Guide to Understanding
Mucopolysaccharidosis II (MPS II)
Hunter Disease

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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.

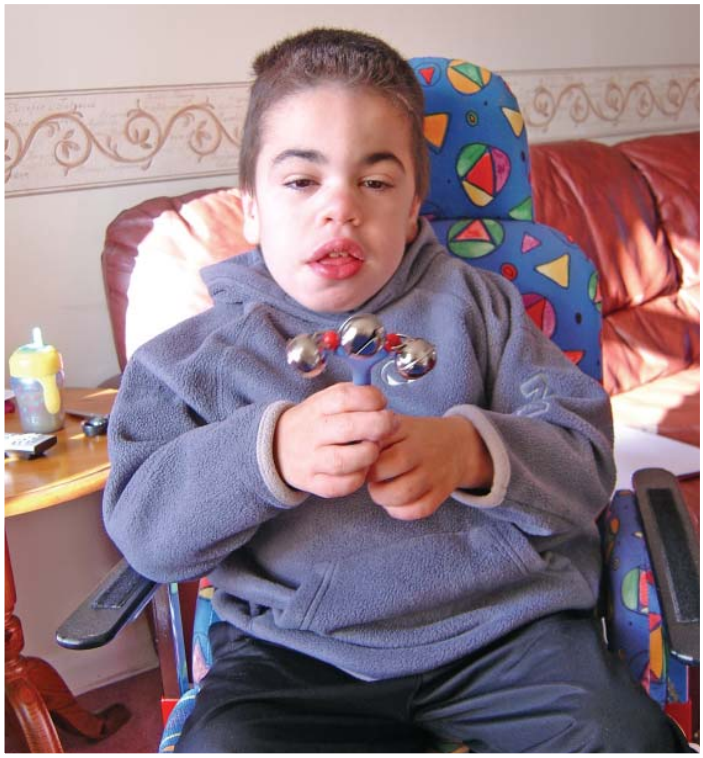
All individuals with Hunter Disease have a deficiency of the enzyme 'iduronate sulphatase' which results in the accumulation of Mucopolysaccharides. The accumulation of Mucopolysaccharides is responsible for many problems that affect individuals with Hunter Disease.

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.



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"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.

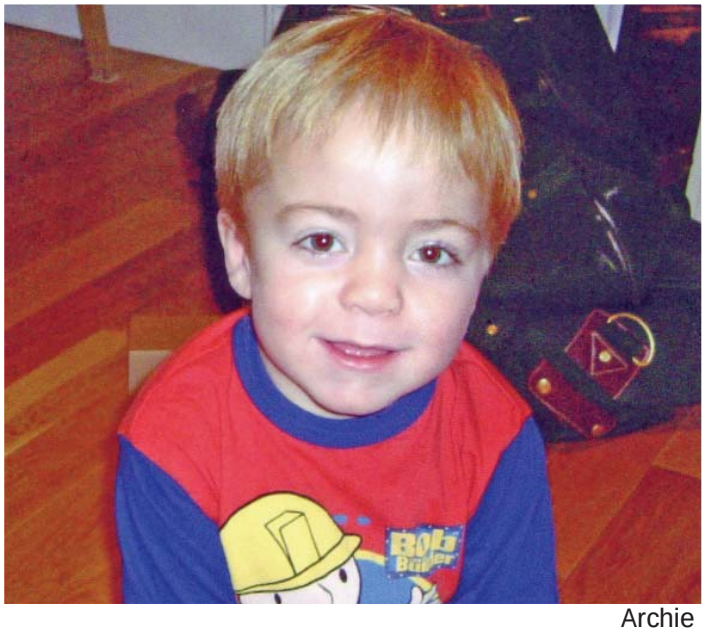
In the course of normal life there is a continuous recycling process of building new Mucopolysaccharides and breaking down old ones.

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 bare 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".

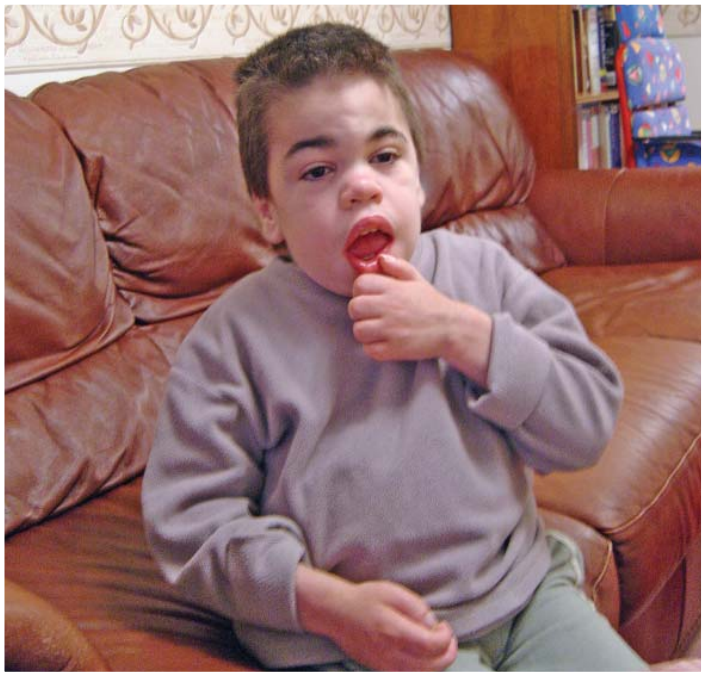
It is thought that there may be a defect in the autonomic nervous system (the system that controls those bodily functions) usually beyond voluntary control. An examination by the paediatrician or physician, supplemented by an X-ray if necessary, will establish which is the cause.

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.



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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

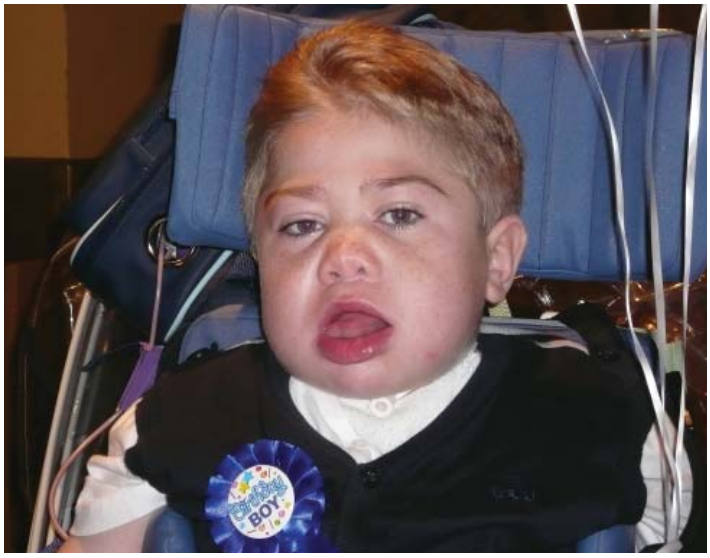
Anaesthetic

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.



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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 Adapations

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.

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