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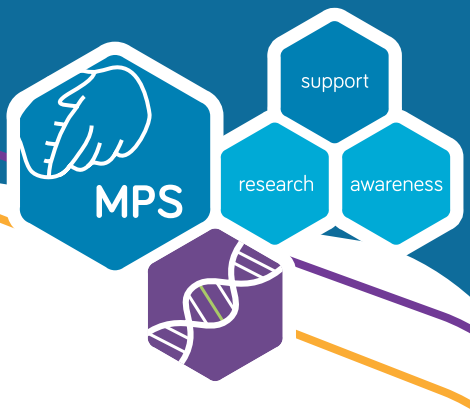

How physiotherapy can help with the physical symptoms of MPS III Sanfilippo

A guide for families, physiotherapists and other supporting professionals

Society for Mucopolysaccharide Diseases

mpssociety.org.uk

Contents

Disclaimer

This publication has been prepared by the MPS Society in collaboration with expert physiotherapists.

This publication does not provide medical advice, it is intended for information purposes only. Always seek the advice of your specialist metabolic centre with any questions you may have regarding your medical condition or treatment.

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Acknowledgements

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Introduction

MPS III (Sanfilippo) is a rare inherited metabolic disorder caused by a lack of an enzyme in the cells that breaks down large sugar molecules (known as heparan sulfate) and recycles them. If these large molecules are not broken down and removed, they build up in the cells and cause damage, particularly in the central nervous system, including the brain.[1-3]

The prominent features of MPS III are neurological decline and behavioural issues, but the buildup of heparan sulfate can affect tissues and organs throughout the body including the bones, muscles and joints. The rate of disease progression and severity can vary between individuals, but most commonly follows three phases starting in early childhood.[1-4]

There are four types of MPS III, which are classified as A, B, C and D. Each type is caused by the lack or reduced activity of a specific enzyme.[3]

MPS III can affect tissues and organs throughout the body including the bones, muscles and joints.

Pre-diagnosis ages 1–4

- The child usually appears healthy at birth, with early symptoms starting between the ages of 1 and 4 years
- The first symptom is often developmental delay, particularly delayed speech
- The child may never complete toilet training and may suffer from diarrhoea
- Recurrent ear, nose and throat infections are common[1]

Behavioural disturbance ages 3–10

- The second stage usually begins around the age of 3 to 4 years and is marked by the development of severe behavioural problems
- These often start with temper tantrums, followed by hyperactivity and a reduced attention span
- The child may start to show aggression and be destructive
- During this phase sleep is often disturbed
- Further effects of MPS III on the brain and movement start to become apparent, with loss of understanding and speech often occurring before problems with walking[1,2]

Neurological and physical decline age 10+

- During the third and final stage of disease behavioural issues are replaced with the continued loss of skills, with most children becoming fully dependent on care in their teenage years[4]
- Walking becomes difficult due to loss of balance and stiffness in the joints and muscles; most will be reliant on a wheelchair by their mid-teens
- The effects of MPS III on swallowing will mean that tube feeding will usually become necessary
- Seizures can become troublesome during this stage[1]

Hearing loss is common, and eyesight may be affected, particularly in the late stages of the disease. Although the symptoms of MPS III usually start in early childhood, some individuals will have a slower progressing disease and not develop symptoms until later in life.[3]

Physiotherapy in the care of MPS III

MPS III is a complex condition affecting multiple systems within the body and as a result needs a multidisciplinary approach to treatment. This means all the different healthcare specialities involved with the individual's care need to work together to provide the best possible support. The physiotherapist is a vital part of this team approach.

Treating those with MPS III can be particularly complex and the physiotherapist will carefully consider the specific difficulties experienced by the individual, and their stage of disease, before thinking about treatment. Problems with movement and posture need to be considered against the background of other MPS III symptoms including behavioural and learning difficulties, developmental delay, hyperactivity, sleep difficulties, seizures and loss of skills.

The aim of physiotherapy is to maintain function and independence for as long as possible.

Before thinking about physiotherapy treatment, a physiotherapist will need to consider the underlying disease process, such as:

Why are there cognitive difficulties?
What happens to bones?
Why are there joint contractures and stiffness?
How do the muscles become weak?
Why is there pain?

Bones and muscles

MPS III is associated with bone and muscle abnormalities, although these are usually less severe than in other MPS diseases. The build-up of heparan sulfate in bones, cartilage, ligaments and muscles leads to joint stiffness or contractures, altered posture and curvature of the spine. Hip joints are usually shallow and can therefore be more prone to wear and tear on the joint surface. The top of the thigh bone where it forms the hip joint can also be 'crumbly' because of poor blood supply (avascular necrosis), making it more likely to deteriorate.[3, 5] As MPS III progresses, those affected can lose muscle strength and flexibility in their joints[6] and the progressive hip deformity can cause pain and discomfort on movement.[3]

The effects of MPS III on the brain can also affect movement and muscles, resulting in the inability to walk normally (gait disorders), hyperactive reflexes, and tight muscles with an inability to control those muscles (spasticity).[3]

Bone, joint and muscle symptoms do not usually appear until after a child is showing developmental delays and behavioural problems.[3]

Behaviour and learning

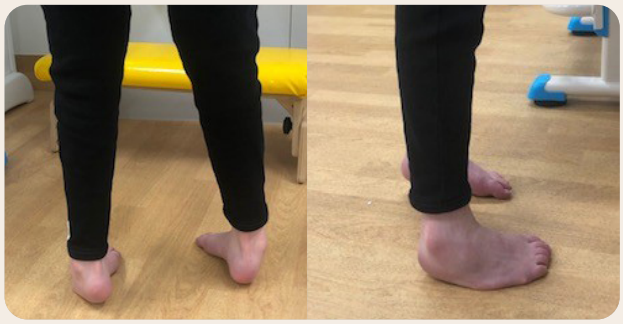
MPS III can cause severe behavioural problems that are characterized mainly by extremely hyperactive behaviour.[1] Children may also exhibit aggression and temper tantrums with no full understanding of danger or fear. Behaviour becomes less problematic with age as the effects of MPS III result in decreased mobility and initiative.[3]

The mental capacity of children with MPS III will vary. Children with the severe or 'classical' type of MPS III will have a normal to near normal development up to the age of 2 years, followed by a slowing and stagnation of development at around the age of 3 to 4 years. This is then followed by the progressive loss of mental capacity.

Adults with MPS III and children with less severe disease may have a stable learning difficulty for many years.[4]

Toe walking

Toe walking is a complication that happens in some children with MPS III. The child walks with the balls and toes, but not the heels, of their feet touching the ground. Toe walking is common in children learning to walk and they usually outgrow it by the age of two but children with MPS III may continue to do it. Although toe walking may trouble parents, most children are still able to mobilise well, and it should not be a cause for concern.



Why is physiotherapy important?

The aim of physiotherapy is to maintain function and independence for as long as possible and quality of life is always a priority. As those with MPS III have complex needs, a physiotherapist works closely with other professionals such as occupational therapists, psychologists and various other specialists to support the individual. Appropriate strategies are developed in collaboration with the child or adult and their family.[7] Another important aspect of the physiotherapist's role is to manage and optimise the posture of those with MPS III.

A physiotherapist also plays an important role in monitoring those with MPS III and can alert other professionals involved in their care to any changes or potential problems that need to be addressed.[7]

Regular review by a physiotherapist and wider specialist team based at a specialist centre is recommended.

How physiotherapy can help

Maintaining good posture, range of movement and strength at hip, knee and ankle joints is a key objective for any physiotherapy care plan. Both children and adults with MPS III are encouraged to keep as active as possible and early use of special equipment to improve posture is recommended.

There are several aspects to consider when treating children with MPS III, including:

- Hip pain
- Hyperactivity and short attention span
- Ability to understand what is happening and why
- Challenging behaviour like aggression and tantrums
- Movement disorders with limb jerking, kicking and muscle spasms

Role of the physiotherapist

A physiotherapist will evaluate an individual's physical abilities and needs, design an appropriate physical programme, and monitor this over time. There are several aspects to consider when designing the physical programme.

The physiotherapist will also closely monitor any physical changes and assess new symptoms of pain, weakness and altered posture to provide the necessary assistance and advice.

They will also ensure the family have referrals to appropriate services such as an occupational therapist who can assist with both static seating within the home and accessing appropriate wheelchairs.

> The aim is to maintain function and independence for as long as possible. Quality of life is always a priority.

Physiotherapy for children

Physiotherapy assessment

The first thing a physiotherapist will do is familiarise themselves with the child's medical history. They may also need to discuss this with other professionals, so they are aware of any safety precautions they should take when delivering physiotherapy.[7]

A physiotherapist will do a thorough assessment of the child's physical abilities before devising a physical therapy plan.

It may not be possible to perform all assessments to measure range or test power due to the behavioural and learning difficulties of children with MPS III, but function can be assessed by observing the child.

Physical examination by a physiotherapist will include the following:

Not all these tests or observations may be carried out due to behavioural and learning issues. Each child will be assessed according to their needs.

- Comprehensive assessment of gross motor abilities (these involve the whole body and use large muscles that are working in activities like sitting, standing and walking)
- Gait (manner of walking, looking for toe walking or any unevenness)
- Comprehensive assessment of fine motor abilities (these use small muscles of the hands, fingers and wrists that are involved in dexterity, such as holding a pencil or playing with bricks)
- Range of movement including restrictions and any pain on movement
- Strength testing/muscle power – often by observation of movement and looking for specific weaknesses
- Posture
- Exercise tolerance (how much physical exercise the child can manage)

A thorough physiotherapy assessment is the first step in any physical therapy plan.

Physiotherapy interventions

A physiotherapist will use a range of different interventions according to the care plan they have designed specifically for the child. This plan may need to be modified over time. These interventions include:

- **Active exercises** – to protect joints
  - These are movements performed by the child on their own
  - Examples of active exercises are walking, cycling (use of a tricycle), movements in water (bath or a pool)
- **Balance and strengthening exercises** – to maintain function
- **Active stretches** – to maintain range of movement, often using creative ideas
  - This includes the use of games and play to achieve stretches, e.g., reaching games
  - Helps to maintain range of movement
  - Encourages standing and walking

A physiotherapist has an important role within the child's care team in monitoring the child's changing abilities and needs and identifying problems early.

- **Movement in water**– a holistic activity with multiple benefits[6]
  - This is important to include in any physiotherapy programme
  - Having access to swimming pools, warm water pools and baths is a great benefit to a child's mobility
  - It allows various interventions to be done in a warm, relaxing and safe environment that also supports the body and joints.[6] The physiotherapist can increase resistance by adding floatation devices or asking the child to walk at different depths
- **Home exercise programmes** – to maintain physiotherapy benefits and establish a routine.
  - Continuing exercises outside of a physiotherapy appointment will help maintain the child's progress and establish a routine
  - The physiotherapist will liaise with parents, carers and community teams to offer advice and training to encourage active movements and stretches at home
- **Passive movements** – exercises performed by the child, but assisted by the physiotherapist

What are passive movements?

These are movements of a limb or extremity done rhythmically and smoothly through the available range of motion of a joint by the child with the help of the physiotherapist.

Passive stretches are different. These involve the limb or extremity being moved beyond its available range of motion. Tissues of children and adults with MPS III do not 'give' with stretch, **so passive stretches are not advised as they do not produce good outcomes.**

Physiotherapy is more than just exercises

A physiotherapist can also advise on what types of equipment may help the child when walking, travelling or sleeping.

Equipment provision

A physiotherapist can also help with arranging special equipment for the child to improve body symmetry and support function, including:

- Special footwear (called 'orthotic footwear') or some external appliances (collectively called 'orthoses') that may help to optimise foot position and assist with walking (splints are not well tolerated by most children with MPS III because they are uncomfortable)
- Seating, standing frames (require careful consideration) and sleep systems
- Mobility aids (walking frames etc.)

They can advise on issues related to safety, such as the use of buggies and helmets and safety in vehicles. In association with other professionals, such as occupational therapists, advice on suitable types of seat belts, car seats and related items, can be given on an individual basis.

Advice for schools and families

School years

Physiotherapy considerations for children attending school

Maintaining mobility
Children with MPS III are often kept in chairs for safety while at school but including movement into their day is important to reduce joint stiffness. These children will lose mobility over time, so it is very important to support and maintain their mobility for as long as possible.

Supervised time on their feet is very important for children with MPS III, but it is also important to remember that:

- The child may have no sense of danger
- Their learning and behavioural issues may make it difficult for them to follow instructions
- They may have balance problems and be at risk of falling

A physiotherapist can advise schools about how best to maintain the child's mobility, while also keeping them safe. For example, regular breaks from sitting in a chair could be timetabled into the daily routine e.g., a walk around at changing and break times. Walking may be supported or using a piece of equipment such as a handling belt or walking aid, which a physiotherapist can also advise on.

Education Health and Care Plan
A physiotherapist can contribute to Education Health and Care plans and identify support needed for the child at school. They can advise on frequency of position changes and activities to build into the school timetable.

Other advice
Children with MPS III often have specific problems which a physiotherapist can make schools aware of. It is important that schools know how to recognise if the child is in pain. The child may not be able to express pain, but staff can be trained to look for signs of pain such as lop-sidedness or grimacing.

Children may show toe walking or their joints may become 'stuck' in one position. Toe walking is common but has no major impact on mobility. Becoming 'stuck' may make it difficult to put on shoes or position feet on wheelchair footplates and a physiotherapist can offer advice to deal with this.

A physiotherapist can liaise with your child's school to make sure they support your child's mobility and understand common problems.

Corrective surgery

Orthopaedic surgery is rare in children with MPS III[8], but may be considered when pain is an issue. If surgery is being considered for a child, a physiotherapist plays an important role in the child's care team who will take a holistic approach and consider various factors associated with surgery, including:

- **The reasons for surgery, and if there are other ways to solve the problem**
- **If there is any equipment that may help, either instead of or alongside surgery**
- **If the child can cope with rehabilitation after surgery**
- **If the family can cope with the surgery and the child's care afterwards**
- **If the family has realistic expectations of what surgery can achieve**

Children with advanced disease

As MPS III naturally progresses to the third stage, the child's mobility will deteriorate and they will become more dependent on wheelchairs and parents or carers for assistance. A physiotherapist will help to maintain the child's available range of motion by encouraging flexibility and mobility of their joints for as long as possible. During this stage a child may not be able to make these movements independently and will therefore need the physiotherapist to perform the movements for them.

Any movement can help to reduce joint stiffness and stop joints 'freezing' from lack of movement. A physiotherapist alongside an occupational therapist will also advise on how to maintain an optimal position for the child when they are sitting or lying.

Physiotherapy still has an important role in the care of children with severe mobility issues.

Adults with MPS III

Children with MPS III may survive into early adulthood, as death usually occurs in the second or third decade of life. In addition, those with milder disease may not start to develop symptoms until mid-to-late adulthood.[3]

Most adult specialist centres have a specialist physiotherapist who will work closely with the care team to provide advice on the physical needs of each adult with MPS III, liaising closely with community services and families.

Physiotherapy assessments, interventions and monitoring will be similar to that provided for children. Additional community support, including learning disability teams, carers, day centres, respite, social services, palliative and hospice care are available and should be considered when providing support to an individual with MPS III.

Useful contacts

Specialist centres – Children

Birmingham Women's and Children's Hospital

Physiotherapy Department
Ground Floor, Parson's House
Birmingham Children's Hospital
Steelhouse Lane
Birmingham
B4 6NH

Tel: 0121 333 9480

bwc.nhs.uk/physiotherapy-and-orthotics

Great Ormond Street Hospital

Physiotherapy Department
Level 5, Frontage Building
Great Ormond Street Hospital
London
WC1N 3JH

Tel: 020 7405 9200 extension 5144

www.gosh.nhs.uk/wards-and-departments/departments/clinical-support-services/physiotherapy/

Royal Manchester Children's Hospital

Paediatric Physiotherapy
Therapy and Dietetic Services
Ground Floor
Royal Manchester Children's Hospital
Oxford Road
Manchester
M13 9WL

Tel: 0161 276 1234
Tel: 0161 701 2640

mft.nhs.uk/rmch

Specialist centres – Adults

Addenbrooke's Hospital

Cambridge University Hospitals
NHS Foundation Trust
Lysosomal Disorders Unit, Box 135, Biomedical Campus
Hills Road, Cambridge
CB2 0QQ

Tel: 01223 245 151

www.cuh.nhs.uk/our-services/inherited-metabolic-disorders/

National Hospital for Neurology and Neurosurgery

Charles Dent Metabolic Unit
UCLH NHS Foundation Trust
Queen Square, Box 102
London
WC1N 3BG

Tel: 0203 448 4778

www.uclh.nhs.uk/patients-and-visitors/patient-information-pages/charles-dent-metabolic-unit

Queen Elizabeth Hospital Birmingham

Mindelsohn Way, Edgbaston
Birmingham
B15 2GW

Tel: 0121 371 2000

www.archive.uhb.nhs.uk/physiotherapy.htm or
www.uhb.nhs.uk/physiotherapy.htm

Royal Free Hospital

Pond Street
London
NW3 2QG

Tel: 020 7794 0500

www.royalfree.nhs.uk/services/services-a-z/occupational-health-and-wellbeing-services/physiotherapy-service/

Salford Royal Hospital

Mark Holland Metabolic Unit
NW2, 2nd Floor, Ladywell Building
Salford Royal NHS Foundation Trust
Stott Lane
Salford
M6 8HD

Tel: 0161 206 1080

www.srft.nhs.uk/about-us/depts/aimd/

Support, advice and advocacy services

The MPS Society provides support for families and individuals with MPS III. The team can support families with Education, Health and Care Plans (EHC), disability benefit applications and with health and social care.

Active listening service and telephone helpline

Education, Health and Care plans and school talks

Peer-to-peer befriending service

Help accessing disability benefits

Referrals to social care for services or respite

Support with end of life, loss and bereavement

Information on housing, home adaptations and specialist equipment

Independent living and transition support

Linking with specialist clinical centres and signposting to expert advice

For all services, contact our support and advocacy team

www.mpssociety.org.uk/advocacy

advocacy@mpssociety.org.uk

0345 389 9901

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My physiotherapy team

Name: _____

Role: _____

Location: _____

Contact tel/email: _____

Appointments

Date: _____
Time: _____
Location: _____
With: _____

Date: _____
Time: _____
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