lead to damage to the ear drums, which contributes to hearing loss and recurrent ear canal infections.

Nerve deafness is damage to the tiny hair cells in the inner ear. It may happen at the same time as conductive deafness (mixed deafness).

Management

When considering the management of hearing loss, it is important to consider how the individual will cope with any strategies put in place.

- Use of communication aids (such as signs and symbols)
- Glue ear can be treated through surgery by inserting grommets (also known as T-tubes) into the ear.
- Nerve deafness is usually managed by fitting hearing aids.
- Mixed deafness can be managed by grommets/ T-tubes and hearing aids.
- Bone-anchored hearing aids and cochlear implants, can be used in special circumstances.

Eyes

Individuals with MPS III may have some level of visual impairment, this can be associated with damage to the optic nerve and the retina. Typically, vision can be difficult to assess due to impaired communication and inability to carry out assessments.

Night blindness is known to be a common feature of MPS III. Often children with MPS III do not want to sleep in the dark or are afraid when waking up at night.

Parents and supporting professionals should be aware of potential issues with vision and the impact that this might have on communication and quality of life.

Bones and joints

People with MPS III present with a range of progressive bone and joint issues that may impact mobility and cause significant pain and distress. Individuals should be assessed by an orthopaedic doctor who will discuss treatment options.

Kyphosis – curvature of the spine which causes the top of the back to appear more rounded

Scoliosis – abnormal twisting and curvature of the spine which may cause one shoulder to be higher than the other

Mobility

The combination of neurological and physical decline results in progressive loss of mobility. Individuals with MPS III will become increasingly unsteady on their feet and will eventually lose the ability to walk.

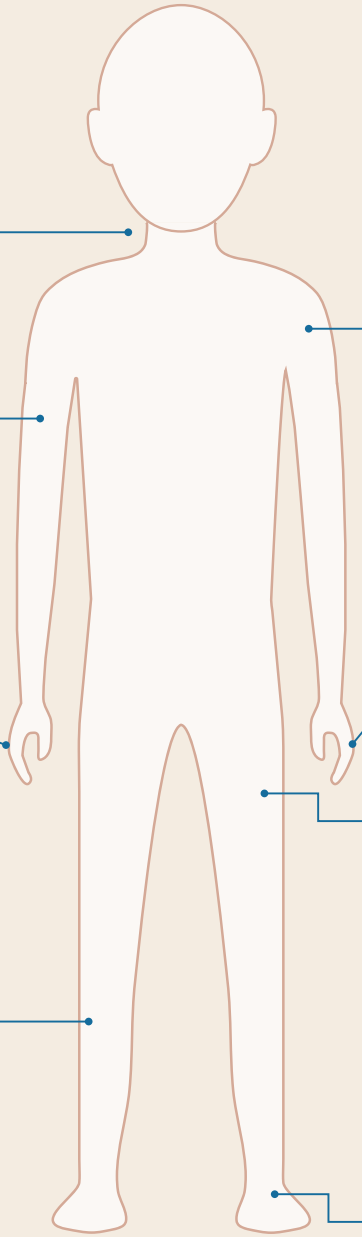
It is important that individuals with MPS III are referred for therapy to support their daily activities and maintain their comfort levels.

Occupational therapy – an occupational therapist will assess for equipment and home adaptations. This may include moving and handling equipment (such as a hoist). They may also provide specialist seating and buggies/wheelchairs.

Physiotherapy – a physiotherapist will support individuals with MPS III in relation to their movement and function. They will assess and develop a programme that will help to maintain range of movement and comfort levels. Please refer to our booklet on 'How physiotherapy can help with the physical symptoms of MPS III Sanfilippo.'

Hydrotherapy – physiotherapy exercises carried out in warm water can help to strengthen muscles, relieve pain and reduce joint stiffness.

Low bone mass and vitamin D deficiency are common and increase the risk of weakened bones (osteoporosis) and fractures.



Spine
Progressive curvature of the spine may develop with an impact on posture and mobility and may cause of pain and discomfort.

Joint stiffness
Progressive joint stiffness can impact daily tasks such as getting dressed.

Low bone mass and vitamin D deficiency
are common and increase the risk of weakened bones (osteoporosis) and fractures.

Trigger finger
Can make it difficult to bend the affected finger or thumb and may appear to be 'locked' in a bent position. This can be painful and may require treatment.

Hands
Clawing of the hands can occur.

Hips
Dislocated hips and deterioration of the hip joint can cause significant pain.

Legs and feet
Knock-knees *(Genu Valgum)* is a common feature. Legs may turn inward, causing the appearance of the knees to be touching while the ankles remain apart.

Toe walking
The Achilles tendon, which connects the calf muscle to the heel, can become tight causing the individual to walk on their toes. This can impact posture and balance. Flat and broad feet with curved toes may also be a symptom.

Nutrition

Managing the nutrition of an individual with MPS III is very important. An individual's needs in relation to nutrition will change significantly over time. It is important that your child is referred to a dietician and speech and language therapist to assess their needs.

A gastrostomy is a surgical opening through which a feeding tube is placed directly into the stomach so that nutrition can be delivered bypassing the mouth and throat.

In a **Jejunostomy**, the feeding device is situated in the lower intestine, rather than the stomach.

Feeding

Individuals with MPS III will develop progressive feeding difficulties.

Sensory issues can have an impact on an individual's food preferences. If this is causing difficulties in maintaining nutrition (your child is losing weight), a dietitian may be able to provide strategies to support you.

One of the most significant changes is the deterioration in the ability to swallow. Parents and carers may notice that their child is slow to swallow mouthfuls of food or is coughing during their meals.

This increases the risk of food entering the lungs and causing infection. It is important that the individual is referred to a Speech and Language Therapist (SALT) to have their swallow assessed.

The SALT will provide advice about safe texture of food and feeding strategies to make mealtimes safer.

Over time the individual's swallow will deteriorate significantly, and oral feeding will no longer be safe. It is likely to be necessary to insert a feeding tube to supplement nutrition and reduce the risk of chest infections. The clinical teams will discuss the timing and most appropriate device. A feeding tube may initially be used to 'top up' a child's nutrition, or assist with administering medication, particularly if the child is unwell. Parents and carers will be trained to use and maintain these devices with support from local clinical teams.

Digestion

Progressive digestive issues in MPS III can be very disruptive and difficult to manage.

Diarrhoea (loose stools) is a common feature in people with MPS III that can become increasingly problematic as time goes on. Treatment of this can be difficult to balance as constipation can also be a feature of the condition. Finding the correct combination of medication and nutrition can take some trial and error.

In the later stages of the condition, the body's ability to process and absorb nutrients is impacted. Maintaining nutrition becomes a focus and individuals will require intensive input from a dietician to manage feeding. It is important for parents, carers, and clinical teams to monitor weight and make changes to feeding programmes as necessary.

As the condition progresses, you may notice that the person with MPS III is having difficulty eating and drinking. Signs of difficulty could include an increase in the time it takes to eat, pooling or holding of food and drinks in the mouth, coughing or choking while eating or drinking. A video fluoroscopy is a special kind of Xray that looks at your ability to swallow safely.

The procedure involves giving different foods and drinks, coated in a small amount of Barium whilst the doctors use Xray to see how the swallow is performing in real time. A Speech and Language Therapist will also review the results who will then be able to advise on types and textures of food and drink to avoid and set out a care plan as needed.

Dysphagia is the medical term for difficulty swallowing.

Lungs and breathing

Individuals with MPS III have narrowing of their airways as well as breathing issues associated with loss of mobility and neurological decline.

Airway

Frequent coughs and colds are a common early feature of MPS III. In the early years, these tend not to cause significant problems; but as the condition progresses, they may be more likely to cause serious illness.

Individuals with MPS III often have noisy breathing which is caused by narrowing of the airways as well as enlarged tonsils and adenoids. This occurs even when there is no illness or infection. It is common for individuals with MPS III to have their tonsils and adenoids removed.

Parents may notice that there are gaps in breathing when their child is asleep, this is called Apnoea and should be investigated by a respiratory doctor.

As the condition progresses, respiratory tract infections become more frequent. Regular antibiotics may be recommended to manage the risk of a serious infection (pneumonia).

The airway is lined with mucous and saliva (secretions) that help it to function correctly. As the condition progresses, these secretions become thicker and more difficult to clear through swallowing and coughing. This will become increasingly problematic and support, such as suctioning, is likely to be required. Respiratory physiotherapists can also provide support in managing secretions.

In the advanced stages of the condition, some individuals may need more breathing support with the use of oxygen throughout the day and night.

Doctors may recommend a sleep study to assess Apnoea symptoms. Some nighttime breathing support, such as Continuous Positive Airway Pressure (CPAP), may be recommended to improve the quality of sleep and maintain safe oxygen levels.

Heart 

The build-up of waste materials in MPS III can result in some specific issues with the heart. These often do not have an impact of daily life but may require monitoring. It is rare for cardiac issues in MPS III to require treatment, but clinical teams will assess each individual and recommend treatment if necessary.

Valves – the valves of the heart can become damaged, resulting in leaking (regurgitation) or blood between the chambers of the heart.

Cardiomyopathy describes the thickening of the heart muscle (or walls of the heart).

Arrythmia – irregular rhythms may be picked up if an individual has an electrocardiogram (ECG). They may be experienced as palpitations or chest discomfort.

Having an operation

Complications during anaesthesia and surgery can occur in patients with MPS III. It is important that the patient is seen by an anaesthetist experienced in MPS conditions for pre-operative assessment and that surgeries are carefully planned in consultation with specialist teams. See mpssociety.org.uk for an anaesthetic guide.

When planning a surgery, there are other practical and wellbeing considerations. These may include:

Reassurance and support
- Allowing a parent or carer to provide support during the induction of anaesthesia
- Care staff to meet care needs before and after surgery

Sensory
- Providing a low-stimulus environment
- The use of distraction techniques and items

Safety
- The ability to secure/close doors to reduce the risk of the patient escaping
- Consideration of safety concerns with regards to impulsivity and hyperactivity

Visit mpssociety.org.uk/our-friends to find out about specialist centres who work with people with lysosomal storage diseases.

Care considerations

Caring for individuals with MPS III is complex. Families will find that all areas of their life are impacted, and they may need support in relation to finance, housing, special education, access to equipment and navigating the health and social care system. The MPS Society Support and Advocacy Team are experienced in supporting families with MPS III. Members of the MPS Society will be connected with a Support and Advocacy Officer who works specifically with MPS III.

Supportive care

The impact the of MPS III on an individual and their family is significant and often involves around the clock care.

Accessing support is essential to protect the health and wellbeing of all those involved. This may include development of a care package with the involvement of health and social care teams.

Palliative care

Palliative care is the support and treatment for people with life-limiting illness (one that is progressive and/or life-threatening). Palliative care can be delivered at any stage of illness, alongside treatments and therapies.

Your specialist team can refer you to palliative care and hospice services at any point following diagnosis to access support, such as:

- Respite
- Therapeutic services
- Counselling
- Pain management and symptom control
- Sibling support
- Support at home

If you would like more information about palliative and supportive services, you can contact the MPS Society Support and Advocacy Team: mpssociety.org.uk/advocacy

Where can I get more information and support?

The Society for Mucopolysaccharide Diseases (MPS Society) is the only registered UK charity providing professional support to individuals and families affected by MPS and related lysosomal storage diseases throughout the UK.

Further information booklets and other resources about MPS, Fabry and related diseases are available from **mpssociety.org.uk**

Our Support and Advocacy team have specialist knowledge of these diseases and a background in social care. We are here for you whenever you need us.

Phone us on **0345 389 9901** Mon to Fri 9am–5pm

Outside these hours you can call us on **07712 653 258**
Mon to Fri 5pm–10pm
Sat and Sun 9am–5pm

Email us at **advocacy@mpssociety.org.uk**

Members in Northern Ireland and Scotland can contact their Support and Advocacy Officer on **07786 258 336**

Please contact us on **0345 389 9901** or visit our website **mpssociety.org.uk/advocacy** if you would like to find out more about how the MPS Society can support you.

Every effort has been made to ensure that the information in this booklet was accurate and up to date at the time of going to press. This booklet is not intended as a substitute for professional medical advice and the MPS Society and other contributors cannot take responsibility for actions taken as a result of this information.

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Registered as a Company limited by guarantee in England & Wales No. 7726882
Registered as a Charity in Scotland No. SCO41012

This booklet was written by MPS Society UK with input from clinical specialists.
Production was supported with unrestricted funding from Ultragenyx Europe GmbH, part of the Ultragenyx Group of companies, Denali Therapeutics and JCR Pharmaceuticals.