

Learning Disability Practice

Sensing what's Important: determining parental preferences of children with Hunters Syndrome/Sanfilippo syndrome for when they are receiving treatment in hospital.

--Manuscript Draft--

Manuscript Number:	LDP1790R1
Article Type:	Research
Full Title:	Sensing what's Important: determining parental preferences of children with Hunters Syndrome/Sanfilippo syndrome for when they are receiving treatment in hospital.
Corresponding Author:	Andrea Page, PhD., MSc., PGCert., RNLD., RT., SFHEA Birmingham City University Birmingham, UNITED KINGDOM
Other Authors:	Charlotte Gayson, MSc., BSc (Hons). Nicola Vanes, RGN (Level 1)., ODP Paul Ashmore, NNEB, Dip HE., RNC. Andrew McDonnell, PhD., MSc., BSc.
Abstract:	Nurses hold children to administer treatment, prevent treatment interference and to undertake clinical assessment. It is important that clinical holding is within a framework which includes effective preparation. Effective preparation successfully reduces the need for undue force when using clinical holding. This includes the introduction of other strategies. This article explores the opinion of parents when interviewed about their child's senses (any of the faculties by which stimuli from outside or inside the body are received and felt, for example the faculties of hearing, sight, smell, touch, taste, and equilibrium) with the aim of making their child's stay with the NIHR/CRF more successful and thereby reduce any behaviours of concern which make it difficult for nurses to effectively treat the child or young person. It is hoped that our findings could be used for all children and young people who have autistic traits when they are receiving treatment in hospital. Five semi structured interviews took place with parents, which were then analysed using thematic analysis. The overarching themes were person centred planning, confidence and interview amendments.
Keywords:	Clinical trials; Hunters syndrome; Sanfilippo syndrome; Sensory profile; qualitative research; children and young people; Autism; behaviours of concern; clinical holding; effective preparation
Additional Information:	
Question	Response
Please confirm that you have read and agree to our Publisher's Agreement that is available here	Yes
Have you submitted this manuscript elsewhere?	No
Has this manuscript already been published?	No
Has the research received ethical permission and comply with ethical guidelines?	Yes
Do you have copyright for all the images, graphics and figures included with your submission?	Yes
Is the research more than 5 years old?	No
What is the word count of your document including references but excluding the	3138

abstract?	
What is the word count of your document excluding both references and the abstract?	2848
Author Comments:	<p>Dear Colin Parish</p> <p>With regards to LDP 1790 we have revised our submission as requested on 12th December 2016.</p> <p>Please thank your reviewers for their comments, they were very useful.</p> <p>We have revised the aim of this article to improve clarity and included the interview script. Details about how the interview schedule was developed and informed by knowledge and experience have now been provided within the introduction. We have added information about the independent ethical process followed. We have added detail about the small sample size and how participants were recruited. Thank you for identifying the number of quotes from parents - we have now included quotes from all 5 parents We have amended the section about thematic analysis to ensure clarity about how the themes were arrived at. The themes are now identified at the onset of findings. The discussion has now been developed with reference to appropriate literature. The limitations of the study have been highlighted with the discussion. The conclusion has been strengthened. We have included a glossary about the two syndromes</p> <p>We look forward to hearing back from you and receiving feedback about this resubmission</p> <p>Best wishes</p> <p>Andrea Page On behalf of the writing team</p>

Sensing what's Important: determining parental preferences of children with Hunters

Syndrome/Sanfilippo Syndrome for when they are receiving treatment in hospital.

Introduction

Introduction

This article explores the opinion of parents when interviewed about their child's senses (any of the faculties by which stimuli from outside or inside the body are received and felt, e.g. hearing, sight, smell, touch, taste, and equilibrium) with the aim of making their child's stay in hospital more successful and thereby reduce any behaviours of concern which make it difficult for nurses to effectively treat the child or young person. The aim of this article is to explore perceptions about using the 'stress sensory preferences interview and recommendations evaluation' (SsPIRE) to elicit this information. This includes understanding the form and questions covered within the profile, how this can impact upon the care provided once strategies most appropriate for their child have been recognised and confidence in staff to think proactively about plans for sensory care and managing behaviours of concern.

Behaviours of concern may prove difficult for nurses to manage a child's quality of care and the intensity of such behaviours can put parents, carers and nurses at risk of injury (Page and McDonnell, 2013; Chan et al, 2011; McDonnell, 2010). Many children diagnosed with life limiting conditions may display behaviours of concern when receiving medical care from healthcare professionals (Page et al, 2015).

The National Institute for Health Research/Wellcome Trust Clinical Research Facility (NIHR/CRF) at Birmingham Children's hospital supports the delivery of academic and commercial research studies, enabling children and young people to take part in high quality, safe and ethically approved clinical research. In supporting clinical research the NIHR/CRF aims to improve clinical care and the lives of children, young people and their families. Many of the children and young people undertaking such clinical research are doing so because they have a life limiting medical condition which often includes conditions and syndromes which gives rise to conditions and syndromes which challenge healthcare practitioners when they are administering medical care (Page et al, 2015). Some of the children and young people visiting the NIHR/CRF have disabilities and traits of autism as part of their

condition, for example those with Hunters Syndrome or Sanfilippo Syndrome. [See Glossary for information about Hunters Syndrome and Sanfilippo Syndrome](#)

There are a vast number of websites and resources offering standardised forms and checklists but none offered the level of insight required to improve the quality of care for the child or young person within the hospital setting. With this in mind we developed a stress sensory preferences interview and recommendations evaluation (SsPIRE) which was trialled within the NIHR/CRF between November 2015 and May 2016.

The authors developed SsPIRE as a result of their experiences of working within this field, noting key information to understand and support individuals who display behaviours of concern and have sensory difficulties. Therefore, their knowledge, understanding and perceptions of an assessment profiling tool such as the SsPIRE was required to gather data on how practical this profile is to implement into a ward setting. The parents feelings about their child being in hospital undergoing treatment can also influence their child's reactions and behaviours when receiving treatment. Therefore it was important to evaluate parents' perceptions of the stress sensory preferences interview and recommendations evaluation (SsPIRE), not just those of the nursing team.

This article is part of a series funded by the NIHR Wellcome Trust using qualitative research to develop best practice on holding and providing treatment to children with behaviours that challenge for clinical procedures and inform policy development.

Methods

This study was conducted at the National Institute for Health Research/Wellcome Trust Clinical Research Facility (NIHR/CRF) at Birmingham Children's Hospital. In supporting clinical research the CRF aims to improve clinical care and the lives of children, young people and their families. The unit opened in 2008 and currently has 107 active studies, with four Hunters Syndrome and Sanfilippo Syndrome trials taking place.

Ethical approval was gained prior to the commencement of this work from Birmingham City University Ethics Committee and Birmingham Children's Hospital Research and Development Strategic Committee.

Participants in this study were parents of children receiving treatment on the unit between February and May 2016. Only parents whose children had Hunters or SanFilippo Syndrome were asked to take part. Four mothers and one father volunteered, which provided 100% uptake from the eligible participant group. In order to preserve the principles of a least intrusive approach, it was considered appropriate for the authors to not observe the interaction between the clinical research nurse and parents when SsPIRE (version 1) was being completed, despite the obvious advantages of such. Also the highly confidential nature of clinical trials taking place would not be jeopardised.

The authors (AP, AM and CG) produced a semi structured interview script to evaluate the ease of completion, the sensory topics and questions covered within the profile, and how this can impact upon the care provided once strategies most appropriate for that child or young person have been recognised. [Please see interview script](#)

Thematic analysis was utilised, which is a useful method for identifying, analysing and reporting patterns (themes) within data (Braun and Clarke, 2006). This approach can also generate unanticipated insights and due to the limited research on using sensory profiles in this area, this was viewed as an advantage. Finally, as noted by Braun and Clarke, this approach is also beneficial for analysis informing policy development, which is an ultimate aim of this research. Primary analysis of the semi structured interviews was completed by all the authors, where the data was examined and certain points highlighted, such as noting contradictions, consistencies and common themes. AP, CG, PA reviewed all transcripts again to develop a rich thematic description of the entire data set and identify predominant themes. The authors organised the data into meaningful groups of interesting patterns (coding) manually. Themes were identified through mind mapping. AM joined the group to review, define and name the themes.

Results/findings

Three overarching themes were generated; person-centred planning, confidence and interview amendments.

Person centred planning.

“Person Centred Planning discovers and acts on what is important to a person. It is a process for continual listening and learning, focussing on what are important to someone now and in the future, and acting on this in alliance with their family and their friends” (Thompson, Kilbane and Sanderson, 2008:27).

With this in mind parents commented that they thought the SsPIRE was able to separate out what is important to the child or young person from what is important for them, and to find a balance for when they are receiving treatment in hospital.

Parent 4 *“My son does not like to wear shoes and will not put on a hospital gown”.*

Parent 1 *“You are wanting to get a better idea of the patient...so do they prefer to be active vast majority of times yes...but in this kind of environment not so much”*

As a framework, parents thought that the SsPIRE encouraged nurses to be thinking about their child’s care. In particular they felt that this process would be valuable for new patients in the unit or if their child was receiving treatment elsewhere in the hospital.

Parent 1 *“This tool would be great for example if he’s having an op somewhere else in the hospital and he’s going off to somebody who doesn’t know him well...this would be a great tool as a handover kind of package for them to be able to see”*

Parent 3 *“My son goes to several wards, hopefully this form will mean that all the nurses work together and do the same thing”*

Parents also suggested that the SsPIRE would be valuable to identify nurse training needs and further equipment that could be purchased.

Parent 2 *“I mean things like visual prompts like signs and symbols used in schools...he uses those and then obviously here he hasn’t got those... so just like a basic signs and symbols package would probably work quite well for him because none of the children with my child’s illness have got communication or Makaton abilities, none of them.....so that would probably benefit the staff here”*

Parent 5 *"The role of sensory play is often left to parents on the ward ...this form suggests that the nurses have a bigger role in this"*

The parents commented that the SsPIRE asked questions that they had not been asked before. One of the goals of the SsPIRE was to synthesise and organise the questions and responses so that it describes not only what is important to and important for each child or young person but also describes the balance between them ensure 'a good day' when they receive treatment on the unit, but also to minimise incidences of behaviours which challenged or interrupted treatment. Therefore whilst surprising to know that parents had not been asked certain questions before, it was reassuring to realise that the questions were viewed as important by the parents.

Parent 2 *"I don't think we have ever been asked about touch, balance and body awareness"*

Parent 1 *"Yeah...a lot of them ... do they like messy hands and that sort of thing I have not been asked before"*

Parent 5 *"It was good for questions on balance like does my son seek motion and what that actually means for him"*

Our experience in using a similar approach to the SsPIRE with children and adults with a learning disability suggested that the use of the tools would encourage sharing of information. We were pleased to note that this was also identified by the parents and prompted them also to think about introducing strategies into the unit.

Parent 1 *"I would share it with staff at the hospice they have a play worker so it would be good if she could have a copy and input into it as well"*

Parent 2 *"I'd share it with school I'd put it in his as part of his care plan... (my child) had quite an in depth sensory input through his OT back at home. He's had the weighted blankets, vest, he's got quite a few different things...but I would never have thought about bringing those kind of activities into this situation where actually it probably would have helped him"*

Confidence

Parents perceived that by using the SsPIRE led them to feel more confident is the ability of the nurses to care for their child.

Parent 4 *“made me have more confidence my son’s needs will be looked after and addressed”*

In this article we have highlighted the difficulties these children and young people have interacting with others, expressing their needs, likes and dislikes. One parent felt that the SsPIRE form would improve her child’s confidence in communicating to others as this form would help others to understand his needs.

Interview Amendments

All interview data are open to interpretation, which is why it is important to undertake a trial and examine people’s interpretation and viewpoints to enable modifications to take place. The parents asked if there could be a variable section within the questions to recognise that their child may behave differently dependent upon their mood.

Parent 3 *“There were a few questions where he was between a yes and a no. One on for example does physical activity distress or relax them or have no known effect...depending on his mood it can relax or it can stress him ”*

Parent 2 *“The only thing that I would point out is that the communication section is a very tricky one because in the main they have no verbal communication....they can’t tell you what they want...it’s a power of dedication type of thing...so the communication section is a really difficult one and could probably do with being expanded a little bit ”*

Parent 4 *“Possibly need additional questions about clothing...my son does not like to wear shoes and he will not wear a hospital gown ”*

Discussion

Thematic analysis yielded three main themes: person-centred planning, confidence and interview amendments when utilising this profile for the future. Parents appeared to find this form useful and empowering, stating it was easy to use and the questions allowed for positive information gathering, which promotes greater person-centred care. Thus, the SsPIRE appears to have good face validity and produce useful information on sensory experiences. It should be noted that the sample size for this study is small. However the setting of the NIHR/CRF being a highly specialised department means that a large sample size would be difficult to achieve.

It has been documented that individuals with an intellectual disability and/or traits of autism may process and experience sensory information differently to their peers without disabilities (Mefford et al, 2012; Thomchek and Dunn, 2007). Such difficulties may be highlighted more so when there is an increase in stress levels due to visiting hospital and receiving medical procedures. The behaviours of concern healthcare practitioners can include aggression, self-harm, destructiveness and disruptiveness. Children with behaviours such as these are also at risk of being denied access to in-patient, acute and emergency care treatments as the intensity of their behaviours puts them, parents/carers and healthcare staff at perceived risk of injury. This can lead to poorer quality of life and on-going health problems as treatment(s) may have been omitted or the results are not as effective (Page and McDonnell, 2013). These behaviours of concern are often seen in children with conditions that affect 'communication' and the 'brain', for example learning disabilities and more rare conditions such as Hunters Syndrome and Sanfilippo Syndrome. Communication is the main way we interact and express our needs, likes and dislikes. If communication is a problem then it can be very frustrating for the child and may result in behaviours of concern. Being in hospital can create stressful and anxiety emotional states which children may have limited coping abilities for (Wright, 1995). The treatment required for children and young people with Hunters Syndrome and Sanfilippo syndrome can be highly invasive and this information has been developed to assist and clarify key behaviours of concern. The nurses working within the NIHR/CRF are highly specialised and tend to see these children at intervals during their treatment process. SsPIRE has led to a changing in thinking from parents and healthcare staffs perspective- sorting what is important to the child from what is important for them within the hospital setting, and then looking for a balance between these two perspectives. The NIHR/CRF nursing team recognise that all too often this is not the reality of how services and professionals operate. Therefore the SsPIRE could also be a tool to facilitate teaching of this skill and so that identifying the balance between what is important to the child from what is important for them within the hospital setting becomes a habit for nursing staff.

SsPIRE follows the general principles of care identified within The NICE Guidelines (2015) in that it takes into account the child's learning disability, their communication needs and sensory needs in order to improve their quality of life when receiving treatment in hospital, and identifies sensory strategies to calm the child down or to divert them when they are demonstrating signs of distress. NHS England is committed to making sure that people with a learning disability and/or autism who display behaviours of concern receive the right care in the right settings, with the right support. The

feedback from parents is that SsPIRE will assist in improving the care given towards their child in hospital and their own confidence in healthcare staff to manage their child's behaviours. Further research is needed to determine whether this greater emphasis on admission and the communication pathway may have apposite impact upon the treatment process. In addition, involving parents in this process at an early stage may lead to the development of more positive working relationships with healthcare staff. Given statistics about the numbers of children affected by autism, the association of stress and sensory issues, future research needs to look at introducing bespoke autism training for nurses and evaluating the impact upon the care given.

Other relevant research currently being undertaken within the NIHR/CRF

Through the NIHR/CRFs sixth formers research programme additional supporting research has been developed and undertaken; the 'Sense not Stress study', designed by sixth formers explores the theory that using sensory equipment can alleviate anxiety in children and young people attending for clinical appointments and undergoing clinical procedures, creating a more personalised and positive experience for children and young people attending the facility. The study findings have highlighted the need for further research with the potential linkage to the sensory assessment profile (SsPIRE) to provide a wider data capture set, to assist in profiling and provision of equipment placement, and pathways for children visiting the facility especially for those children having sensory challenges.

A study has also been conducted investigating the perceptions of nursing staff using the SsPIRE in the same unit as the present study. Staff reported that the use of form has reiterated and increased their understanding of sensory processes and noted how this form promotes a person-centred approach for their patients. Using feedback from both nursing staff and parents we are now in the process of disseminating SsPIRE for use with children who have been identified.

References

- Braun V & Clarke V (2006) Using thematic analysis in psychology. *Qualitative Research in Psychology* 3 (2): 77-101
- Chan, J., Arnold, S., Webber, L., Riches, V., Parmenter, T & Stancliffe, R (2012) Is it time to drop the term 'challenging behaviour'? *Learning Disability Practice*, 15 (5):36-38
- Department of Health (DH) (2007) *Palliative Care Services for Children and Young People in England* London, DH Publications
- McDonnell, A, A., (2010) *Managing aggressive behaviours in care settings; Understanding and applying low arousal approaches* UK, Wiley- Blackwell
- Mefford, H, C., Batshaw, M, L., and Hoffman, E, P., (2012) Genomics, Intellectual Disability, and Autism *New England Journal of Medicine*; 366:733-743
- Muenzer J1, Wraith JE, Beck M et al. (2006) A phase II/III clinical study of enzyme replacement therapy with idursulfase in mucopolysaccharidosis II (Hunter syndrome). *Genetics in Medicine* 8(8):465-73.
- National Institute for Health and Care Excellence (2015) Challenging behaviour and learning disabilities: prevention and interventions for people with learning disabilities whose behaviour challenges *London, NICE*
- Page, A & McDonnell, A, A (2013) Holding children and young people: defining skills for good practice *British Journal of Nursing*, 22(20):1153-1158
- Page, A; McDonnell, A, A; Gayson, C; Moss, F; Mohammed, N; Smith, C and Vanes, N (2015) Clinical holding with children who display behaviours that challenge *British Journal of Nursing* 24(21):1004-1009
- Thomchek, S, D., and Dunn, W., (2007) Sensory Processing in Children With and Without Autism. *American Journal of Occupational Therapy*.61(2):190-200
- Thompson J. Kilbane J. Sanderson H. (2008) *Person Centred Practice for Professionals*. Berkshire, Open University Press
- Wright, M, C (1995) Behavioural effects of hospitalisation in children *Journal of Paediatrics and child health*. 31(3) p165-167

Interview Script – Sensory assessment forms (Parents)

Q1. Were there any questions you didn't understand being on the form?

Q2. Can you think of any additional questions that should be asked?

Q3. Were there any questions you hadn't been asked before with regard to hospital visits?

Q4. Has the use of these questions impacted on your confidence in the care given when visiting this unit?

Q5. If you were to have a copy of this form, who would you think about sharing it with?

Q6. Has using this form prompted you to think more proactively about plans for sensory care than before?

Q7. If your child has been visiting the unit prior to November 2015, has using this form and the sensory room impacted your visits to the unit?

Glossary

Hunters syndrome (mucopolysaccharidoses MPS II) is a disease in which long chains of sugar molecules (mucopolysaccharides) are not broken down correctly and build up in the body. It's an inherited condition where mainly boys are affected. The condition is caused by a lack of the enzyme iduronate sulfatase and without this enzyme, mucopolysaccharides build up in various body tissues, causing damage. The early-onset, severe form of the disease begins shortly after age 2. A late-onset, mild form causes less severe symptoms to appear later in life. Over a ten year period between 1992 and 2002, 52 babies with Hunters Syndrome were born in the UK.

Juvenile form (early-onset, severe form) can lead to spasticity, aggressive behaviours, hyperactivity, declining mental functioning over time and severe intellectual disability. A late (mild) form can lead to mild to no mental deficiency (Muenzer, Wraith & Beck, 2006).

Sanfilippo syndrome (mucopolysaccharidoses MPS III) is an inherited, autosomal recessive, metabolism disorder that where the body is unable to effectively break down long chains of sugar molecules called glycosaminoglycans. Sanfilippo syndrome occurs when the substances (enzymes) needed to break down the heparan sulfate sugar chain are missing or are defective. There are four main types of Sanfilippo syndrome, which type a person is depends on which enzyme is affected. Approximately 1 in 70,000 births are affected by Sanfilippo syndrome. The symptoms often appear after the first year of life. This can be characterised by a decline in learning ability typically occurring between ages 2 and 6. Delayed development is followed by deteriorating mental status and associated behavioural problems. These children can suffer significant neurological symptoms which include severe intellectual disability. These symptoms unfortunately mean that these children have severe challenging behaviours that parents, careers and health care providers have to deal with in order to look after the children (Muenzer, Wraith & Beck, 2006).

Guide to Understanding Mucopolysaccharidosis III (MPS III) Sanfilippo Disease



0845 389 9901
mps@mpssociety.org.uk
www.mpssociety.org.uk



Oliver (MPS IIIC)

Contents

What is Sanfilippo Disease?
What Causes Sanfilippo Disease
Are There Different Forms of Sanfilippo Disease?
How Common is Sanfilippo Disease?
How is Sanfilippo Disease Inherited?
Can You Test for Sanfilippo in Pregnancy?
Genetic Counselling
Clinical Presentation of Sanfilippo Disease
Progression of Sanfilippo Disease
General Management of Sanfilippo Disease
Living with a Child with Sanfilippo Disease
Treatment of Sanfilippo Disease
About the MPS Society

What is Sanfilippo Disease?

Sanfilippo Disease is a Mucopolysaccharide storage disorder, also known as Mucopolysaccharidosis Type III. The disease takes its name from Dr. Sanfilippo who was one of the first doctors from the United States to describe the condition in 1963.

Whilst there is no cure for individuals affected by MPS III, this factsheet explores the presentation and clinical management of the disease.

This factsheet is produced by the Society for Mucopolysaccharide Diseases (MPS Society) drawing on the experiences of parents and doctors with reference to medical literature.

What Causes Sanfilippo Disease?

Mucopolysaccharides are long chains of sugar molecules used in the building of bones, cartilage, tendons and many other tissues in the body. “Muco” refers to the thick jelly-like consistency of the molecules, “poly” means many and “Saccharide” is a general term for the sugar part of the molecule.

An alternative word for Mucopolysaccharides is glycosaminoglycans (or GAGS) but the term Mucopolysaccharides will be used for continuity throughout this Factsheet.

In the course of normal life there is a continuous recycling process of building new Mucopolysaccharides and breaking down old ones. This process requires a series of biochemical tools called enzymes.

Individuals with MPS III are missing an enzyme which is essential in cutting up the used Mucopolysaccharides. Incompletely broken down Mucopolysaccharides remain stored inside parts of the cells called lysosomes. The lysosomes become swollen and disrupt cell functioning causing progressive damage. Babies may show little sign of the disease but symptoms start to appear as more and more cells become damaged by the accumulation of Mucopolysaccharides.

Are There Different Forms of Sanfilippo Disease?

To date, four different enzyme deficiencies have been found to cause MPS III and thus the condition is described as MPS III Type A, B, C or D. Type A is the most common form found in most populations.

- MPS III A is missing the enzyme heparan N sulphatase
- MPS III B is missing the enzyme alpha-N-acetylglucosaminidase
- MPS III C is missing acetyl-CoA:alpha-glucosaminide acetyltransferase
- MPS III D is missing N-acetylglucosamine-6-sulphatase

It is important to note that there are no significant, clinical, physical differences between the different subtypes of MPS III disease, although there have been cases of late onset MPS III Type B where the individuals have remained relatively unaffected into adult life. The latest understanding is that some people seem to produce some enzyme activity which helps to slow down the progression of the disease whilst those with more severe symptoms appear to have no enzyme activity at all.

How Common is Sanfilippo Disease?

The MPS Society, which co-ordinates the Registry for Mucopolysaccharide and Related Diseases, has shown that MPS III is a rare condition affecting one in 85,000 live births. Over a ten year period between 1988 and 1998, 97 babies were born with MPS III in the United Kingdom.

How is Sanfilippo Disease Inherited?

MPS III is an autosomal recessive disease whereby both parents must carry the same defective gene and each pass this same defective gene to their child. Where both parents are carriers of the MPS III gene there is a 25% (1:4) chance of having an affected child in each pregnancy. There is a 50% (1:2) chance of a child receiving only one copy of the defective gene and therefore being a carrier. A carrier will not be affected but can pass the defective gene to his/her offspring. The remaining 25% (1:4) will be neither affected nor a carrier. Using information from an affected individual's DNA, it may be possible to determine whether brothers and sisters are carriers of, or if they are affected by MPS III.

Can you Test for Sanfilippo Disease in Pregnancy?

For each pregnancy the chances of a baby inheriting MPS III are totally independent of whether a previous child was affected by MPS III. Pre-natal tests can be arranged early on during a pregnancy for those families who already have a child with MPS III. Both amniocentesis and chorionic villus sampling can be used to diagnose MPS III in utero.

Genetic Counselling

All parents of children with a lysosomal storage disease should consider asking for genetic counselling before having other children. The counsellor should be able to advise on the risk to close relatives and to suggest whether the wider family should be informed.

Clinical Presentation of Sanfilippo Disease

Physical Appearance

Children with MPS III grow to a fairly normal height and changes in appearance may be less than in other MPS diseases. The hair is thick and coarser than usual and their bodies may be hairier than normal. The eyebrows are often dark and bushy and may meet in the middle. Noses tend to be upturned and flat on the bridge.

Physical Problems

Of all the MPS diseases, MPS III produces the mildest physical abnormalities. It is important, however, that simple and treatable conditions, such as ear infections, are not overlooked because behavioural problems make examination difficult. Parents may need to search until they find a doctor with the patience and interest in treating a child with a long-term condition. Do not hesitate to consult your doctor if you think your child may be in pain.

Intellectual Ability

Whilst the majority of children affected by MPS III will lose their intellectual ability progressively through childhood, a small number may retain intellectual skills into adulthood.

Neurological Problems

During the later stages of MPS III some children may experience seizures. Seizures are as a result in disruption in the electrical activity in the brain, which may also be referred to as epilepsy or fits.

Eyes

Corneal clouding does not occur in the eyes as it does in other MPS diseases.

Night blindness

Many families have reported that children with Sanfilippo Disease do not want to walk in the dark or are afraid when waking up at night. Determining the reason for this problem is difficult.

Putting a night light in a hall or bedroom may prove beneficial.

Seizures/Epilepsy

At a later stage of the disease a number of children with MPS III will start to have frequent, minor seizures when they momentarily alter their level of consciousness (absences). This could be a stare for a few seconds, lack of response or a slight twitch.

On days when this occurs the child may seem more out of touch or harder to feed. Some may have more generalised seizures (grand mal) involving either loss of consciousness or physical jerking.

During the seizure you should place your child on his or her side to prevent the inhalation of vomit. The child should be left in that position until the seizure is over. You should check that the airway is clear but do not put anything in the child's mouth.

Both forms of seizure can be treated with medication if indicated. Sometimes this may involve a number of drugs and the child may be drowsy until they become used to the medication.

Ears

Some degree of deafness is common in individuals suffering from all types of MPS III. It may be Conductive Deafness, Nerve Deafness or both (Mixed Deafness) and can be made worse by frequent ear infections.

It is important that individuals with MPS III have their hearing checked regularly and for problems to be treated early to improve or maintain the ability to communicate and learn.

Conductive Deafness is concerned with impaired transmission of sound waves through the ear canal, the ear drum and the middle ear. Correct functioning of the middle ear depends on the pressure behind the ear drum being the same as that in the outer ear canal and the atmosphere.

This pressure is equalised by the eustachian tube which runs from the middle ear to the back of the nose. If the tube is blocked the pressure behind the eardrum will drop, the drum will be drawn in and the transmission of sound waves will be impaired.



Amy (MPS IIIB)

If this negative pressure persists, fluid from the lining of the middle ear will build up and in time become thick like glue. Hence the condition being referred to as “glue ear”.

Conductive Deafness (Glue ear)

Under general anaesthetic a small incision behind the eardrum can be made (myringotomy) and the fluid sucked out. A small ventilation tube called a “grommet” may then be inserted to keep the hole open and allow air to enter from the outer ear canal until the eustachian tube starts to work properly again. Grommets will eventually fall out. If the conductive deafness recurs T-tubes may be used. T-tubes are a type of grommet which stay in place longer. Due to the anaesthetic risks for individuals with MPS III, the surgeon may decide to use T-tubes on the first occasion.

Sensorineural Deafness (Nerve Deafness)

In most cases the cause of Nerve Deafness is damage to the tiny hair cells in the inner ear. It may accompany Conductive Deafness, in which case it is referred to as “Mixed Deafness”.

Nerve Deafness is managed by the fitting of hearing aids. Children with MPS III may keep pulling out their hearing aids at first but it is important to persevere at wearing them so that communication can be maintained.

Respiratory Infections

Medication may affect individuals with MPS differently so it is essential to consult your doctor rather than using over the counter medication. Decongestants usually contain stimulants that can raise blood pressure and narrow blood vessels, both of which are undesirable for individuals with MPS III. Cough medicines that have a sedating effect may cause more problems with sleep apnoea by depressing muscle tone and respiration. Individuals with MPS III may end up with secondary bacterial infections which should be treated with antibiotics.

Heart

It is generally accepted that the heart is not usually affected by the disease.

Nose & Throat

The problems described in this section are common to children with MPS diseases but occur less often in individuals with MPS III. The severity of the problem depends greatly on the individual child.

Frequent coughs, colds and throat infections are common problems. The tonsils and adenoids often become enlarged and can partly block the airway. For this reason they may be removed (See breathing difficulties).

The neck is usually short which contributes to the problems in breathing. The windpipe (trachea) becomes narrowed by storage material and is often more floppy, or softer than usual, due to abnormal cartilage rings in the trachea. Nodules or excess induration of tissue can further block the airway.

Typically, the bridge of the nose is flattened and the passage behind the nose is smaller than usual due to poor growth of the bones in the mid-face and thickening of the mucosal lining.

The combination of abnormal bones and storage of Mucopolysaccharide in the soft tissue can cause the nose to become easily blocked. Some children with MPS III can have chronic discharge of clear mucus from the nose (rhinorrhea) and sinus infections.

Mouth & Teeth

The lips are thick, the gum ridges are broad and the tongue becomes enlarged. Teeth are widely spaced and poorly formed with fragile enamel. It is important that the teeth are well cared for as tooth decay could be a cause of pain.

Dental Hygiene

Teeth must be well cared for to avoid the need for extractions. If the water in your area has not been treated with fluoride then your child should have fluoride tablets or drops daily. Cleaning around the mouth with a small sponge or a stick coated in mouthwash will help keep the mouth fresh and avoid bad breath.

It may be safer for any treatment to be carried out in hospital. If teeth need to be removed under anaesthetic this should be carried out in hospital under the care of an experienced anaesthetist and never in the dental surgery.

Dribbling is a common problem and a plastic backed bib under the clothes may prevent soreness.

Breathing Difficulties

Children with MPS III may breathe very noisily, even when there is no infection. At night they may be restless and snore. Sometimes the child may stop breathing for short periods while asleep (sleep apnoea).

This noisy breathing, which stops and starts, can be very frightening for parents to hear and they may fear that their child is dying. In fact many children may breathe like this for years. Many children with MPS III have frequent colds, blocked noses and chest infections. This may be managed using C-PAP.

Night Time C-PAP

(Continuous Positive Airway Pressure)

A Night Time C-PAP may be recommended where a sleep study has shown that an individual is experiencing sleep apnoeas. This is where the individual suffers from low oxygen levels at night and stops breathing for short periods, leading to day time drowsiness and headaches.

C-PAP involves placing a mask or canula on the face each night and having air pumped into the airway to prevent it from collapsing. This may seem to be an extreme measure but it can greatly improve the quality of sleep and help prevent or reduce the risk of heart failure caused by low oxygen levels at night.

In severe cases of sleep apnoea with heart failure, a tracheostomy (a hole in the airway made in front of the neck) may be needed. Those who have received an early tracheostomy claim to feel much better after improving their night time breathing.

Liver, Spleen & Abdomen

The liver and spleen may be slightly enlarged due to the storage of Mucopolysaccharides (hepatosplenomegaly). The enlarged liver does not actually cause problems or lead to liver failure but its volume can interfere with eating and breathing.

Skin

Individuals with MPS III tend to have thickened and tough skin which lacks elasticity. Excess hair on the face and back occurs in some individuals.

Bowel Problems

Many individuals with MPS III suffer from loose stools and diarrhoea. Occasionally it is caused by severe constipation and leakage of loose stools from behind the solid mass of faeces. However, parents often describe it as “coming straight through”. It is thought there may be a defect in the autonomic nervous system which controls those bodily functions usually beyond voluntary control. Examination by a paediatrician or physician supplemented by an X-Ray if needed will establish the cause.

This problem may disappear as the child gets older but it can be worsened by antibiotics prescribed for other problems. If there is diarrhoea (and it is not secondary to constipation) simple medication, such as loperamide (Immodium) can be very useful. A diet low in roughage may also be helpful.

Constipation may become a problem as a child gets older and becomes less active, resulting in the muscles weakening. If an increase in roughage in the diet does not help or is not possible, the doctor may prescribe laxatives or a disposable enema.

Bones & Joints

Bones

Individuals with MPS III tend to have minimal problems with bone formation and growth.

Joints

Joint stiffness is common in all MPS diseases. All the joints become stiff and their movement may become limited. Later in the child's life this can cause pain which may be relieved by warmth and ordinary pain killers. The limited movement in the shoulders and arms may make dressing difficult.

Legs and Feet

Many individuals with MPS III stand and walk with their knees and hips flexed. Combined with the tight Achilles Tendon this may cause them to walk with their toes curved under, rather like the hands.

Hands

The fingers of children with MPS III occasionally become bent over (clawed) and the arms may not be able to extend fully. Later on there may be some limitation of movement in the large joints such as shoulders, elbows, hips or knees.

Hips

Individuals with MPS III may suffer from dislocated hips but treatment may not be advisable or necessary.

Hip Disease

Many children with MPS III have periods of limping or apparent pain in their hips. If an X-Ray is taken it is often thought that they have a condition called Perthes' Disease. The changes seen are in fact very common with MPS III and are probably part of the bone disease that occurs in all children with the disease.



Harrison (MPS IIIA)

Progression of Sanfilippo Disease

The disease will affect children differently and its progress will be much faster in some cases more than others. Change will usually be very gradual and therefore easier to adjust to. There could also be periods where there is a significant amount of change all at once, followed by a period of time where the rate of these changes slows down considerably. This is referred to as a plateau stage. The disease tends to progress through the following stages:

The First Stage

The first stage takes place during a child's pre-school years. This may be a very frustrating time for the parents. They begin to worry as their child starts to lag behind their friends' children in development and they may feel that they are being blamed for the child's overactive and difficult behaviour. The diagnosis is often made very late due to the fact that some children do not look different in any way and their symptoms (such as diarrhoea, frequent respiratory and ear infections as well as boisterous behaviour) are among the most common seen in all children. The doctor has to be perceptive enough to recognise that something serious is wrong and to ask for urine and blood tests to help reach a diagnosis. It is not unusual for families to have had more than one affected child before the diagnosis is established.

The Second Stage

The second phase of the disease is characterised by extremely active, restless and often very difficult behaviour. Some children sleep very little at night. Many will be into everything. Many like to chew hands, clothes or anything they can get hold of. Sadly, language and understanding will gradually be lost and parents may find it hard not being able to have a conversation with their child. Many will find other ways of communicating, perhaps by using sign language such as the Makaton System. Some children never become toilet trained and those who do will eventually lose this ability.

The Third Stage

In the third phase of the disease children with MPS III begin to slow down. They become unsteady on their feet and tend to fall frequently as they walk or run. Eventually they will lose the ability to walk. Life may be more peaceful in some ways but parents will need help with the tiring physical aspects of caring for an immobile child or teenager. Parents will also need to be advised on how best to cope with these changes so they are equipped and able to provide their child with the best care possible.

General Management of Sanfilippo Disease

Anaesthetic

Giving an anaesthetic to an individual with MPS III requires skill and should always be undertaken by an experienced anaesthetist. Where a child is concerned this should be a paediatric anaesthetist.

The airway can be very small and may require a very small endotracheal tube. This is a tube that is inserted into the trachea through the mouth or nose in order to maintain an open air passage to deliver oxygen

or to permit the suctioning of mucus. Placing this tube may prove difficult and require the use of a flexible bronchoscope. In addition, the neck may be somewhat lax. Repositioning the neck during anaesthesia or intubation could cause injury to the spinal cord. For some individuals, it is difficult to remove the breathing tube after surgery is completed.

There is a more detailed explanation of this complex subject in the specialist anaesthetic booklet published by the MPS Society.

Physiotherapy & Hydrotherapy

Both physiotherapy and hydrotherapy can be useful to help individuals with MPS III achieve specific and realistic goals in daily life. It can also help to drain mucus from the chest.

Individuals should be as active as possible in order to improve their general health. A physiotherapist may be able to suggest ways of achieving this. The best forms of physiotherapy exercises are those that are introduced through play in the younger individuals.

In adults it is important to remember that passive stretching maybe painful and should only be used with caution.

Medication

Children with MPS may be affected differently by drugs so it is essential to consult your doctor rather than purchase over the counter medication.

Drugs may be tried for controlling mucus production but some may make the mucus thicker and harder to dislodge. They may also make the child more irritable. The use of sedatives can increase the problem of sleep apnoea by depressing respiration.

It is now recognised that frequent use of antibiotics may make them less effective when really needed. Repeated use can also cause thrush, a fungal infection which commonly affects the mouth or vagina and produces a white curd-like deposit. It causes irritation and discomfort and will need to be treated. Your doctor may, therefore, wish to limit the number of times antibiotics are prescribed for coughs and colds.

Diet

Most children with MPS III have hearty appetites but if your child is faddy it can be difficult to achieve a balanced diet. Ask your doctor or dietician if supplementary vitamins should be given.

There is no scientific evidence that a particular diet has any beneficial effects. Symptoms such as diarrhoea tend to come and go naturally. Some parents, however, find that a change in their child's diet can ease problems such as excessive mucus, diarrhoea and hyperactivity. Cutting down on milk, dairy products and sugar as well as avoiding foods with too many additives and colourings have all been said to help individual children.



Oliver (MPS III) with his brother Samuel

It would be advisable to consult your doctor or a dietician if you plan any major changes to ensure that the proposed diet does not leave out any essential nutrients.

If your child's problems are eased by a new diet you could try reintroducing foods one at a time to establish if there are any foods in particular that increase your child's symptoms.

Feeding

Children's early feeding very rarely causes problems, however, some do not progress to eating food that requires chewing. Others learn to chew but increasingly find solid food difficult to manage. Children with MPS III can find lumps in food particularly difficult, especially if mixed with food of a smooth texture. The child may also become quite faddy and reject a number of foods for no particular reason.

Children may become very difficult to feed. If possible parents should encourage their child to take an active part in feeding themselves. This, as well as seeing and smelling their food, encourages them to prepare for the process of swallowing. Some parents find a musical box or the television calms their child during a meal. As children lose the rhythm of swallowing they may start to splutter and cough while eating. In addition, dribbling or drooling may be more obvious. It is better to serve food of a mashed consistency. Meat will be coped with more easily if it is made soft through slow cooking rather than just chopped into small pieces.

Thickeners added to liquids can often make swallowing drinks easier. Moving the hand gently backwards under the chin and slowly down the throat can help the tongue to move and encourage swallowing.

Choking is frightening so you can provide reassurance by rubbing your child's back and holding hands.

Cold Hands & Feet

It cannot be said whether icy cold hands and feet are a feature of the disease or purely due to lack of activity. If this becomes a problem it may be beneficial for the child to wear warm socks and gloves.

In the later stages of the disease the body's natural temperature control mechanism may become damaged and the child may sweat at night and have cold hands and feet by day. Some children have episodes when their body temperature drops (hypothermia). If this happens you should warm your child up and ask your doctor for advice on the best ways of managing the problem.

Gastrostomy

For some individuals swallowing will become increasingly difficult and unsafe.

In these circumstances a gastrostomy is recommended. This is a tube placed in the stomach during a short operation. Most parents prefer a gastrostomy as this does not interfere with the airway or irritate the nose.

Although a gastrostomy requires a short general anaesthetic, there are usually no complications as long as the operation is done before the child becomes too frail.

Gastrostomies can leak and occasionally the skin around the tube insertion becomes inflamed. You will be given advice from your surgeon on how to deal with this should the need arise.

Chewing

As they become more out of touch with their environment, many children with MPS III will amuse themselves by rocking or by chewing their fingers, clothes or whatever they can lay their hands on. As this behaviour cannot be stopped, it is best dealt with by providing an acceptable and interesting range of things to chew such as rubber toys or teething rings. If the problem becomes so severe that the child is causing harm to their fingers and hands, it is possible to splint the elbows for periods of the day to prevent the hands from reaching the mouth.

Hyperactive Behaviour

Most Sanfilippo children go through a hyperactive stage when they are difficult to control and unaware of danger. It is not so easy to modify this by drugs and it is better, if possible, to adapt the home (as described earlier). A garden where the child can run about safely is a great asset. It might be possible to get a grant for fencing if this is necessary to make it secure. It is most helpful if the child can get out to a playgroup or to school where a variety of activities can be provided. Children with MPS III do not concentrate for long periods of time and cannot be expected to sit still. Ideally there should be space for the child to run about and use up energy as well as keeping fit for as long as possible. Many children are calmed by the movement of a car and will travel well.

Sleeping Difficulties

Many Sanfilippo children are very restless at night, not sleeping for more than a couple of hours at a time. The reason for this is not known.

It is sometimes possible to improve the situation with medication, but it may take a period of trial and error to establish which drug will work best for your child. Drugs often lose their effect after a while so some parents choose to either ration their use to a few nights a week or accept that after a few weeks the drug will have to be discontinued for a period. Some parents find they can achieve a longer period of unbroken sleep by putting the child to bed later and with a regular routine. It is vitally important that parents get enough sleep if they are to cope in the day, so do not hesitate to ask your doctor for help.

Living with a Child with Sanfilippo Disease

Home Adaptations

During the hyperactive stages of MPS III parents find it very helpful if they can set aside a room or part of a room within hearing distance. The room, which can be fitted with a half door latched on the outside, can be made safe for the child to play without constant supervision so that the parent can get on with day to day tasks in relative peace and quiet. Furniture which is fragile or that has sharp edges should be removed and replaced by large cushions or a mattress on the floor. Windows may need to be fitted with strengthened glass and the floor should be easy to clean. The child's favourite playthings and durable toys should be provided and a television can be placed on a high shelf or suspended from the ceiling. These could be operated by the parents using a remote control.

Children in the later stages of the disease will become progressively less mobile and increasingly dependent on their parents and carers to meet their everyday needs in areas of incontinence, personal hygiene and nutrition.

It is important to give thought early on to the ways in which the families and carers will manage when weight bearing and walking or climbing the stairs is no longer possible. An ensuite bathroom for the child's bedroom is ideal with plenty of space for a buggy and a carer to manoeuvre around in. When weight bearing is no longer possible a hoist is beneficial with tracking from bed to bath directly in line for ease of use. Adaptations can often take a long time so it is prudent to plan ahead as far as possible. The MPS Society has considerable experience of the options available to families caring for a child with MPS III.

Education

Whilst some children with MPS III may benefit from having a mainstream education in their primary school years and enjoy the social interaction with peers, a majority will equally benefit from a Special Educational Needs Placement with small class sizes and a range of communication systems in place. Children with MPS III may have a Statement of Special Educational Needs or need an Individual Education Plan (IEP) with regular reviews.

Many will need the help of a classroom assistant during the school day as well as during breaks and lunchtimes to ensure they are able to access all the educational opportunities open to them. This is also to maintain their health and safety throughout the day.

This provision will enable the class teacher to set specific tasks for the child. The classroom assistant will be on hand to help keep the child on task and maintain concentration. Many individuals with MPS III have a very short concentration span requiring them to be constantly kept on short tasks.

Having a Break

Caring for a child affected by MPS III is hard work and parents or carers need a break to rest and enjoy activities which may not be possible when in their caring role. Many families use children's hospices; social services respite care or have someone close, such as a friend or family member, on hand to help out at busy times. Further details of hospices throughout the UK offering respite care to families are available from the MPS Society.

Palliative Care

Palliative care is provided to both the family and the child with a life limiting disease in situations where curative treatment is not an option. This support encompasses aspects such as respite care, symptom management and bereavement support and may extend over a considerable period of time. In addition, considerable personal care may be required including feeding and personal hygiene. This can take up a large amount of time. The stress involved can put a family under immense strain. An assessment of medical needs and a care plan should lead to an approved package of support being provided to both the child and the family. Once in place this should enable both to experience a better quality of life.



Sophie (MPS IIIA)



Lewis (MPS IIIA)

Puberty

Children with MPS III will go through the normal changes associated with puberty, sometimes at an early stage. There are a number of ways of managing periods in older girls and you should ask your doctor for advice.

Life Expectancy

Whilst many children will lose their lives to MPS III in their teenage years, a significant number will survive into adulthood. Survival into the 30s, 40s and even 50s is known.

Treatment of MPS III

At present there is no cure for MPS III. Various experimental methods have been used to try to replace the missing enzyme but none have proven to have any significant long term benefits. Bone Marrow Transplant has been tried on patients with MPS III but with disappointing results and is now never recommended for this condition.

About the MPS Society

The Society for Mucopolysaccharide Diseases (MPS Society) was founded in 1982 representing over 1200 children and adults suffering from MPS and related diseases including Fabry, their families, carers and professionals throughout the UK. It is a registered charity entirely supported by voluntary donations and fundraising and is managed by the members themselves.



Nathan (MPS IIIA)

Society for Mucopolysaccharide Diseases

MPS House, Repton Place, White Lion Road, Amersham, Buckinghamshire, HP7 9LP, UK
Registered Charity No. 1143472. Registered as a Charity in Scotland No. SCO41012
Registered as a Company limited by guarantee in England & Wales No. 7726882



0845 389 9901

mps@mpssociety.org.ukwww.mpssociety.org.uk

Guide to Understanding Mucopolysaccharidosis II (MPS II) Hunter Disease

Contents

What is Hunter Disease?
 What Causes Hunter Disease?
 Does Hunter Disease Affect Individuals Differently?
 How Common is Hunter Disease?
 How is Hunter Disease Inherited?
 Can you Test for Hunter Disease in Pregnancy?
 Genetic Counselling
 Life Expectancy
 Clinical Presentation of Hunter Disease
 General Management of Hunter Disease
 Living with a Severely Affected Child or Adult
 with Hunter Disease
 Living with a Less Severely Affected Child or Adult
 with Hunter Disease
 Specific Treatments for Hunter Disease
 About the MPS Society

What is Hunter Disease?

Hunter Disease is a Mucopolysaccharide storage disorder also known as Mucopolysaccharidosis Type II (MPS II).

Hunter Disease takes its name from Charles Hunter, a Professor of Medicine in Manitoba, Canada, who first described two brothers with the disorder in 1917.

In the past just two types of Hunter Disease have been described, mild and severe. It is now clear, however, based on current understanding of the enzyme and its gene, that Hunter Disease comprises a wide spectrum of severity.

All individuals with Hunter Disease have a deficiency of the enzyme 'iduronate sulphatase' which results in the accumulation of Mucopolysaccharides. The accumulation of Mucopolysaccharides is responsible for many problems that affect individuals with Hunter Disease.

Whilst there is no cure for individuals affected by Hunter Disease, this fact sheet explores the disease's presentation and clinical management.

This fact sheet is produced by the Society for Mucopolysaccharide Diseases and draws on the experiences of parents and doctors with reference to medical literature.

What Causes Hunter Disease?

Mucopolysaccharides are long chains of sugar molecules used in the building of bones, cartilage, skin, tendons and many other tissues in the body.



Matthew

"Muco" refers to the thick jelly like consistency of the molecules, "poly" means many and "saccharide" is a general term for a sugar molecule. An alternative word for Mucopolysaccharides is Glycosaminoglycans or GAGs but the term Mucopolysaccharide will be used for continuity throughout this fact sheet.

In the course of normal life there is a continuous recycling process of building new Mucopolysaccharides and breaking down old ones.

The breakdown and recycling process requires a series of special biochemical tools called enzymes.

Individuals with Hunter Disease are missing, or are deficient in, an enzyme called 'iduronate sulphatase' which is essential in the breaking down of the Mucopolysaccharides called 'dermatan' and 'heparan sulphate'.

The incompletely broken down dermatan and heparan sulphate remain stored inside the cells of the body and begin to build up causing progressive damage.

Babies may show little sign of the disease but as more and more cells become damaged by accumulation of Mucopolysaccharides, symptoms start to appear.



Archie

Does Hunter Disease Affect Individuals Differently?

Until recently Hunter Disease had been described as either mild or severe. However, based on current understanding of the enzyme and its gene, Hunter Disease comprises a wide spectrum of severity. Some individuals with Hunter Disease will have progressive developmental delay and severe progressive physical problems. Others will have normal intelligence and progressive physical problems, some being more severely affected than others.

It is important to remember that Hunter Disease is extremely varied in its effects. A whole range of possible symptoms are outlined in this factsheet, however, the affected individual may not experience all of them. Hunter Disease may even vary in its effects between siblings and generations.

How Common is Hunter Disease?

Except in very rare cases, only males will be affected by Hunter Disease. The MPS Society, which co-ordinates the European Registry for Mucopolysaccharide and related diseases, has shown that this is a rare condition affecting 1 in 100,000 male births.

Over a 10 year period, between 1992 and 2002, 52 babies with Hunter Disease were born in the United Kingdom.

How is Hunter Disease Inherited?

Hunter Disease has a different form of inheritance from all other MPS Diseases as it is an X-linked recessive disease (also called sex-linked) like haemophilia.

Females may be carriers, but except in very rare cases only males will be affected. If a woman is a carrier for Hunter Disease there is a 50% (1:2) risk that any male born to her will have the disorder. Further more, there is a 50% (1:2) risk that any female born to her will be a carrier for the disorder. This means that there is a 25% (1:4) chance of having an affected child with each pregnancy.

The sisters and maternal aunts of an individual with Hunter Disease may be carriers of the disorder and would

also have a 50% risk of passing the abnormal gene to any male born to them. In many families it is possible to detect female carriers by direct analysis of genetic material. The doctor may wish to take a sample of blood from your affected child so that the exact genetic abnormality can be detected. In most families it is possible to identify the exact genetic fault on the X-chromosome responsible for Hunter Disease (mutation analysis). This can help with pre-natal diagnosis and carrier testing. However, the mother is not always found to be a carrier for Hunter Disease in all cases. In this situation the disorder may have occurred in the male for the first time, a new mutation.

There is a more detailed explanation of this complex subject in the booklet on inheritance available from the MPS Society.

Can you Test for Hunter Disease in Pregnancy?

There are three situations when pre-natal testing is possible in early pregnancy to detect Hunter Disease:

- If you are already a mother to a child with Hunter Disease.
- If you know you are a carrier.
- If you are a female relative on the mother's side to a male with Hunter Disease and have not been carrier tested.

It is important to contact your doctor when planning a pregnancy, or as soon as you suspect that you may be pregnant if you wish tests to be arranged.

Both amniocentesis and chorionic villus sampling can be used to diagnose Hunter Disease in utero.

Genetic Counselling

All parents of children with a lysosomal storage disease should consider asking for genetic counselling before having other children. The counsellor should be able to provide non-directive advice on the risk to close relatives and the reproductive choices available as well as advice on whether the wider family should be informed.

Life Expectancy

There is a wide range of life expectancy depending on whether there is involvement of the Central Nervous System (CNS). Those individuals who do not have CNS involvement may have a reasonably normal life span if their physical problems, such as chest and heart disease, are not severe.

Survival into the fifth and sixth decades of life, however, is rare. Sadly, those who do have CNS involvement are likely to die before reaching their mid to late teens. Some children may die much earlier.

Clinical Presentation of Hunter Disease Growth

Babies with Hunter Disease may be larger than average and may grow faster than normal during the first two years of life. In individuals with severe Hunter Disease their final height is likely to be between four feet (120cm) and four feet seven inches (140cm). Individuals with less severe Hunter Disease usually grow to a slightly less than normal height of 150-165cms (5 foot to 5 foot 6 inches).

Physical Appearance

Individuals with Hunter Disease tend to have a close resemblance to each other with many similar features. Their faces are often chubby with rosy cheeks and their heads are rather large with a prominent forehead. The neck is short and the nose is broad with a flattened bridge. The lips are often thickened and the tongue enlarged. The hair tends to be thick, the eyebrows bushy and there may be more hair than usual on the body.

Individuals with Hunter Disease have prominent bellies and a characteristic way of walking and holding their arms due to joint contractures at their hips, shoulders, elbows and knees.

Those in receipt of Enzyme Replacement Therapy (ERT) have noticed a marked improvement in certain characteristics including softening of the hair and facial features as well as a noticeable improvement in their height. Individuals have also noticed that their tummies are far less prominent due to the reduction in the size of the internal organs.

Intellectual Ability

Individuals with severe Hunter Disease usually experience progressive storage of Mucopolysaccharides in the brain that is primarily responsible for the slowing of intellectual development by 2 to 4 years of age.

This is often followed by a gradual loss of skills until death; however the pattern is very varied. Some individuals will only learn to say a few words while others learn to talk well and to read a little. They can enjoy nursery rhymes and simple puzzles.

Emphasis should be on helping infants and children with Hunter Disease to learn as much as they can before the disorder progresses.

Even when the child starts to lose skills they have learned there may be some surprising abilities left. Children will continue to understand and find enjoyment in life even if they lose the ability to speak.

Individuals with less severe Hunter Disease may have normal intelligence. They usually have the same physical features as those seen in severe Hunter Disease, but with a reduced rate of progression.

Brain

The brain and the spinal cord are protected from jolting by the cerebrospinal fluid that circulates around them. In some individuals with Hunter Disease the circulation of the fluid may become blocked over time.

The blockage (communicating hydrocephalus) causes increased pressure in the head which can press on the brain. If this remains untreated it will result in headaches, visual impairment and delayed development.

Other aspects of Hunter Disease that can affect brain function include inadequate oxygen levels and sleep deprivation due to sleep apnoea.

Hydrocephalus

Hydrocephalus (also known as 'water on the brain') can be confirmed using a CT or MRI scan. A lumbar puncture (known as a spinal tap) with pressure measurement is another way to assess if hydrocephalus exists. If hydrocephalus is confirmed it can be treated by insertion of a thin tube (shunt) which drains fluid from the brain. The shunt has a pressure sensitive valve which allows spinal fluid to be drained when the pressure around the brain becomes too high. A lack of swelling around the optic disc does not rule out hydrocephalus in an individual suffering from Hunter Disease.

Epilepsy

A number of individuals who are severely affected by Hunter Disease will develop epilepsy. This may take different forms, e.g. absence episodes (where the individual may appear to be staring into space with or without jerking or twitching movements of the eye muscles), or more generalised tonic-clonic seizures (a type of generalized seizure that affects the entire brain). Tonic-clonic seizures are more commonly associated with epilepsy. Fortunately most individuals will respond favourably to anticonvulsant medication.

Eyes

Clouding of the cornea, which is a feature of some of the other MPS diseases, is not normally found in individuals with Hunter Disease. Occasionally there may be problems with vision caused by changes to the retina or glaucoma (due to increased fluid pressure inside the eye) which should be checked during an eye examination. Storage in the retina can result in loss of peripheral vision and night blindness.

Night Blindness

Many families have reported that children with Hunter Disease do not want to walk in the dark or are afraid when waking up at night. Sometimes the simple addition of a night light in a hall or bedroom may prove beneficial. It is often difficult to determine what is responsible for this problem.

Ears

Some degree of deafness is common in individuals suffering from Hunter Disease. It may be Conductive Deafness, Nerve Deafness or both (Mixed Deafness) and may be made worse by frequent ear infections. It is important that individuals with Hunter Disease have their hearing checked regularly and for problems to be treated early to improve or maintain the ability to communicate.

Correct functioning of the middle ear depends on the pressure behind the ear drum being the same as that in the outer ear canal and the atmosphere. This pressure is equalised by the eustachian tube which runs from the middle ear to the back of the nose. If the tube is blocked, the pressure behind the eardrum will drop and the drum will be drawn in. If this negative pressure persists, fluid from the lining of the middle ear will build up and in time become thick like glue. Hence the condition being known as "glue ear".

Conductive Deafness (Glue Ear)

Under general anaesthetic a small incision behind the eardrum can be made (myringotomy) and the fluid sucked out. A small ventilation tube called a "grommet" may then be inserted to keep the hole open and allow air to enter from the outer ear canal until the eustachian tube starts to work properly again. Grommets will eventually fall out. If the Conductive Deafness recurs the surgeon may decide to use T-tubes (a type of grommet which stays in place much longer). In view of the anaesthetic risks for individuals with Hunter Disease, the surgeon may decide to use T-tubes on the first occasion.

Sensorineural Deafness (Nerve Deafness)

In most cases the cause of Nerve Deafness is damage to the tiny hair cells in the inner ear. It may accompany Conductive Deafness, in which case it is referred to as Mixed Deafness. Nerve Deafness is managed by the fitting of hearing aids in most individuals with Hunter Disease. More severely affected children may keep pulling out their hearing aids at first but it is important to persevere at wearing them so that communication can be maintained. Other children with Hunter Disease have found radio aids and the loop system helpful at school and at home.

Nose & Throat

Typically, the bridge of the nose is flattened and the passage behind the nose is smaller than usual due to poor growth of the bones in the mid-face and thickening of the mucosal lining.

The combination of abnormal bones and storage of Mucopolysaccharide in the soft tissues in the nose and throat can cause the nose to become easily blocked.

One of the common features of children with Hunter Disease is the chronic discharge of clear mucus from the nose (rhinorrhea), and sinus infections.

Frequent coughs, colds and throat infections are common problems for many individuals with Hunter Disease. Individuals will have narrowing of the large airways and increased secretions which can lead to 'asthma-like' episodes. Many individuals with Hunter Disease are helped by treatment of asthma medication during viral illness.

Many affected individuals breathe very noisily, even when there is no infection. At night they may be restless and snore. Admission to hospital overnight for a sleep study may be advised. Monitors are placed on the skin and connected to a computer.

These monitors measure brain waves, the levels of oxygen in the blood and the breathing effort that is required during sleep. From this study doctors can assess how much blockage to breathing is present, how much trouble your child is having moving air into the lungs during sleep and how much of an effect this has on their body.

Removal of tonsils and adenoids may help in some cases to lessen the obstruction and make breathing easier, but adenoid tissue may grow back.

The windpipe (trachea) becomes narrowed by storage material and is often more floppy, or softer than usual due to abnormal cartilage rings in the trachea. Nodules or excess induration of tissue can further block the airway making swallowing difficult.

Night-time C-PAP

A Night-time CPAP may be recommended where a sleep study has shown that an individual is experiencing sleep apnoeas with low oxygen levels at night. Sleep apnoea is where the individual stops breathing for short periods during sleep. This leads to daytime drowsiness and headaches.

C-PAP involves placing a mask or canula on the face each night and having air pumped into the airway to prevent it from collapsing. This may seem to be an extreme measure but it can greatly improve the quality of sleep as well as help prevent or reduce the risk of heart failure caused by low oxygen levels at night.

In severe cases of sleep apnoea with heart failure, a tracheostomy (a hole in the airway made in front of the neck) may be needed. Most individuals with Hunter Disease will try to avoid a tracheostomy because it is invasive and seemingly destructive of normal function. In fact those who have received an early tracheostomy claim to feel much better after improving their night time breathing.

Mouth & Teeth

Individuals with Hunter Disease usually have thicker lips and an enlarged tongue. Gum ridges are broad. The teeth are widely spaced and poorly formed with fragile enamel.

Dental Hygiene

It is important that the teeth are well cared for to avoid the need for extractions. If the water in your area has not been treated with fluoride, a child with Hunter Disease should have fluoride tablets or drops daily. Cleaning around the mouth with a small sponge or a stick soaked in mouthwash will help keep the mouth fresh and avoid bad breath. Dribbling is a common problem and a plastic-backed bib under the clothes may prevent soreness.

Regular checks at the dentist are important as tooth decay could be a source of pain. If your child is severely affected it may be safer for treatment to be carried out at a hospital.

It is important that you inform the dentist if your child has a heart problem. You will probably be advised that s/he should be given antibiotics before and after any dental treatment. This is because certain bacteria in the mouth may get into the blood stream and cause an infection on the heart valves. If teeth need to be removed under anaesthetic, this should be carried out in a hospital under the care of an experienced anaesthetist and never in the dental surgery. It may be possible for the hospital to carry out other treatment or investigations under the same anaesthetic.

Chest & Respiratory Infections

The shape of the chest is abnormal and the junction between the ribs and the breastbone (sternum) is not as flexible as it should be. The chest is therefore rigid and unable to move freely to allow the lungs to take in a large volume of air.

The muscles at the base of the chest (diaphragm) may be pushed upwards by an enlarged liver and spleen, further reducing the space for the lungs. When the lungs are not fully cleared, there is an increased risk of respiratory infections that may lead to pneumonia.

Respiratory Infections

Medication may affect individuals with MPS differently, so it is essential to consult your doctor rather than using 'over the counter' medication.

Medication for controlling mucus production may not help. Medication such as antihistamines may dry out the mucus making it thicker and harder to dislodge. Decongestants usually contain stimulants that can raise blood pressure and narrow blood vessels, both undesirable for individuals with Hunter Disease. Cough medicines that have a sedating effect may cause more problems with sleep apnoea by depressing muscle tone and respiration.

Individuals with Hunter Disease commonly end up with secondary bacterial infections which should be treated with antibiotics.

Heart

Heart Disease is fairly common in individuals with Hunter Disease but may not develop or cause problems until much later in life. The heart may be affected in different ways. The valves, which open and shut as the blood is pumped from one chamber of the heart to another, may be weakened by storage of Mucopolysaccharides. The valves may fail to close tightly enough allowing small amounts of blood to leak back again. The muscles of the heart may also be damaged by storage of Mucopolysaccharides (cardiomyopathy) and the heart may be put under strain by upper airway obstruction, repeated chest infections or by having to pump blood through stiffened lungs (cor pulmonale).

Heart Problems

Some individuals with Hunter Disease may develop problems with the aortic or mitral valve: they may have slowly progressive valvular Heart Disease for years without any apparent clinical effects. If the condition worsens an operation may be needed to replace the damaged valves. As heart problems occur so frequently in individuals with Hunter Disease, a test known as an 'echocardiogram' should be carried out annually (or as often as your doctor thinks necessary) to show whether any problems are developing. The test is painless and similar to the ultra sound screening of babies in the womb. It can identify problems with heart muscle, heart function and heart valves.

Liver, Spleen & Abdomen

In most individuals with Hunter Disease both the liver and spleen become enlarged by storage of Mucopolysaccharides (Hepatosplenomegaly).

The enlarged liver does not usually cause liver problems or lead to liver failure, but it can interfere with eating and breathing.

In individuals with Hunter Disease, the abdomen bulges out due to posture, weakness of the muscles and the enlarged liver and spleen.

Frequently, part of the abdominal contents will push out behind a weak spot in the wall of the abdomen. This is called a hernia.

Hernias

The hernia can come from behind the navel (umbilical hernia) or from the groin (inguinal hernia). Inguinal hernias should be repaired by an operation.

Umbilical hernias are not usually treated unless they are large and cause entrapment of the intestine, or are very large and are causing other problems.

It is very common to have a recurrence of both inguinal and umbilical hernias after a repair has been made.

Bowel Problems

Many individuals with Hunter Disease suffer frequently or periodically from loose stools and diarrhoea. The cause of this is not fully understood.

Occasionally the problem is caused by severe constipation and leakage of loose stools from behind the solid mass of faeces. More often however, parents describe it as "coming straight through".

It is thought that there may be a defect in the autonomic nervous system (the system that controls those bodily functions) usually beyond voluntary control. An examination by the paediatrician or physician, supplemented by an X-ray if necessary, will establish which is the cause.

The problem may disappear as the child with Hunter Disease gets older, but it can be made worse by antibiotics prescribed for other problems.

If there is diarrhoea (and it is not secondary to constipation) simple medication, for example loperamide (Imodium) can be very useful.

A diet low in roughage may also be helpful. Constipation may increasingly become a problem as a child with Hunter Disease gets older, less active and the muscles progressively weaken.

If an increase in roughage in the diet does not help or is not possible, the doctor may prescribe laxatives or a disposable enema.



Matthew

Skin

Individuals with Hunter Disease tend to have thickened and tough skin which lacks elasticity. This can cause irritation and soreness, particularly in areas where the skin folds i.e at the back of the neck.

Some individuals with Hunter Disease have a characteristic pebble-like texture to their skin. These small skin-coloured bumps around the shoulder blades, upper arms and thighs are caused by storage of Mucopolysaccharides (nodular skin lesions).

Excess hair on the face and back occurs in some individuals with Hunter Disease. Sweating and cold hands and feet are also common problems due to poor temperature control, as the centre of the brain which regulates the temperature becomes damaged.

Bones & Joints

Individuals with Hunter Disease tend to have significant problems with bone formation and growth. This leads to both bone and neurological problems if nerves are compressed by bone.

Joint stiffness is a common feature of Hunter Disease and the maximum range of movement of all joints may become limited. Joint stiffness may cause pain, which may be relieved by warmth and ordinary painkillers.

The limited movement in the shoulders and arms may make dressing difficult. Anti-inflammatory drugs, such as ibuprofen, can help with joint pain, but they should be taken with or after food and monitored closely under medical supervision to make sure that irritation and ulcers of the stomach do not occur.

Spine

The bones of the spine (vertebrae) normally line up from the neck to the buttocks. Individuals with Hunter Disease can have poorly formed vertebrae that may not stably interact with each other. One or two of the vertebrae in the middle of the back are sometimes smaller than the rest and set back in line.

This backward slippage of the vertebrae can cause an angular curve (kyphosis or gibbus) to develop, but is usually mild and generally does not need treatment.

Older children and adults with Hunter Disease occasionally develop compression of the spinal cord due to thickening of the ligaments around the bones of the neck. The doctor will want to monitor this carefully and arrange surgical treatment if necessary.

Hands

The shape of the hand is very characteristic and the MPS Society's logo depicts the hands of a boy with Hunter Disease. The hands are broad with stubby fingers which gradually curve over or become clawed.

Individuals with Hunter Disease often experience pain and loss of feeling in the fingertips caused by Carpal Tunnel Syndrome. The wrist (or carpus) consists of eight small bones known as the carpals which are joined by fibrous bands of protein called ligaments. Nerves have to pass through the wrist in the space between the carpal bones and the ligaments. Thickening of the ligaments causes pressure on the nerves. This can cause irreversible nerve damage. The nerve damage will cause the muscle at the base of the thumb to waste away.

Carpal Tunnel Syndrome

Although a child or adult with Hunter Disease may not complain of pain they may already have Carpal Tunnel Syndrome. Doctors may advise for this to be monitored with a test called a nerve conduction study which will show whether there is carpal tunnel syndrome present. This test would also be carried out if there is any weakness or numbness in the hand or decreased muscle mass at the base of the thumb. This disorder can be treated by a minor operation.

Legs & Feet

Many individuals with Hunter Disease stand and walk with their knees and hips flexed. This, combined with the tight achilles tendon, may cause them to walk on their toes. Sometimes these individuals have knocked knees but this is very unlikely to need treatment. The feet are broad and may be stiff with the toes curved under, rather like the hands.

General Management of Hunter Disease

Anaesthetic

Giving an anaesthetic to an individual with Hunter Disease requires skill and should always be undertaken by an experienced anaesthetist.

Where a child is concerned this should be a paediatric anaesthetist.

The airway can be very small and may require a very small endotracheal tube. Placing the tube may prove difficult and require the use of a flexible bronchoscope.

In addition, the neck may be somewhat lax and repositioning the neck during anaesthesia or intubation could cause injury to the spinal cord.



Jack

For some individuals, it is difficult to remove the breathing tube after surgery is completed. There is a more detailed explanation of this complex subject in the specialist anaesthetic booklet published by the MPS Society.

Physiotherapy & Hydrotherapy

Physiotherapy and Hydrotherapy can be useful to help individuals with Hunter Disease achieve specific and realistic goals in daily life or to drain mucus from the chest. At other times it is common sense for the individuals to be as active as possible to improve their general health and the physiotherapist may be able to suggest ways of achieving this. For younger individuals the best forms of physiotherapy are exercises that are introduced through play. In adults it is important to remember that passive stretching may be painful and should only be used with caution.

Living with a Severely Affected Child or Adult with Hunter Disease

Children with Hunter Disease may be overactive, strong, usually cheerful and affectionate but hard work to look after. They have limited powers of concentration and less understanding than you would expect for their age and physical development. They could, for example, lock the bathroom door but be unable to understand how to get out again, even when told. They enjoy rough and tumble play, making a lot of noise and throwing toys rather than playing with them. They may be unaware of danger, stubborn and unresponsive to discipline as they cannot understand what is required. Some may have outbursts of aggressive behaviour. Toilet training may be achieved briefly by some individuals but most will remain in nappies. Getting enough sleep may be difficult for parents who should not hesitate to ask their doctors for help.

Feeding

Most children with Hunter Disease enjoy their food, although some will eat only a very limited range of foods. They often drink a great deal. Many do not progress to using a knife and fork or an ordinary cup and eventually it may be necessary to feed your child as you would a baby. In the later stages, your child may find it harder to chew properly and food may have to be mashed or liquidised.

When a child or adult cannot chew and has difficulty swallowing, there is a risk of choking. Food, especially meat, should be cut up very small, but even this may not prevent the possibility of choking. For some individuals, swallowing will become increasingly difficult and in these circumstances a gastrostomy may be recommended.

Education

Whilst some children with severe Hunter Disease may benefit from having a mainstream education in their primary school years and enjoy the social interaction with peers, a majority will equally benefit from a Special Educational Needs placement with small class sizes and a range of communication systems in place.

Children with Hunter Disease may have a statement of Special Educational Needs or need an Individual Education Plan (IEP) with regular reviews. Many will need the help of a classroom assistant.

Home Adaptations

Children and adults with severe Hunter Disease will become progressively less mobile and increasingly dependent on their parents and carers to meet their everyday needs in areas of incontinence, personal hygiene and nutrition.

It is important to give thought early on to the ways in which the families and carers will manage when weight bearing and walking or climbing the stairs is no longer possible.

The MPS Society has considerable experience of the options available to families caring for an individual with severe Hunter Disease.

Gastrostomy

For some individuals swallowing, and therefore eating and drinking, will become increasingly difficult and unsafe. In these circumstances a gastrostomy would be recommended. This is a tube (called a nasogastric tube) that is placed in the stomach during a short operation. Most parents prefer a gastrostomy as this does not interfere with their airway or irritate the nose.

Although a gastrostomy requires a short general anaesthetic, as long as the operation is done before the child becomes too frail there are usually no complications. Gastrostomies can leak and occasionally the skin around the tube insertion may become inflamed. If this happens you will be given advice on how to deal with this from your surgeon.

Having a Break

Caring for an individual severely affected by Hunter Disease is hard work and parents or carers need a break to rest and enjoy activities which may not be possible when in their caring role.

Many families use children's hospices, social services respite care and/or have a friend or family member close by that is available on a regular basis to help at busy times.

The Quieter Stage

The change from the overactive, noisy period is likely to be gradual. Families will realise that their affected child no longer runs everywhere and is happier sitting than standing. Many will be easily pleased, perhaps by looking through the same little book of photographs, having stories read or watching the same video many times over. Children and adults with Hunter Disease may doze off quite often.

Weight will be lost gradually as muscles waste away and chest infections may be more frequent. Many affected individuals die peacefully after an infection or from the heart's gradual failure. Family and friends may find it helpful to prepare for the time of death.

If you feel you would like to do this please contact the MPS Society who have information you may find helpful.

Living with a Less Severely Affected Child or Adult with Hunter Disease

Less severely affected children suffering from Hunter Disease may be completely normal in behaviour and they are often affectionate, sunny natured children.

They can be short tempered at times from frustration when their physical limitations make life difficult.

Independence

As a child with Hunter Disease grows up they should be encouraged to be as independent as possible since many adults with Hunter Disease can live full and enjoyable lives. The teenage years may be difficult: if ordinary adolescents worry about a pimple on the chin, think how much more teenagers with Hunter Disease must worry about their appearance and the restrictions imposed by their condition. They may be helped by meeting or being in touch with other teenagers and adults with Hunter Disease. Ask the MPS Society to put you in touch through their Befriending Scheme. Those who are less severely affected by Hunter Disease will go through the normal stages of puberty but possibly a year or so after their peers.

Home Adaptations

Appropriately adapted living accommodation will greatly enhance the ability of an individual with Hunter Disease to develop independent living skills. A few less severely affected adults with Hunter Disease have found satisfying work as social workers, civil servants, teachers of the deaf and one is known to be a marine architect.

There is every reason to encourage a young person with Hunter Disease to lead as full and as independent a life as possible.

The Future

A few adults with Hunter Disease have married and had children. The sons would not have Hunter Disease unless the mother happens to be a carrier.

Daughters of a father with Hunter Disease will all be carriers of the disease.

Specific Treatments for Hunter Disease

Bone Marrow Transplant (BMT)

For some years Bone Marrow Transplants (BMT) have been used to treat children with Mucopolysaccharide and related diseases but this procedure is currently not recommended for children with Hunter Disease. Bone Marrow Transplant in Hunter Disease has not been shown to have any effect on preventing the damage to the brain that occurs in individuals severely affected.

Enzyme Replacement Therapy (ERT)

Enzyme Replacement Therapy (ERT) for Hunter Disease has undergone a number of clinical trials and is now available for affected individuals. This therapy is based on the principle that the recombinant form of the enzyme that is missing or malfunctioning is given via repeated intravenous infusion in order to reduce the symptoms and clinical manifestations associated with the disease.

This involves the recombinant enzyme being given by repeated intravenous infusion every week.

ERT will help some of the physical problems associated with Hunter Disease, but unfortunately the 'blood-brain barrier' prevents the medication from directly helping the brain. As a result, the enzyme infusions do not lead to improvement in those who have Central Nervous System involvement.

ERT has demonstrated benefits in energy levels, reduction in the size of the liver and spleen, walking ability and lung function tests over a period of 12-18 months.

In the longer term it is hoped that ERT will encourage the stabilisation of the physical condition and will continue to show an improvement within the quality of life in most individuals.



Colin

Future Treatments

There is a great deal of research being carried out that may lead to other treatments in the future.

As previously indicated, ERT poses a challenge in successfully introducing enzyme into the brain cells. To try and get round this in an attempt to develop a treatment for patients with severe MPS II, research workers are looking at the possibility of giving the enzyme directly into the fluid around the brain; so called intra-thecal (IT) therapy.

It has been proposed that, if sufficient enzyme is infused by this route, a small quantity may cross the blood-brain barrier and enter the brain. Initial studies have commenced in the USA but there are currently no results available.

Gene therapy, which is the concept of replacing the faulty gene with a copy of a normal gene, may be a realistic possibility in years to come. Experiments on animals with MPS II suggest that this may be a successful route of treatment in the future however; it is possible that not all those affected by Hunter Disease will be able to benefit from these advances.

Your paediatrician or physician may be able to give you up to date information on treatment options.

You can also contact the MPS Society.

About the MPS Society

The Society for Mucopolysaccharide Diseases (MPS Society) was founded in 1982 and represents over 1200 children and adults suffering from MPS and related diseases including Fabry, their families, carers and professionals throughout the UK.



Society for Mucopolysaccharide Diseases

MPS House, Repton Place, White Lion Road, Amersham, Buckinghamshire, HP7 9LP, UK
Registered Charity No. 1143472. Registered as a Charity in Scotland No. SCO41012
Registered as a Company limited by guarantee in England & Wales No. 7726882