Canadian MPS Society
for Mucopolysaccharide & Related Diseases

June, 2010

Dear Friends:

We are pleased to provide you with this resource called "Mucopolysaccharidosis II: A resource for individuals and families living with MPS II." This comprehensive binder is designed to provide you with a source of information on the manifestations, concerns, and management of MPS II disease, along with space for you to record and store your own experiences.

This resource is not intended to constitute medical advice or care, legal advice, or financial advice. You should evaluate the information in this resource in the context of your individual situation, and consult appropriate professionals to assess what makes sense in your particular case. The Canadian Society for Mucopolysaccharide & Related Diseases (Canadian MPS Society) and Shire Human Genetic Therapies Inc. (Shire HGT) make no representation or warranty with respect to, and disclaim any legal liability or responsibility for, the accuracy or completeness of the contents of this resource, and they specifically disclaim any implied warranties or fitness for a particular use.

Begin by reading the table of contents. Recognizing the heterogeneity of MPS II and the differences medical management may make on its progression, you may find sections that are not relevant to you or your child at this time, but may be in the future. The chapters are separated by colour so they can be easily updated when there is new information available.

The binder includes special extras, such as a business card holder to make it easy to keep track of all the physicians and other medical professionals you see. There are handouts about MPS II that you may copy and give to physicians, teachers, and support workers. You may also download and print additional copies of these handouts from our website (www.mpssociety.ca). The MPS II journal is designed to be a special place to write down information you receive during medical appointments, information on treatments, and observations about you or your child.

We encourage you to ask for copies of medical reports and evaluations and include these items in the journal section of the binder. By having all information in one location, you can easily answer the questions asked by the various specialists you see.

We welcome your comments and suggestions about this resource and hope you find it to be a beneficial and functional tool.

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Canadian MPS Society
for Mucopolysaccharide & Related Diseases

Contents

Mucopolysaccharidosis II (MPS II)

Management options

Anesthesia considerations

Living with MPS II

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Educational strategies

MPS II journal

Information handouts

Glossary

Author acknowledgments

Updates and new information

Developed in collaboration with Shire HGT

Shire
Human Genetic Therapies

Mucopolysaccharidosis II (MPS II)

What is MPS II disease?

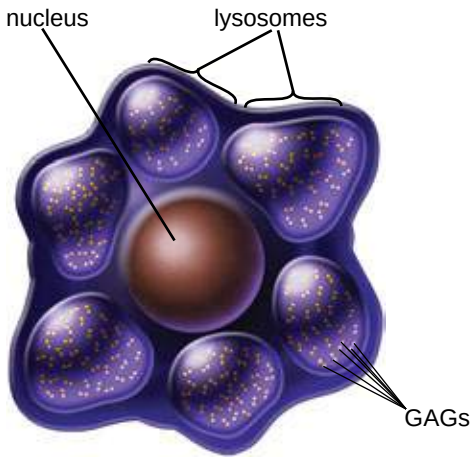
Mucopolysaccharidosis II (MPS II; pronounced **mew**-ko-**pol**-ee-**sak**-ah-ri-**doh**-sis **two**) is a rare genetic disorder that affects many body systems and may lead to damage of different body organs (e.g., the heart). MPS II is also known as Hunter syndrome. It is caused by a defect in the gene that instructs the body to make an enzyme called iduronate-2-sulfatase (pronounced eye-**dur**-o-nate **two sul**-fa-tace), also called I2S. Because of this defect, cells either produce the enzyme in low amounts or cannot produce it at all. The enzyme is needed to break down substances called glycosaminoglycans (GAGs; pronounced **gly**-cose-a-**mee**-no-**gly**-cans). If GAGs are not broken down, they build up in the cell, eventually leading to damage to cells, tissues, and organs.

GAGs, previously called mucopolysaccharides, are long chains of sugar molecules joined together, and are located mostly on the outside surface of cells. The body uses them in the building of bones, cartilage, skin, tendons, and many other tissues in the body. GAGs form part of the structure of the body and also give the body some of the special features that make it work. For example, the slippery, gooey joint fluid that lubricates your joints contains GAGs. The rubbery cartilage in your joints is another example. All tissues have some of this substance as a normal part of their structure. However, individuals with MPS II have too great an amount of GAGs in their body.

To understand how GAGs accumulate and cause MPS II, it is important to understand that in the course of normal life, there is a continuous process of building new substances like GAGs and breaking down old ones. This ongoing process is required to keep your body healthy. The breakdown and rebuilding of GAGs requires a number of different enzymes, which are chemicals that are made by the body.

To break down GAGs, a series of enzymes work in sequence one after another. Each enzyme in the process has its special purpose in the body and does one very specific action, just like a screwdriver works on screws and a hammer works on nails. As mentioned earlier, people with MPS II are missing one specific enzyme called iduronate-2-sulfatase (I2S), which is needed for the breakdown of certain GAGs called dermatan sulfate and heparan sulfate. If these GAGs are not completely broken down, they remain stored inside cells throughout the body and begin to build up. This buildup of GAGs interferes with how the cell normally works and causes damage that gets worse with time. Some individuals with MPS II may initially show few signs of the disorder, but as more and more GAG builds up, signs of too much GAG in the body's tissues start to appear. GAGs are produced in the body, not obtained from sugar in foods, so it is not possible to reduce the buildup by eating a special diet.

Mucopolysaccharidosis II (MPS II) is a rare genetic disorder that affects many body systems and that may lead to damage of body organs.



Cell with GAG buildup

Courtesy of Genzyme Corporation

How is MPS II related to other MPS syndromes?

The enzymes involved in breaking down GAGs are contained inside special parts of the cell, called lysosomes. The GAGs that accumulate as a result of the missing enzyme, therefore, are all stored within the lysosomes in the cells. It is for this reason that MPS II is part of a larger family of diseases called the "lysosomal storage disorders" (or "LSDs" for short).

There are more than 40 different conditions that are known to be caused by lysosomal storage disorders. These disorders have some features in common: they are genetic; most are caused by a deficiency in a particular enzyme normally present in lysosomes; and they cause by-products of chemical reactions to build up in the body's cells, leading to symptoms in a variety of body systems. However, because the exact enzyme is different in each case, individual LSDs can cause completely different types of symptoms and affect different organ systems. Individual LSDs can also differ in the age that they begin to cause symptoms and in how they affect life expectancy.

Within the larger family of LSDs, however, there are certain groups of conditions that have many clinical features in common. The mucopolysaccharidoses (MPS) are an example of such a group. The MPS disorders include 7 main types: MPS I, MPS II, MPS III, MPS IV, MPS VI, MPS VII, and MPS IX. All MPS disorders result in the buildup of various GAGs in the lysosomes. Each type of MPS is caused by a deficiency of a different specific enzyme. Although each of the individual MPS disorders can cause a variety of different symptoms, the disorders collectively have many symptoms in common (which are explained later in this resource) – for example, short stature, joint stiffness, speech and hearing problems, hernias, runny nose, and heart problems.

How common is MPS II?

MPS II is a rare disease, but it is hard to say exactly *how* rare because there are so few studies. MPS II occurs in at least 1 of every 170,000 live male births. MPS II is extremely rare in females.

It is worth noting that while MPS II is rare, the larger family of lysosomal storage diseases collectively occur in about 1 in every 5,000 live births.

How do people inherit MPS II?

MPS II is a genetic disorder. To understand how MPS II is inherited, it is important to understand some basics of genetics.

Most cells in the human body have 46 chromosomes: 23 from the mother, and 23 from the father. Females have two X chromosomes (one from the mother and the other from the father). Males have an X chromosome (from the mother) and a Y chromosome (from the father). MPS II is an X-linked recessive disorder. This means that the gene needed to make I2S (the enzyme that is deficient in MPS II) is found on the X chromosome.

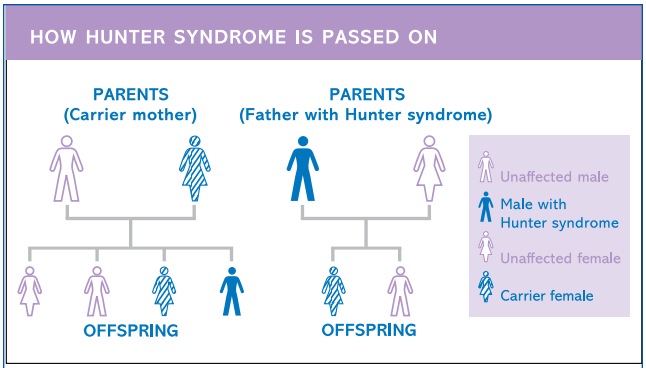
Because MPS II is an inherited disorder, family screening is extremely important.

Females who have one "MPS II" X chromosome and one unaffected X chromosome generally have enough enzyme to stay healthy because the unaffected X chromosome contains the gene needed to make enzyme. Although the body may only produce about 50% of the normal level of enzyme, the body is quite resilient, and even the 50% level of enzyme is more than enough to keep the person healthy. Basically, half the enzyme can work twice as hard. This is why MPS II is very rare in females, although some cases have been reported. Females with one "MPS II" X chromosome are called "carriers," since they may pass this affected chromosome on to their children.

Males who have one "MPS II" X chromosome lack the gene to make enzyme, because they do not have another X chromosome, just a Y

chromosome (which does not have the gene needed to make enzyme). Therefore, they do not make enough enzyme and are affected by MPS II. Affected males may inherit a damaged X chromosome from their mother or their X chromosome may become damaged spontaneously.

The figure below shows how the MPS II gene may be passed from one generation to the next.



Hunter syndrome (MPS II) shows X-linked inheritance. On average, a carrier mother will pass on the abnormal I2S gene to 50% of her sons and 50% of her daughters. A father with Hunter syndrome will pass on the abnormal I2S gene to all of his daughters and none of his sons.

Courtesy of Shire HGT

Because of the way genetic inheritance works, in a family with no previous MPS history it is impossible to predict in advance of a pregnancy whether parents are carrying the defective gene for MPS II and if they will have an affected child. However, it is possible to calculate the probability that this will occur if the parents are known to be carriers. As the figure above illustrates, female children born to a carrier mother and an unaffected father have a 1 out of 2 (50%) chance of becoming a carrier. Male children born to a carrier mother and an unaffected father have a 1 out of 2 (50%) chance of having MPS II. All female children born to a father affected by MPS II and an unaffected mother will become carriers, and none of the male children will be affected. It is rare for males with MPS II to father children.

Because MPS II is an inherited disorder, family screening is extremely important. When a diagnosis for an individual is made, it is important to find out whether other siblings may also be affected. A genetic counsellor can be a valuable resource to help you understand how MPS II is inherited in families and to help determine if others in the family will want to consider genetic testing for MPS II as well. Testing for MPS II is done by measuring the enzyme level in a blood or skin sample, and your doctor can arrange for this test. With early diagnosis, the disease may be better managed.

Prenatal testing allows pregnant women who have already had a child with MPS II to find out if the baby they are carrying is also affected by MPS II. There are 2 tests that can be used: chorionic villus sampling (taking a sample of the placenta) and amniocentesis (taking a sample of the amniotic fluid). Your doctor can advise you on your specific prenatal testing options. The decision to have prenatal testing is complex and personal. It is important to consult your doctor early in the pregnancy if you wish tests to be arranged. Talking with your genetic counsellor or doctor can help you explore these options and other strategies for having additional children while limiting the probability that they will have MPS II.

MPS II disease spectrum

Our understanding of the molecular basis and the signs and symptoms of MPS II has become clearer over time. It is becoming increasingly clear that this disease is more complex and more variable than was previously assumed.

It is now perhaps appropriate to view MPS II as a continuous spectrum of disease, with the most rapidly progressing (severely affected) individuals on one end and the more slowly progressing individuals on the other end, and a whole range of different severities in between. Some physicians choose to use the term “attenuated” for the more slowly progressing individuals. It is important to note that rapidly progressing or severe disease does not always include neurologic decline or delay.

Spectrum of disease severity in MPS II:

Rapidly progressing — *Slowly progressing*

Courtesy of the National MPS Society and Shire HGT

It is now perhaps appropriate to view MPS II as a continuous spectrum of disease, with the most rapidly progressing (severely affected) individuals on one end and the more slowly progressing (attenuated) individuals on the other end, and a whole range of different severities in between.

Is it possible to predict the severity of MPS II?

MPS II is a progressive disease with a broad spectrum of disease severity. There is currently no 100% reliable test that indicates how severe an individual's MPS II will eventually become. People with MPS II either completely lack the I2S enzyme needed to break down GAGs or only produce small amounts.

Many different kinds of mutations (defects in the make-up of genes) in the gene that produces I2S have been identified, all of which result in I2S enzyme deficiency. This enzyme deficiency results in MPS II disease. The severity of MPS II has not been found to be related to the amount of I2S or its activity.

Therefore, there is still today no perfectly reliable way to determine the exact effects that the disease will have on many individuals with MPS II. Even with the same small amount of enzyme activity, and even within the same family, there can be variations in severity of disease that cannot be explained by the enzyme level or DNA mutation. It is important to remember that MPS II is a spectrum with a variety of symptoms, and the disorder is extremely varied in its effects. A whole range of possible symptoms is outlined later in this learning guide, but any given person may not experience them all or to the degree pictured in those sections.

As MPS II is a condition that gets worse with time, all individuals will experience progression of symptoms, no matter where they are on the spectrum of disease severity.

Is it possible to predict lifespan?

Disease severity varies significantly for people with MPS II and it is not possible to predict the expected lifespan for a given individual. Lifespan depends on many factors, such as position on the spectrum of disease severity and the specific kinds of symptoms a person experiences. Those on the more slowly progressing end of the disease spectrum may have a reasonably normal lifespan.

The oldest known individual with MPS II survived to 87 years of age.

Individuals with more rapidly progressing disease are likely to have a shortened lifespan: they may live into their mid-teens, although some may die much earlier.

How is a diagnosis made?

Doctors may consider testing for MPS II when signs and symptoms of the condition are present and are not explained by other causes. All diagnostic tests should be overseen by a doctor with expertise in lysosomal storage disorders, as the tests are complicated and the results may be difficult to interpret.

A clear diagnosis requires enzyme tests conducted by experts. Early diagnosis is critical.

To diagnose MPS II, the doctor will typically first do a urine test to look for levels of glycosaminoglycans (GAGs) that are higher than normal. The results are compared to levels of GAGs that are known to be normal for various ages. Most, but not all, individuals with MPS disorders have GAG levels in their urine that are higher than those of individuals without MPS.

A urine test is only one of the first steps in diagnosing MPS II; a clear diagnosis requires a test to measure levels of enzyme activity in the blood or skin cells. In healthy individuals, the tests show white blood cells, serum, and skin cells that contain normal levels of enzyme activity. In individuals with MPS II, the enzyme activity levels are much lower or absent.

Early diagnosis of MPS II is critical. The earlier that MPS II is diagnosed, the sooner potential treatment options can be explored and supportive care may be started to help you or your loved one.

An update on newborn screening

Newborn screening is the testing of newborn babies to see whether they have specific disorders. The goal is to help with early diagnosis and management. Each province makes its own decisions about which health conditions should be included in their newborn screening programs.

The factors that are considered when deciding on newborn testing include:

- Is the disorder clearly defined?
- Does the disorder occur fairly often?
- Does early diagnosis help?
- Are tests available to diagnose the disorder accurately and cost-effectively?
- Can the tests be done quickly or is there a long waiting time for results?

Lysosomal storage disorders (such as MPS II) meet some of these criteria:

- Lysosomal storage disorders occur in about 1 in every 5,000 live births, which is more frequent than some conditions already included in newborn screening.
- Early diagnosis helps affected people get interventions sooner.
- There are a number of “markers” (substances that can be measured, such as GAGs in the urine) for lysosomal storage disorders that could potentially be used to develop screening tests. However, developing tests for lysosomal storage disorders is a complex process.

Currently, there is a growing movement promoting newborn screening for MPS disorders such as MPS II. It is now more widely recognized that to many families, information about the diagnosis alone is helpful with the opportunity for genetic counselling and education about additional management options.

Considering the potential benefits, the current aim is to develop a test that would allow for these options for children with lysosomal storage disorders. To develop a good screening strategy,

researchers have identified features common to all lysosomal storage disorders. This is important because there would not be enough time, money, and labour available to measure each enzyme that is deficient in each individual disorder, which is the usual method of making a diagnosis, so different methods for screening are being studied.

Some researchers suggest a tiered, or step-by-step, screening strategy. The first screening would involve measuring certain markers to identify a person who is at risk for the condition. For the person identified as "at risk," a second test would be performed to identify higher levels of stored materials. A positive result on both tests would need to be found before a family is referred for counselling, additional necessary testing, and management.

Research into newborn screening for LSDs is still in early stages. Important questions remain about the screening process and the testing methods. There will likely continue to be debate over the appropriateness of screening. There may also be concern about the effect on the parent-child relationship when a newborn is identified with a condition before symptoms appear. The test may also not be able to tell how severe the child's symptoms may become. This leaves many questions for families and healthcare professionals who want to choose the best management options. As a community, those whose lives have been touched by MPS II will likely continue to become more involved in the promotion of newborn screening.

Signs and symptoms of MPS II and their management

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Overview

MPS II has a wide range of symptoms, and people may experience different degrees of disease severity and disability. The table below provides an overview of the signs and symptoms that may occur in individuals who have MPS II. The content in later sections will provide more details about some of these signs and symptoms. Where possible, symptoms that are similar or linked or relate to a common organ system have been included together. Within the section devoted to each symptom or group of symptoms, there is also some information about disease management and relevant surgical procedures.

MPS II affects many areas of the body. Because its signs and symptoms are so variable, it affects each individual differently.

It is important to note that many individuals with MPS II may never experience some of the symptoms described in this section, and that those who do experience such symptoms will not necessarily do so to the degree featured in some of the pictures.

Table 1 provides a brief overview of MPS II signs and symptoms. Further information is provided in the sections to follow.

Table 1: MPS II signs and symptoms

General symptoms
- reduced endurance

Physical appearance
- abnormal facial features (such as broad, flat-bridged noses; thick lips; flat faces; eyes that stick out slightly; and an enlarged tongue that may stick out)
- large head (macrocephaly)
- short stature

Mouth and teeth
- enlarged tongue
- abnormal teeth (widely spaced and poorly formed, with fragile enamel)

Eyes, ears, nose, and throat
- vision problems, such as reduced field of vision or optic nerve damage (some people may have glaucoma, but this is not common in MPS II)
- hearing loss
- recurrent ear infections (otitis media)
- recurrent sinus infections (sinusitis)

Respiratory system (lungs and breathing)
- lung problems and reduced lung function
- sleep apnea
- recurrent lung infections

Heart and blood vessels
- heart valve problems
- damage to the heart muscle (cardiomyopathy)
- irregular heartbeat (cardiac arrhythmia)
- high blood pressure in the blood vessels supplying the lungs (pulmonary hypertension)

Gastrointestinal system (stomach and intestines)
- enlarged liver and spleen (hepatosplenomegaly)
- umbilical and inguinal hernias*

Musculoskeletal system (bones and joints)
- joint stiffness
- skeletal abnormalities (dysostosis multiplex)
- abnormal hip formation (hip dysplasia)

Brain and nerves
- pressure on the neck area of the spinal cord from abnormal tissue growth nearby (cervical spinal cord compression)
- fluid build-up in the brain (hydrocephalus)
- carpal tunnel syndrome
- potential developmental delays (i.e., the child takes longer to reach milestones such as sitting up, walking, and speaking) – however, some individuals may have normal intelligence.
- potential slowing of mental development by 2 to 4 years of age, followed by regression in skills

**Hernias: When part of an organ (such as the intestine) protrudes from a weak spot in the muscular wall surrounding the abdomen, producing a bulge in the skin, this is called a hernia. With an umbilical hernia, the bulge is in the belly button area. With an inguinal hernia, the bulge is in the groin area.*

Physical appearance

Stature
People with MPS II are often tall for their age until they are about 4 or 5 years old. After this point, their growth slows down and they tend to be shorter than unaffected individuals. They may have a bent-over stance due to joint contractures (stiffness). The may also have protruding bellies.

Facial features
MPS II individuals have similar facial features, which include a large head, a bulging forehead, and a broad nose with a flat bridge and wide upturned nostrils. They often have a large tongue, which may stick out. Their cheeks often appear rosy. These typical facial features are caused by the abnormal bones and cartilage of the face along with the GAG storage in the soft tissues of the face. You may hear doctors use the phrase “coarse facial features.” This is not meant to be insensitive but rather to help identify the features that point to the correct diagnosis.

Individuals with more slowly progressing disease will vary considerably in appearance and may not have the typical features described above or may show only subtle changes.

Spectrum of disease severity in MPS II:

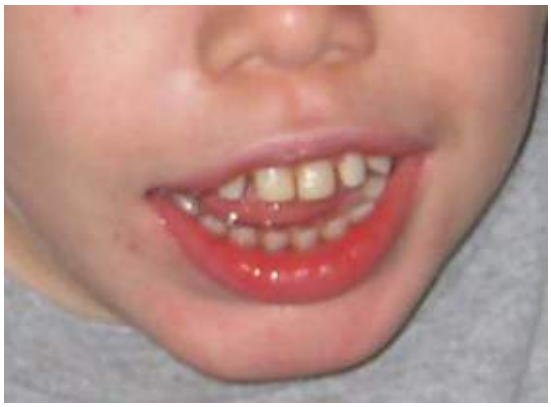
Rapidly progressing — *Slowly progressing*

Courtesy of the National MPS Society and Shire HGT

Mouth and teeth

People with MPS II generally have thick lips and an enlarged tongue. Their teeth have an irregular shape (sometimes described as being "peg shaped"), and their gums tend to be large and overgrown.

If teeth need to be removed while under an anesthetic, this should be done in the hospital under the care of both an experienced anesthetist and a dentist, never in the dentist's office.



Mouth and teeth of an individual with MPS II

Courtesy of Shire HGT

Skin

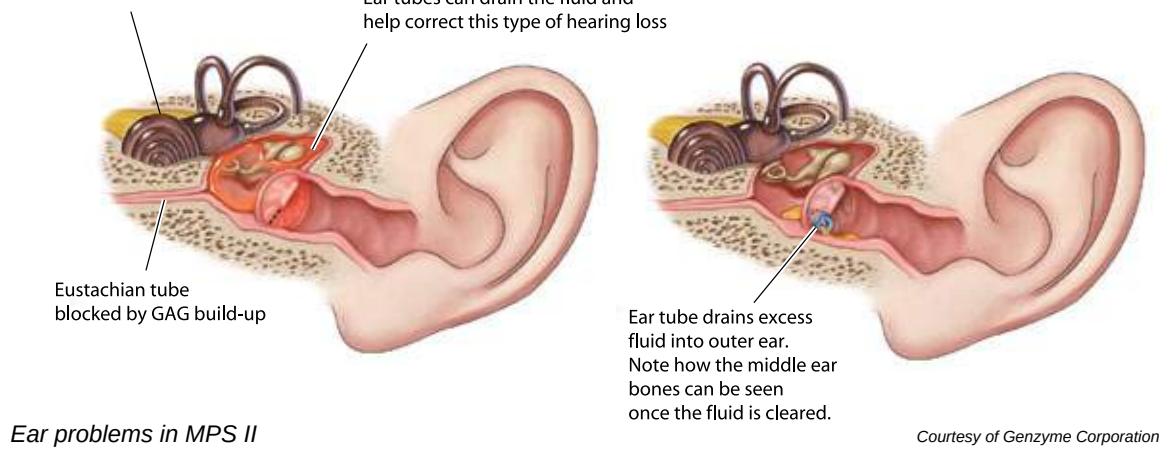
People with MPS II often have thickened and tough skin. Severely affected individuals may have more abundant or coarser hair on their face and body.

Some individuals with MPS II may have an ivory-coloured pebble-like texture to the skin on their back and shoulders, and in some cases on the arms and lower trunk. This is not a medical concern and is believed to be caused by GAG storage in the skin.

Eyes

MPS II symptoms affecting the eyes occur occasionally and include the following:

- *Glaucoma:* There may be problems with vision caused by glaucoma (increased pressure in the eye) that should be checked during an eye examination. However, glaucoma is not common in people with MPS II.
- *Retinal degeneration:* There may be problems with vision caused by changes to the retina resulting in night blindness. Night blindness can result in an individual not wanting to walk in a dark area at night or waking up at night and being afraid. Sometimes the addition of a night-light in a hall or bedroom is helpful. Regular annual eye exams are recommended.



Ear problems in MPS II

Courtesy of Genzyme Corporation

Ears

Deafness

Some degree of deafness is common in individuals with MPS II. This may be made worse by frequent ear infections. It is important that individuals with MPS II have their hearing checked regularly so that problems can be treated early to maximize their ability to learn and communicate.

Deafness in people with MPS II may be conductive deafness, sensorineural deafness, or both (see definitions below).

- *Conductive deafness:* Conductive deafness occurs when the transmission of sound through the outer and middle ear is blocked. In order for the middle ear to work properly, the pressure behind the eardrum has to be the same as that in the outer ear canal and the atmosphere. This pressure is equalized by the Eustachian tube, which runs to the middle ear from the back of the throat. 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, over time, become thick like glue. This is called middle ear effusion, and is an important factor contributing to hearing loss in people with MPS II. A small incision through the eardrum can be made (myringotomy) to remove the fluid. A small ventilation tube 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. It is expected that once the ventilation tube is in place, fluid should drain out and hearing should improve.
- *Sensorineural (nerve) deafness:* Sensorineural deafness occurs from damage to the sensory hair cells in the inner ear or to the nerves that transmit sound to the brain. In most cases, nerve or conductive deafness can be managed with a hearing aid or aids.

Otitis media

For parents of children with MPS II

What is otitis media?

Otitis media (OM) is the medical term for an infection of the middle ear. OM is a common problem encountered by healthy children, but it is one of the more stubborn problems for children with MPS II. Children with MPS II tend to have many recurrent ear infections. This is complicated by the fact that ear infections occur most frequently in the toddler and preschool years, often before a diagnosis of MPS or a related disease has been made.

There are 2 types of otitis media:

- *Acute otitis media:* This occurs when fluid is present in the middle ear, along with signs or symptoms of ear infection such as bulging eardrum often with pain, ear tugging, fever, irritability, decreased appetite, vomiting, and diarrhea. Complications, although rare, can include tympanic membrane perforation (broken eardrum), acute mastoiditis (an inflammation in the area surrounding the middle and inner ear), cholesteatoma (a mass of cells and cholesterol in the middle ear), or meningitis (a serious, potentially life-threatening inflammation of the membranes covering the brain). Language development can also be affected by repeated ear infections. In unaffected children, ear infections often result from Eustachian tube dysfunction related to allergies or a respiratory tract infection that causes swelling of the Eustachian tube and surrounding tissues.
- *Otitis media with effusion (OME):* OME is diagnosed when there is fluid in the middle ear without signs or symptoms of middle ear infection.

Otitis media (OM) is the medical term for an infection of the middle ear.

The child may not have any symptoms, but hearing can be affected. Any child who has fluid in the middle ears for at least 3 months should have a hearing test. Ear, nose, and throat (ENT) specialists, also called otolaryngologists, can help diagnose MPS II by identifying children with recurrent infections and abnormalities seen under examination. Once a diagnosis of MPS II has been made, the ENT specialist can be very helpful with many of the issues regarding managing the symptoms associated with the ears, nose, and throat.

Medication

Children with MPS II tend to have many ear infections that can be very difficult to treat. Many experts feel that if a child has ear infections that are hard to get rid of, it may be necessary for the doctor to do a "culture" of the fluid in the middle ear. The doctor will take a sample of this fluid and test it to see which bacteria, viruses, or fungi are living in the fluid. Identifying the bacteria, virus, or fungus that may be causing the infection allows the doctor to prescribe the appropriate medication. If the infection is fungal, frequent antibiotic use will only worsen the situation.

Antibiotics are the usual treatment for otitis media. There is a wide array of antibiotics available for treatment; however, some require refrigeration or frequent dosing. Antibiotic injections can be considered for a child who has difficulty taking medications by mouth. Some common side effects of antibiotics include diarrhea, nausea, and vomiting. Antibiotics may also cause skin rashes and allergic reactions.

In most cases of repeated ear infections, inserting tubes into the eardrum is recommended to allow the fluid to drain.

Corticosteroid medications (drugs like prednisone that reduce inflammation) may also be helpful. Children can receive a vaccine to protect against infection with Streptococcus pneumoniae, which is one of the more common bacteria causing problems.

Use of ear tubes

In most cases of repeated ear infections, inserting tubes into a hole in the eardrum (tympanostomy) is recommended to allow the fluid to drain. Tympanostomy tubes (also called ear tubes) may become blocked or infected. They may also damage or scar the eardrum. It is important to consult with an ear, nose, and throat (ENT) specialist experienced with MPS II to determine which tube is best. (Please note that many experts

Courtesy of Shire HGT

respond differently to various treatments, so every option should be tried if needed. Speak to your doctor before trying a new treatment, including herbal or alternative treatments.

MPS II can cause frequent ear infections, hearing loss, an enlarged tongue, and blocked airways. Any of these symptoms may lead to speech and language problems. A speech therapist may help those with MPS II with their speech. Hearing aids and sign language may also be useful for people with hearing loss.

A speech therapist may help those with MPS II with their speech.

recommend that ear tube insertion should be done at a properly equipped hospital and only after consultation with the anesthesiologist because of anesthesia concerns for children with MPS II. These concerns are covered in detail in the "Anesthesia considerations" section of this resource.) After the procedure, a culture should be made from the drained fluid to identify the offending organism.

Some experts believe that removal of the adenoids (tissues at the back of the nasal cavity) and tonsils might also be useful for children who have recurrent acute OM. If the child is to have general anesthesia for the placement of ear tubes, removal of the adenoids and tonsils should also be considered at the same time. This avoids some of the risk by reducing the number of procedures requiring anesthesia.

Prevention

Some experts feel that children may benefit from a vaccine for *Streptococcus pneumoniae*, which is one of the bacteria that cause ear infections. Vaccines may cause a fever or pain, redness, or swelling at the site of injection. More serious side effects include allergic reactions, but these are rare.

Some experts also state that exposure to second-hand cigarette smoke is recognized as a risk factor for OM, and every effort should be made keep children away from smoke exposure.

Ear infections occur repeatedly in children with MPS II, and anything that can help relieve the symptoms may be warranted. Each child may

Nose and throat

Runny nose: Typically, the bridge of the nose is flattened and the passage behind the nose may be smaller than usual because the bones in the mid-face have not grown well and the nose lining is thicker. GAG buildup in the soft tissues of the nose and throat, combined with abnormal bones, can cause the airway to become easily blocked. One of the most common features of children with MPS II is a chronic (long-term) discharge of clear mucus from the nose (rhinorrhea), and chronic ear and sinus infections.

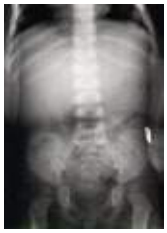
Throat: The adenoids (tissues at the back of the nasal cavity) and tonsils often become enlarged and can partly block the airway. The windpipe (trachea) becomes narrowed by stored GAGs and may be floppy or softer than usual. Excess tissue can further block the airway.

Respiratory system

Overview

Airway obstruction (blockage) is commonly seen in individuals with MPS II, who tend to have unusually narrow airways. In addition, the tonsils and adenoids (tissues at the back of the nasal cavity) can become enlarged and block the airway, which can also contribute to breathing difficulties. Finally, the shape of the chest is often

stiff, so it cannot move freely to allow the lungs to take in a large volume of air. The muscle at the base of the chest (diaphragm) may be pushed upwards by an enlarged liver and spleen (hepatosplenomegaly), further reducing the space for the lungs. A combination of these things can prevent the individual from breathing in adequate amounts of oxygen and can lead to difficulty breathing while awake or asleep. Individuals with MPS II have an increased risk of infection (pneumonia).



Chest X-ray of child with MPS I, a disease with similar clinical presentation to MPS II

Courtesy of Hodder Arnold Publishers

Many affected individuals breathe very noisily. At night they may be restless and snore. Sometimes the individual may stop breathing for short periods while asleep (sleep apnea). A sleep specialist can evaluate the individual with MPS II using a sleep study (see below for more information) to determine the presence and severity of sleep apnea. Sleep apnea can be treated in some cases by removing the tonsils and adenoids and/or opening up the airway with nighttime CPAP (continuous positive airway pressure), or by tracheostomy.

Managing breathing problems

The doctor may want you or your child to be admitted to the hospital overnight for a sleep study. Monitors are placed on the skin and connected to a computer to measure the levels of oxygen in the blood, breathing effort, brain waves during sleep, and other indicators of the body's function. From this study, doctors can assess how much blockage to breathing is present, how much trouble the person is having moving air into the lungs during sleep, and how much effect this has on the body.

Infections

Consult your doctor before using over-the-counter medications to treat respiratory infections. Drugs for controlling mucus production may not help. Drugs such as antihistamines (allergy medications) 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. Cough suppressants or drugs that are too sedating may cause more problems with sleep apnea by decreasing muscle tone and breathing rates.

People with MPS II are more prone to recurrent ear and sinus infections. Based on expert opinion, bacterial infections of the sinuses or middle ear should be treated with antibiotics as prescribed by the doctor.

There are many different antibiotics that may be used, and each one has side effects. Some common side effects of antibiotics include diarrhea, nausea, and vomiting. They may also cause skin rashes and allergic reactions. Since the sinuses and middle ear don't drain properly, overcoming infections can be difficult. It is common to have infections seem to go away while the individual is taking antibiotics and then come back after the antibiotic course is over.

Some people become allergic to antibiotics or may develop resistant infections. Your doctor can prescribe other antibiotics to help manage this problem. You will need a doctor with whom you can develop a good working relationship to manage the infections.

Sleep apnea

Obstructive sleep apnea is a common airway problem for individuals with MPS II. It is defined as temporary breathing interruptions during sleep that occur when the airway in the neck becomes blocked as muscles in the airway relax. The risk of the airway becoming blocked is increased by some of the physical effects of MPS II, including a narrow airway, and enlarged tonsils and adenoids.

This airflow can be improved by using a CPAP (continuous positive airway pressure) machine. An airway can also be opened using the tracheostomy procedure.

CPAP and BiPAP

Sleep apnea can be improved in some individuals by opening the airway with nighttime CPAP

treatments. An airway can also be opened using a tracheostomy (a surgical procedure to insert a breathing tube in the throat). For more information on tracheostomies, see "Tracheostomy" in this section.

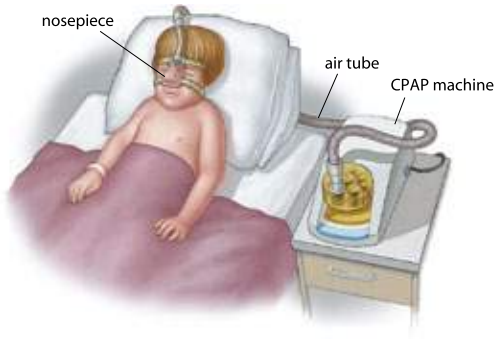
Both CPAP and BiPAP (bi-level positive airway pressure) are non-invasive. This means that they do not involve surgery, breaking the skin, or inserting a device into a body cavity. Instead, the treatments require that the individual wear a mask covering the nose and sometimes the mouth while sleeping in order to receive the positive airway pressure to keep the airway open. CPAP and BiPAP treatments are very effective treatments for sleep apnea, but they do not cure the underlying problem.

ficult. The mask does not breathe for the person. The airflow creates enough pressure during inspiration to keep the airway open. Another method for delivering air involves placing a tube into the nose in order to supply gentle air pressure to the airway.

Occasionally, CPAP can increase the work involved in breathing. In those cases, BiPAP is used instead. BiPAP stands for "bi-level positive airway pressure." It is also called non-invasive face mask ventilation. Bi-level means that the air pressure rises during inhalation and drops during exhalation, making breathing easier. BiPAP therapy is usually prescribed for people with sleep apnea if the CPAP therapy is too difficult. Occasionally the BiPAP machine will

Sleep apnea can be improved in some individuals by opening the airway with nighttime CPAP (continuous positive airway pressure) or BiPAP (bi-level positive airway pressure).

While CPAP and BiPAP are generally very similar, there are slight differences between them. BiPAP differs from CPAP in that the pressure during expiration (breathing out) may be adjusted separately from the pressure delivered during inspiration (breathing in). CPAP means there is a continuous supply of air at the same pressure being delivered to the patient with each breath. Nasal CPAP delivers air pressure through the nose. The mask is placed securely over the person's nose and slight positive air pressure is used to increase the amount of air being inhaled without making the work of breathing more difficult.



CPAP device *Courtesy of Genzyme Corporation*

be spontaneously timed (BiPAP SP), meaning that if for some reason the patient does not take a breath, the machine will automatically start a breath for them.

Obstacles to using CPAP and BiPAP

The major obstacle that most people must overcome is getting used to the CPAP or BiPAP system. The person must get used to sleeping while wearing the mask and mouthpiece. Approximately 20% of people never adjust or get accustomed to the treatment method. Some people feel that the device makes them feel claustrophobic. Others find that it is difficult to take with them when traveling.

Some complications can arise through the use of positive airway pressure devices. These complications can be associated with the equipment or with the person's condition. Mucus can build up in the nasal tubes. The person can also become uncomfortable if the pressure is set too high.

Determining whether it's needed

To determine whether someone has sleep apnea and may benefit from CPAP or BiPAP, a doctor can arrange for a sleep study. During a sleep study, the person is continually monitored for apnea episodes or decreases (desaturations) in his or her oxygen level. An abnormal sleep study may suggest that CPAP or BiPAP would be helpful to maintain an open airway during the night. Before considering any therapy or treatment, consult your pulmonologist (lung doctor).

How well a person does following a tracheostomy procedure depends greatly on their well-being prior to the surgery and on the specific reason the tracheostomy was performed. A person can expect to spend approximately 3–5 days in the hospital after a tracheostomy operation and usually an additional 2 weeks recovering.

A tracheostomy is generally a routine procedure; however, as with any other surgical procedure, there are risks. With the anesthesia, there is a risk

A tracheostomy (also called an artificial airway or "trach") is a surgically created opening through the neck into the trachea (windpipe). A tube is usually placed through the opening into the trachea. The function of the tube is to open an airway and to remove secretions from the lungs.

Tracheostomy

A tracheostomy (**tray**-kee-**oss**-ta-mee; also called an artificial airway or "trach," pronounced "trake") is a surgically created opening through the neck into the trachea (windpipe). A tube is usually placed through the opening into the trachea. This tube is referred to as a tracheostomy tube or a "trach" tube. The function of the tube is to open an airway and to remove secretions from the lungs.

A tracheostomy is usually performed under general anesthesia. After the area is cleaned, incisions are made to expose the outer wall of the trachea, which is made up of tough cartilage rings. A surgeon inserts the tracheostomy tube into the trachea after creating an opening through the cartilage rings.

There are 3 parts to the tracheostomy tube: the outer cannula, the inner cannula, and the obturator. The obturator is used for inserting the tube. The outer cannula is a tube that stays in the trachea all of the time, except for cleaning. The inner cannula is a safety valve to keep the airway open. This can be removed for cleaning.

of adverse reactions to medications and problems with breathing. Because people with MPS II are at a higher risk of problems with anesthesia, the tracheostomy should be done in a hospital that is fully equipped to deal with these issues. It is recommended that the anesthesiologist for the procedure has experience with MPS. See "Anesthesia considerations" for more information about anesthesia and MPS II.

Adjusting to a tracheostomy

Having a tracheostomy may also lead to significant differences in a person's lifestyle during the adjustment to having a trach placed. It is important to discuss trach care in detail with the doctors. The surgical incision needs to be cleaned frequently as it heals, perhaps as many as 4–5 times per day. Once the skin heals, it should be kept clean and dry. Most people use soap and water to clean the skin. Some people use a small amount of water-soluble antibiotic ointment around the skin incision.

Mucus secretions or blood can block the tracheostomy tube and interfere with breathing. The tube may be blocked if you notice bubbles in the trach tube, if you hear loud gurgles coming from the trach tube, or if the individual with the tube seems to be having difficulty breathing (for babies, the signs may include agitation, flared nostrils, increased heart rate, or pale or blue skin). If this occurs, the tube should be suctioned. People with tracheostomies can learn to suction

Individual with MPS II with tracheostomy tube

Courtesy of Shire HGT

their trach by using a suction machine and catheter as needed. If a child in your care has a tracheostomy, you may need to suction the trach for them.

From time to time, the tracheostomy tube will need to be changed. Changing an old tube for a fresh new tube can be challenging but becomes easier with time. Shortly after surgery, if the entry site has not healed properly, it may cave in when the tube is removed and block the trachea. When the new tube is being inserted, there is also a risk of the tube accidentally entering incorrectly. As the wound heals, the chance of either situation will decrease. Many people are eventually able to change their trach tubes in their home. If your child has a trach tube, you may need to change the tube for them.

One of the biggest challenges that people face following the insertion of the trach is adjusting to new breathing patterns and the changes to the vocal cords. Communication is perhaps the biggest adjustment because it may be impossible for the person to talk or make sounds. However, with proper training, many individuals can learn to speak with a tracheostomy tube.

Water-related activities can be hazardous to the person with a trach because there is not an easy way to hold their breath underwater and water could enter their lungs. Tub baths may be a reasonable solution for bathing. If a shower is preferred, it can be done with special care to shield the tracheostomy tube opening from the water.

A person with a trach also may benefit from using a cotton cover or scarf as a protection from inhaling dust and other particles.

With proper planning, discussion with doctors, and after-surgery care, a tracheostomy may significantly help individuals with MPS II whose upper airway is blocked.

Heart and blood vessels

Most people with MPS II have heart disease. It is a major cause of death for people with MPS II.

Effects on the heart valves

Many individuals with MPS II develop problems with their heart valves. Heart valves may leak or become narrowed because of GAG storage. There are 4 valves in the heart:

- The *tricuspid valve* is on the right side of the heart between the atrium (a collecting chamber for blood flowing back from the body) and ventricle (a muscular pumping chamber that pumps blood to the lungs). The valve prevents blood from flowing backwards into the right atrium when the right ventricle of the heart contracts.
- The *mitral valve* is on the left side of the heart between the atrium (a collecting chamber for blood flowing back from the lungs) and the ventricle (a muscular pumping chamber that pumps blood to the rest of the body). The valve prevents blood from flowing backwards into the left atrium when the left ventricle of the heart contracts.
- The *pulmonary valve* sits between the right ventricle and the pulmonary artery (the vessel that brings blood from the heart to the lungs).

The valve prevents blood from flowing backwards into the heart between its contractions.
- The *aortic valve* sits between the left ventricle and the aorta (the vessel that brings blood from the heart to the rest of the body). The valve prevents blood from flowing backwards into the heart between its contractions.

The doctor may hear heart murmurs (sounds caused by turbulence in blood flow in the heart) if the valves become damaged by stored GAGs. The heart valves are designed to close tightly in order to stop blood from flowing back in the wrong direction as blood passes from one chamber of the heart to another. If a valve is damaged by GAG accumulation, 2 different conditions may occur:

Heart valve damaged by MPS I, a disease with clinical presentation similar to MPS II.

Courtesy of J.E. Wraith

- *Regurgitation:* This occurs when the weakened valve cannot shut firmly enough and a small amount of blood may shoot backwards, leading to turbulence and a murmur. Conditions that involve regurgitation include mitral valve regurgitation (where the valve within the left side of the heart does not shut firmly enough) and aortic valve regurgitation (where the valve between the left side of the heart and the rest of the body does not shut firmly enough).
- *Stenosis:* This term refers to a stiffened heart valve. A stiffened heart valve may not be able to open completely. This means that the opening through which the blood is pumped will be smaller.

Eventually the damaged heart valves may need to be replaced surgically.

Effects on the heart muscle

MPS II-related damage to the heart valves can lead to left and right ventricular hypertrophy (enlargement of the pumping chambers of the heart) and heart failure (a condition where the heart cannot pump enough blood to meet the needs of the body). MPS II can also cause cardiomyopathy (abnormal heart muscle).

Effects on heart rhythm

Cardiomyopathy (see above) can increase the risk of arrhythmias (abnormal heart rate or rhythm).

Effects on the blood vessels

People with MPS II may suffer from pulmonary hypertension, a condition that causes high blood pressure in the blood vessels supplying the lungs. They may also have narrowing of the blood vessels that supply the heart with blood (the coronary arteries).

Rarely, adults with MPS II may develop a left ventricular aneurysm, a weakened out-pouching of the wall of the heart's main pumping chamber. These aneurysms can be identified by MRI. Left untreated, they may rupture and cause sudden death.

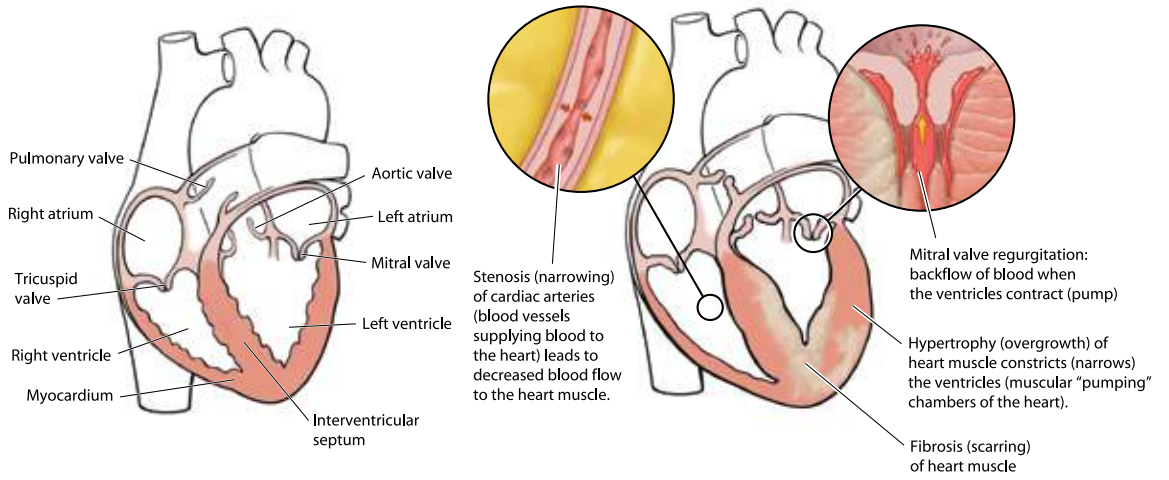
Effects on the electrical system of the heart

GAGs can get into the heart's electrical system and interrupt electrical signals to the pumping chambers of the heart. About 11% of individuals with MPS II experience unexpected sudden death, and in some cases this may be due to an abnormality of the heart's conduction system (heart block). If heart block occurs, a cardiac pacemaker should be considered. Speak to your doctor to learn more.

The importance of regular heart checkups

Since heart problems occur so frequently in MPS II, all individuals with MPS II should have a test known as an echocardiogram regularly (as often as your doctor thinks necessary) to show whether any problems are beginning.

Because of the unusual special problems that can occur in these disorders, you should choose a cardiologist with some knowledge of MPS. If this is not possible, you should inform the doctor about the heart problems experienced by individuals with MPS II.



Normal heart (left) and heart problems in MPS II (right)

Courtesy of Genzyme Corporation

Some experts believe that medications may be used to help manage the heart problems that occur as a result of MPS II.

Gastrointestinal system

Liver and spleen

Accumulation of GAGs may cause enlargement of the liver (hepatomegaly) in most people with MPS II. The large liver does not usually cause liver or spleen problems or lead to liver or spleen dysfunction.

Abdomen and hernias

In individuals with MPS II, the abdomen may bulge out due to the weakness of the abdominal 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. The hernia can come from behind the navel (umbilical hernia) or in the groin (inguinal hernia). Inguinal (groin) hernias should be repaired by an operation, but hernias will sometimes recur. Umbilical (navel) hernias are not usually treated unless they cause entrapment of the intestine (intestine gets caught in the abdominal opening, which cuts off its blood supply) or are very large and are causing problems. It is very common for an umbilical hernia to reappear after it has been repaired.

Hernia in a child with MPS VI, a condition with clinical presentation similar to MPS II

Courtesy of the National MPS Society

Bowel problems

Many individuals with MPS II suffer periodically from loose stools and diarrhea. The cause of this is not fully understood. It is thought that there may be a problem with the autonomic nervous system, the system that controls those bodily functions usually beyond voluntary control. Studies have found GAG buildup in the nerve cells of the intestine, and it is possible that the diarrhea is caused by abnormal movement of the bowel.

An examination by a physician, supplemented by an X-ray if necessary, may establish the cause of diarrhea. For children, the problem may disappear as the child gets older, but it can be made worse by antibiotics prescribed for other problems. Diet changes may help control diarrhea. If antibiotics have caused the diarrhea, eating plain live-culture yogurt is often helpful during

episodes of diarrhea. This provides a source of lactobacillus (a "friendly" bacteria in the bowel) to help prevent the growth of harmful organisms within the bowel wall, which can cause diarrhea or make it worse.

Constipation may become a problem as children with MPS II get older and less active and as the muscles weaken.

Dietary considerations

Some experts believe that reducing intake of milk, dairy products, and sugar, as well as avoiding foods with too many additives and colouring, may help some individuals. It would be advisable to consult your doctor or a dietitian if you plan major dietary changes to make sure that the proposed diet does not leave out any essential items. If your child's problems are eased, you could try reintroducing foods one at a time to test whether any particular item seems to increase the child's symptoms.

Feeding tubes

Why use tube feeding?

Individuals with MPS II may have problems chewing and swallowing as the disease progresses. People with lysosomal storage disorders (such as MPS II) who have chewing and swallowing problems are at risk of poor nutrition and aspiration. "Aspiration" means inhaling food or other substances into the lungs. Aspiration can lead to pneumonia. Because of the difficulties with swallowing and the risk of aspiration, individuals with lysosomal storage disorders (such as MPS II) may need a feeding tube.

A process called "tube feeding" can be used to help individuals with MPS II get the nutrition they need and protect them from choking or aspirating. It also makes it easier and faster for a caregiver to feed the individual with MPS II. A flexible feeding tube is inserted that bypasses the mouth and throat and goes directly into the stomach or intestine. Tube feeding is also called "enteral nutrition."

Making the decision

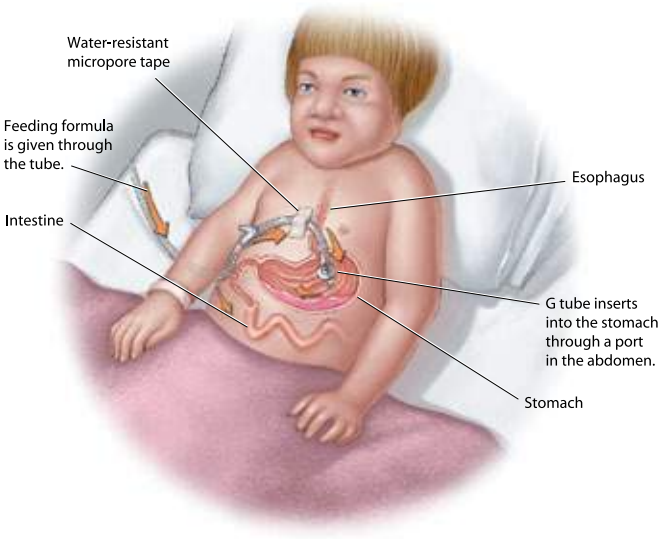
The decision to change to enteral nutrition is a difficult one. You may wish to consult your pediatrician, geneticist, gastroenterologist, and nutritionist. To help with the decision, it's important to keep track of the affected person's intake of food, weight gain or loss, choking and gagging, episodes of pneumonia, and time required for feeding. This will help you and your health professionals decide whether enteral nutrition is needed.

G-tubes and J-tubes

Tube feeding is usually done through either a gastrostomy tube (G-tube) or a jejunostomy tube (J-tube). A G-tube goes into the stomach through a surgical opening in the abdominal wall. A special kind of G-tube tube may be inserted by means of an endoscopic procedure (which uses a camera on a flexible tube to see inside the body) and is called a percutaneous endoscopic gastrostomy (PEG) tube. A J-tube is usually surgically placed through the abdominal wall into the part of the small intestine called the jejunum. Each tube is a flexible (usually silicone) catheter that remains in place at all times and is clamped between feedings to prevent leakage of stomach contents.

Because of difficulties with swallowing and the risk of aspiration, individuals with lysosomal storage disorders (such as MPS II) may need a feeding tube.

G-tube feeding can be done at regular mealtimes. It can be given at once, called bolus feeding, or it can be given slowly over a period of several hours using the gravity (drip) method or the pump-controlled method. Each method has its advantages and disadvantages, and many factors



Gastrostomy tube (G-tube) *Courtesy of Genzyme Corporation*

must be considered when choosing a method. J-tube feedings are continuously infused because the small intestine is pressure sensitive.

After the decision is made to insert a feeding tube, the doctor will perform X-rays of the gastrointestinal tract (stomach and intestines) to help decide which type of tube to use. The doctor will also check for gastroesophageal reflux disease (GERD) because tube placement may worsen existing GERD and a J-tube may be a better choice. A J-tube may also be an option if there is very poor motility (spontaneous movement) of the stomach. Because of special concerns regarding anesthesia in MPS II patients, you or your doctor should consult with an anesthesiologist before surgery is done to insert the tube. See "Anesthesia considerations" for more information about anesthesia and MPS II.

Caring for the tube

The surgical opening for the G-tube or J-tube is called a stoma. The stoma can be slow to heal after surgery. Proper care of the stoma site is very important to avoid infection or irritation from stomach and intestinal juices. The area should be kept covered with a dressing and changed as often as needed to keep dry. The skin around the stoma should stay snug around the tube. Swimming in lakes or ponds should be avoided because the bacteria living in these areas may infect the stoma site.

A G-tube is anchored inside the stomach by a small balloon at the tip of the tube. The balloon can deteriorate and deflate and the tube can fall out. The doctor will provide you with a replacement tube and instructions on how to insert it. Only a doctor can reinsert the J-tube. Contact the doctor immediately if the J-tube falls out. Also, these tubes can become clogged. Prepare for this by discussing with your doctor appropriate methods to unclog them.

The Mic-key low-profile gastrostomy feeding tube/kit is a skin-level device to replace the gastrostomy tube. Because this device is level with the skin, it is less likely to be pulled out and can easily be covered by clothes. A special connector allows the G-tube to be removed between feedings.

When and how to feed

The best tube feeding schedule will allow the person to maintain an adequate weight, tolerate the tube feedings comfortably, and be fed at convenient times. Caregivers should have contact with a nutritionist to regularly discuss the individual's feeding needs. For most individuals, regular solutions such as PediaSure® (Abbott Laboratories), Resource® (Nestlé Nutrition), or Kindercal® (Mead Johnson Nutrition) are sufficient to fill their needs. The formulas are generally tolerated with little difficulty.

Good positioning during feedings is critical. If the individual is not positioned well, he or she may have trouble receiving food through the tube or breathing properly. The person should not be sitting slumped over, as this can put too much pressure on the stomach. If the individual has trouble maintaining an upright position, special equipment and supports are available to help.

If a person with MPS II begins to have trouble eating, it is important to begin keeping track of food intake and weight. This will help to determine if another method of feeding needs to be considered. The decision to switch to enteral nutrition is not an easy one to make, but your healthcare team can help you weigh the risks and benefits. Difficulties that may be encountered are best dealt with by the medical team in charge of the person's medical care. Continued contact with the team is essential for successful enteral feeding (i.e., enteral feeding that meets the individual's nutritional needs).

Problems that may be associated with tube feeding

Feeding tubes may become blocked or clogged. Fortunately, they can often be unclogged at home. If the tube cannot be unclogged, it may be changed. Check with your doctor to find out how to unclog and change the tube so that you will be prepared in case this occurs.

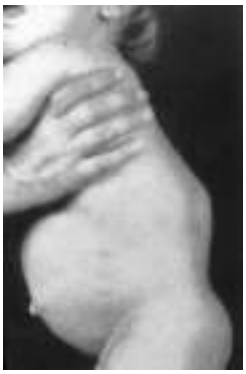
The person receiving tube feeding may accidentally inhale food or liquid into the lungs. This is called "aspiration," and it can lead to pneumonia. Coughing around feeding time is a sign of possible aspiration. Coughing, difficulty breathing, and fever are signs that you or your child may have pneumonia. If any of these things occur, contact a physician.

The stoma may become infected. Signs of infection include fever, pain, swelling, warmth, or increased redness near the stoma. If you notice signs of infection, contact the doctor.

A person who is being tube fed may also experience bloating, diarrhea, or vomiting. These problems may be due to changes in feeding formula, giving too much feeding formula at once, problems digesting the feeding formula, or contamination of the feeding formula with germs.

Musculoskeletal system (bones and joints)

People with MPS II tend to have significant problems with bone abnormalities and joint stiffness. The bone problems are also called dysostosis multiplex.



Gibbus in a child with MPS I, a disease with clinical presentation similar to MPS II

Courtesy of Dr. Emil Kakkis

Spine

The bones of the spine (vertebrae) normally line up from the neck to the buttocks. More severely affected individuals with MPS II often have poorly formed vertebrae that may not stably interact with each other. Backward slippage of the vertebrae can occur, causing an angular curve, called kyphosis or gibbus, to develop.

Neck

The spinal canal can be narrowed in individuals with MPS II, the transverse atlantal ligament can become overgrown (the transverse atlantal ligament is a strong tissue band that helps hold the odontoid process, an important bone for stabilizing the neck, in place), and the bones that stabilize the connection between head and neck can be malformed (odontoid dysplasia) in more rapidly progressing individuals, making the neck unstable. This puts people with MPS II at risk of spinal cord compression (a condition where fluid or tissues such as bones are pressing on the spinal cord). Parents of children with MPS II should be cautious about how the area of the spine around the neck is handled. Children with MPS II should avoid "high risk" activities such as contact sports and gymnastics. In addition, these children should be treated with caution when undergoing positioning for anesthesia. If there is severe pain or pain associated with

Joint stiffness in an individual with MPS VI, a condition with a clinical presentation similar to MPS II

Courtesy of the National MPS Society

weakness or tremors in the lower legs, the person should have studies of the neck to evaluate for slippage of the neck vertebrae.

Joints

Joint stiffness is common in individuals with MPS II, and the maximum range of movement of all joints may become limited. Joint stiffness and tight heel cords cause many people with MPS II to walk on their toes.

Hands

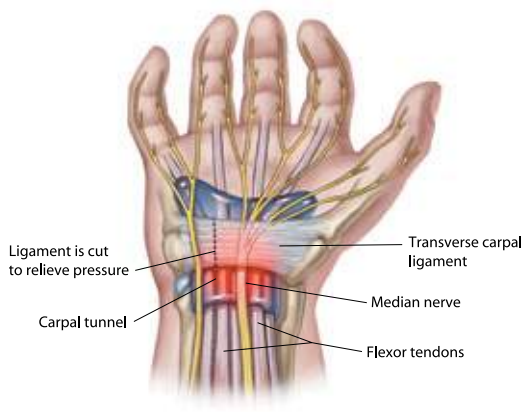
The hands of people with MPS II are short and broad with stubby fingers. Over time, the fingers stiffen and gradually become curved, due to limited joint movement. The tips of the fingers can become permanently bent over, giving rise to the characteristic bent, stiff hand. The medical term for bent and stiff hands is camptodactyly, but they are often referred to as "claw hands."

Bent/stiff hands

Courtesy of Shire HGT

Carpal tunnel syndrome

People with MPS II sometimes experience pain and loss of feeling in the fingertips as a result of carpal tunnel syndrome. Other symptoms of carpal tunnel syndrome may include clumsiness with the hands, problems grasping objects, avoidance of using the hands for daily activities, gnawing of hands, and weakness in the hand. Some individuals affected by MPS II may have few or no symptoms of carpal tunnel syndrome because the nerves have been damaged for so long.



Carpal tunnel syndrome and carpal tunnel release surgery

Courtesy of Genzyme Corporation

The causes of carpal tunnel syndrome in MPS II are related to GAG storage in the hand tissues, and trapping of the nerves within the hand and wrist. In some cases, surgery may be used to treat carpal tunnel syndrome.

As with any surgical procedure for a person with MPS II, it is important to meet with the anesthesiologist prior to the surgery.

Hips

Like the spine, the hip joints suffer from altered bone formation. The hip joints are ball-and-socket joints situated at either side of the pelvis. The "ball" is the head of the femur (thigh bone) and the "socket" is the cupped part of the pelvis (the acetabulum) that surrounds the ball. In abnormal formation of the hip, or hip dysplasia, there is a shallow acetabulum, the head of the femur is underdeveloped, and the top of the thigh bone at the neck of the femur is straightened (a condition called coxa valga). This combination of bone defects results in hip instability and sometimes dislocation.

Hip damage in MPS II can be difficult to manage surgically. Some experts believe that surgery on the hips is done more easily at a younger age, around 5–7 years old, for the best results. Successful surgery (i.e., surgery that is able to correct the hip dysplasia) becomes much more difficult at older ages. Once the hips have been severely damaged, the surgery becomes technically very difficult, and the results are much less predictable.

Hip surgery for dysplasia is a combination of precise bone cuts, or osteotomies, which allow the surgeon to reposition the bones and optimize the working of the hip. Cuts are made in the pelvis and sometimes the femur. The surgery on the bones may be performed in conjunction with tightening the soft tissues around the hip.

Without hip surgery, there is progressive pain and stiffness, and eventually dislocation of the hips, resulting in a greatly decreased ability to walk.

Hip surgery carries a number of risks, including the risks associated with anesthesia (see "What are the special considerations when planning an anesthetic for a child with MPS II or a similar condition?" in "Anesthesia considerations" for more information), infection, bleeding, or blood clots. Physical therapy may also be helpful for people with MPS II-related hip problems. It can help preserve hip joint function and should be started while the child is young.

Physical therapy may help relieve symptoms and improve the person's ability to function.

Legs and feet

Many people with MPS II walk on their toes because of tight heel cords and joint stiffness. In MPS II, all joints, including those in the legs and feet, may be affected by joint damage and restriction of motion.

Physical and occupational therapy

Physical therapy has not been well studied in MPS II, but range-of-motion exercises (passive stretching and bending of the limbs such as arms and legs) may offer some benefits in preserving joint function, and should be started early. If joint motion is already limited, these exercises may limit further losses of flexibility. The doctor or physical therapist may be able to suggest appropriate exercises to help with joint motion.

Physical therapy session

Courtesy of Genzyme Corporation

Brain and central nervous system (CNS)

Overview

Many people on the severe end of the spectrum notice a progressive and possibly severe decline in developmental function. However, it is important to note that not all rapidly progressing individuals experience this neurologic decline. Individuals with mild (slowly progressing) MPS II are less likely to be affected this way and often maintain normal or near-normal intelligence.

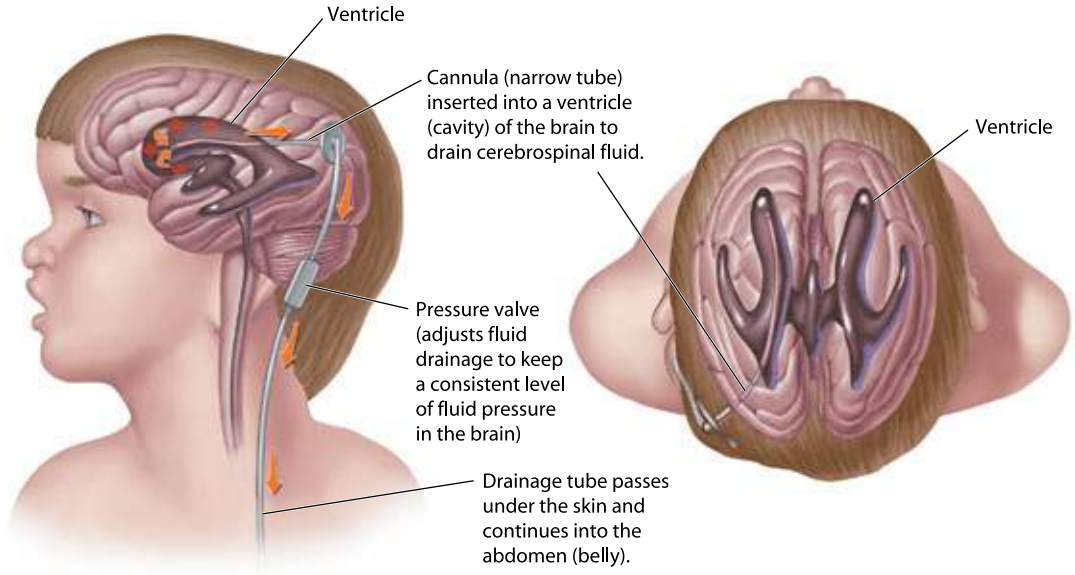
Other aspects of MPS II that may affect intellectual function include impaired hearing, sleep deprivation due to sleep apnea and increased fluid pressure in and around the brain (hydrocephalus).

Cognitive function

At birth, individuals with MPS II may seem to have normal cognitive function (the ability to think and process information). Many infants with MPS II reach early developmental milestones in a normal way. However, after this, there is a wide spectrum of possible developmental outcomes.

More severely affected individuals with MPS II will have some developmental delay by 18 to 24 months of age. Then, slow progress usually continues until the child reaches a developmental plateau by the age of 3 to 5 years. After this, intelligence and mental abilities gradually decline. Eventually, most severely affected individuals have severe mental handicaps and are usually dependent on caregivers for their activities of daily living.

On the other end of the spectrum, people with attenuated MPS II may have normal or near-



Shunt for hydrocephalus

Courtesy of Genzyme Corporation

normal intelligence. However, their brain scans may still show abnormalities.

Hydrocephalus

MPS II can cause hydrocephalus, a condition where fluid accumulates in the brain, causing a pressure buildup that can lead to brain damage. This is more common in individuals with severe neurological (brain and nerve) symptoms.

Hydrocephalus was once known as "water on the brain." The "water" is actually cerebrospinal fluid (CSF), a clear fluid surrounding the brain and spinal cord. The CSF protects the brain and spinal cord from injury by providing a liquid cushion, and is continually being produced, circulated, and absorbed. Communicating hydrocephalus (also known as "non-obstructive hydrocephalus") is caused when the CSF is not absorbed properly. This causes the CSF to build up, leading to an abnormal enlargement of the spaces in the brain called ventricles. This causes potentially harmful pressure on the tissues of the brain.

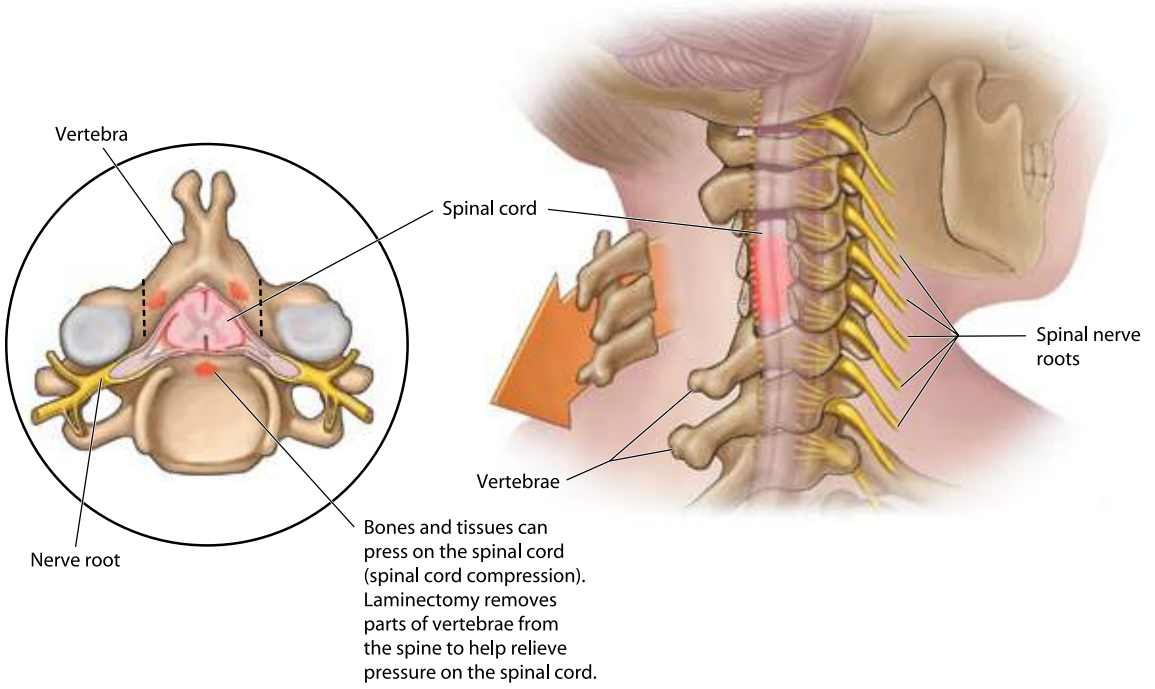
Effects of hydrocephalus

In infants, the most obvious sign of hydrocephalus is often a rapid increase in head circumference or an unusually large head size. In older children and adults, typical symptoms may include a headache followed by vomiting, nausea, blurred or double vision, downward deviation of the eyes (called "sunsetting"), problems with balance, poor coordination, abnormal walking patterns, urinary incontinence (difficulty holding urine), slowing or loss of development, lethargy, drowsiness, irritability, memory loss, or other changes in personality or thinking. If hydrocephalus develops slowly, these typical signs and symptoms may not be seen.

Diagnosing hydrocephalus

Hydrocephalus is diagnosed through clinical neurological evaluation (where the doctor checks the individual's brain and nerve function); by using imaging techniques such as ultrasound, computer tomography (CT), and magnetic resonance imaging (MRI); and through techniques to measure pressure, such as lumbar puncture (spinal tap).

Measuring intracranial pressure (pressure inside the brain) allows the doctor to diagnose hydrocephalus. Once the fluid buildup is too severe, the doctor may recommend a shunt (see below).



Laminectomy for spinal cord compression

Courtesy of Genzyme Corporation

Use of shunts

Hydrocephalus is most often treated with the surgical placement of a shunt. A shunt is a flexible plastic tube (cannula) that diverts the flow of CSF from the brain to another area of the body where it can be absorbed as part of the circulatory process. If a shunt is placed, specialists recommend a high-pressure shunt to prevent rapid decompression (reduction of fluid in the ventricles). Shunts must be inserted surgically. Before surgery, doctors should check for signs of blockage in the form of spinal cord compression, which is described below.

Spinal cord compression

As a result of GAG accumulation, over time the tissues and ligaments around the spinal cord may gradually become thicker and start pressing against the spinal cord. This may result in a condition called spinal cord compression. As a result of this compression, individuals with MPS II may experience a range of symptoms, including spasticity, weakness and altered sensations below the neck, or difficulty urinating.

The main method used to relieve this condition is surgical decompression. Surgical decompression may be done through a surgical procedure called a laminectomy. Possible complications that may occur during surgical decompression include a slowing of the heart rate or difficulty breathing.

As with any surgical procedure for a person with MPS II, it is important to meet with the anesthesiologist prior to the surgery.

Seizures

Seizures (convulsions) are common in severely affected individuals with MPS II: over half of severely affected individuals with MPS II who reach the age of 10 years will suffer from seizures. In attenuated individuals, seizures are not common.

Seizures in MPS II individuals can be treated with anticonvulsant medications.

Management options

Overview

Currently there is no cure for MPS II. Management options for MPS II include those aimed at disease management and supportive or palliative care (care that makes a person with a disease that cannot be cured more comfortable), as well as those targeted to replace the deficient enzyme.

The decision of which interventions and treatments are best for you or your child is an important and complex one. The information in this section is not intended to be medical advice or recommendations. Decisions about interventions and treatments for you or your child are best discussed with medical professionals with experience in MPS.

Importance of multi-disciplinary care

As described earlier in this resource, people with MPS II usually have a wide range of signs and symptoms. As a result, they often need to be managed by a large number and variety of medical specialists, including cardiologists, gastroenterologists, pulmonologists, otolaryngologists, ophthalmologists, orthopedic surgeons, physical therapists, and speech therapists. All such health professionals involved in the care of people with MPS II should have a basic understanding of the disease and how the condition may affect treatment decisions.

However, it can also be very helpful to have a single physician with experience in MPS II who takes responsibility for overseeing the overall care across medical specialties and who keeps track of the "big picture." This physician can then refer the person to other specialists as needed and help make sure the individual is receiving the best possible care. For many people with MPS II, the physician who performs this role is usually either their primary care physician (who might be a pediatrician) or a geneticist.

Disease management and supportive care

Supportive and disease management therapies may improve quality of life for people with MPS II and their caregivers, and may increase an affected person's lifespan. Supportive care measures – such as the use of heart valve replacement surgery or shunts – have already been described earlier in this resource, and are included along with the description of the symptoms that they are intended to address. For people who plan to have surgical interventions, it's important that the health professionals involved in the surgery, especially the anesthesiologist, have a good understanding of the potential risks and complications for individuals with MPS.

Enzyme replacement therapy (ERT)

Overview of ERT

Enzyme replacement therapy (ERT) provides the body with an external source of I2S, the enzyme that is missing or deficient in people with MPS II. It is given via an intravenous (IV)

Enzyme replacement therapy (ERT) is designed to address the underlying deficiency of iduronate-2-sulfatase (I2S) enzyme in MPS II, which leads to GAG buildup in the cells of various organs.

Child receiving ERT

Courtesy of Genzyme Corporation

infusion. The enzyme travels through the bloodstream and enters cells in various organs of the body, where it helps break down GAG buildup.

ELAPRASE® (idursulfase) is an enzyme replacement therapy that provides a manufactured version of the body's natural I2S enzyme. It is indicated for people with mucopolysaccharidosis II (MPS II). It has been shown to improve walking capacity in these patients.

WARNINGS AND PRECAUTIONS

Serious Warnings and Precautions

RISK OF HYPERSENSITIVITY REACTIONS.

Anaphylactoid reactions, which have the potential to be life threatening, have been observed in some patients treated with ELAPRASE.

Patients with compromised respiratory function or acute respiratory disease may be at risk of serious exacerbation of their respiratory dysfunction due to infusion related reactions. These patients require additional monitoring. Late-emergent anaphylactoid reactions have been observed after ELAPRASE administration. Patients who have experienced severe and refractory anaphylactoid reactions may require prolonged observation times.

Due to the potential for severe infusion reactions appropriate medical support measures should be readily available when ELAPRASE is administered.

ELAPRASE® Safety Information

The safety and efficacy of ERT have not been established in children less than 5 years of age or in people aged 65 years and over.

Patients who are hypersensitive to this drug or to any ingredient in the formulation or component of the container should not receive ELAPRASE®.

The most common side effects associated with ELAPRASE® treatment include:

Very common side effects (more than 1 in 10):

- headache
- increased blood pressure
- chest pain
- hives, rash, itching
- fever and infusion site swelling

Common side effects (more than 1 in 100):

- dizziness, tremor
- teary eyes
- changes in the way your heart beats, bluish skin
- decreased blood pressure, flushing (redness)
- difficulty breathing, wheezing, blood clot in the lung artery, cough, quickened breathing
- abdominal pain, nausea, diarrhea, swollen tongue
- facial swelling, skin lesions (redness, eczema)
- pain in the joints
- swelling of the extremities

Serious side effects, how often they happen and what to do about them

Frequency	Symptom/effect	What to do
Common (occurring in ≥ 5% of patients)	Drop in blood oxygen level from difficulty in breathing	Stop taking drug and call your doctor or pharmacist
Uncommon (occurring in < 5% of patients)	Seizure, blood clot in the lungs, missed or extra heartbeats	Stop taking drug and call your doctor or pharmacist

In clinical trials with ELAPRASE®, 11 of 108 patients (10%) experienced allergic reactions during 19 of 8,274 infusions (0.2%). Reactions

have included temporary breathing difficulty, decreased blood pressure, or swelling. In more severe reactions, such as in one particular patient, a seizure occurred because of a drop in blood oxygen level from difficulty in breathing.

Inform your doctor immediately if you have any of these side effects or other side effects not mentioned here. **This is not a complete list of side effects. If you experience any unexpected effects, contact your doctor or pharmacist.**

For the most up-to-date prescribing information, including important safety information, please refer to the ELAPRASE® Canadian Product Monograph. For more information about treatment with ELAPRASE® please call Paladin Labs Inc. at 1-888-550-6080.

Permanent catheter (port) insertion

Some patients prefer to have a permanent catheter (port) surgically implanted under the skin to reduce the trauma of having to find a vein each week. One end of the port is stitched into a major vein. The other end has a large rubber septum (the place where medication is injected into the catheter), which is easier to use for injecting medication into than the traditional method of finding a new IV site each week. You should discuss with your physician whether this approach is appropriate for you or your child.

Where can I get more information and help?

Whether you are still making a decision regarding treatment options, or you have already made a decision and have subsequent questions, below you will find additional sources of information to help you:

The Canadian Society for Mucopolysaccharide & Related Diseases

The Canadian Society for Mucopolysaccharide & Related Diseases (Canadian MPS Society) provides support for individuals and families living with MPS II and other MPS disorders. The Society can connect you with other families affected by MPS II as well as families of people who are

Courtesy of the Canadian MPS Society

currently receiving ELAPRASE® (idursulfase) ERT. The staff in the office can talk with you about the disease and treatment information you are looking for. Please call 1-800-667-1846 or visit www.mpssociety.ca. The Society also performs a variety of other functions that are not related to treatment, and that are described in the "Living with MPS II" section of this resource.

Shire HGT

Shire HGT is committed to helping treat individuals with MPS II and to supporting people with MPS II and their families as they face the daily challenge of living with the disease.

Additionally, Shire HGT has a reimbursement support program for patients receiving ELAPRASE and who have access to private health insurance coverage. For more information about this program call 1-866-773-6302.

If you have medical inquiries about ELAPRASE you can contact Paladin Labs Inc. at 1-888-550-6060.

Other websites

Other websites can also provide you with more information about your current and future treatment and management options, including:

- **www.mpssociety.ca**
  The website of the Canadian MPS Society offers information and support for Canadians affected by MPS II.

- **www.mpssociety.org**
  The website of the National MPS Society offers an MPS II booklet that provides information on the features and treatment of MPS II.

- **www.raredisorders.ca**
  This is the website of the Canadian Organization for Rare Disorders, which is a national network representing all Canadians with rare disorders.

- **www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=309900**
  This website, Online Mendelian Inheritance in Man (OMIM), provides detailed information on inherited conditions such as MPS II.

- **www.nlm.nih.gov/medlineplus/ency/article/001203.htm**
  MedlinePlus, a service provided by the US National Library of Medicine, offers a summary of information about MPS II, including causes, symptoms, medical tests, treatment, and prognosis.

- **www.clinicaltrials.gov**
  A website maintained by the US government that includes a listing of active clinical studies, including those that are related to the study of various treatment options for individuals with MPS II.

Rare disease research

Overview

There are many types of ongoing research for a host of rare diseases. Not all research described here is necessarily for MPS II or any MPS disorder. Research is considered investigational, as evidence does not exist that it will be safe or effective. The following descriptions are intended to provide you with a better understanding of the research areas. Please check with the Canadian Society for Mucopolysaccharide & Related Diseases (Canadian MPS Society) or your healthcare provider for more specific information. Additionally, the websites on the previous page may be a good resource.

The mission of the Canadian MPS Society is to find cures for MPS and related diseases, and as part of that mission, the Society funds research grants. The Society recognizes the need for targeted research for treatment of bone and joint problems and for treating the brain, and recent research funding from the Society has focused on these areas, as well as on biomarker research. Information about the Society's research program can be obtained through the Society office by calling 1-800-667-1846, or on the Society website at www.mpssociety.ca .

Hematopoietic stem cell transplant (HSCT)

Hematopoietic stem cells are blood cells at their earliest stage of development. At this stage, they can develop into three different types of cells: red blood cells, which carry oxygen in the blood; white blood cells, which help the immune system function; and platelets, which help the blood clot.

In a hematopoietic stem cell transplant (HSCT), the recipient is first conditioned with chemotherapy medications to eliminate the cells in the bone marrow, which cannot produce the enzyme. Then stem cells are given intravenously (through a vein) to the recipient. The healthy stem cells travel to the recipient's bone marrow, where they

grow and produce blood cells capable of making the missing enzyme. The goal is to help replace the enzyme that is missing.

HSCT may use stem cells from different sources including bone marrow and umbilical cord blood. Bone marrow transplants are procedures that make use of stem cells from the marrow within the bones of healthy donors, while cord blood transplants make use of stem cells from the blood of the umbilical cords of healthy newborns.

HSCT is not usually recommended as a treatment for MPS II.

To learn more about HSCT research, check with the Canadian MPS Society or your healthcare provider, or visit www.clinicaltrials.gov.

Gene therapy

Gene therapy involves replacing the defective gene with a healthy gene capable of producing enzyme (iduronate-2-sulfatase [I2S]). The main issue with gene therapy is delivery (how to get the healthy gene to all cells of the body). One possible delivery method is cell transplantation. With this technique, a sample of the person's own cells is removed and then treated outside the body. The treatment alters the genes in the cells so that they produce the enzyme. Then the cells are put back into the person's body, where they begin to produce cells with a healthy copy of the gene for the enzyme.

Another potential delivery method for gene therapy is in utero gene therapy. With this therapy, a healthy version of the gene would be injected into the fetus while it is still in the mother's womb, with the goal of producing a baby with healthy enzyme activity.

To learn more about gene therapy research, check with the Canadian MPS Society or your healthcare provider, or visit www.clinicaltrials.gov.

Cell therapy

Another area of research for rare diseases is cell therapy. This type of therapy involves implanting cells, such as muscle or blood cells, into a person's body with the intention of treating a disease. These cells may come from the person's own body or from a donor. Cells may need to undergo treatments to increase their ability to help fight the disease. For example, the cells may be modified to increase the amount of enzyme they produce. The cells may be enclosed in a special coating called a "microcapsule" to help protect them from attacks from the body's immune system. They can then be implanted into the body.

To learn more about cell therapy research, check with the Canadian MPS Society or your healthcare provider, or visit www.clinicaltrials.gov.

Intrathecal therapy

Intrathecal therapy is a technique involving the injection of a medication or enzyme into the fluid around the brain and spinal cord (the cerebrospinal fluid). The goal is for the therapy to reach the tissues in the brain. It can be difficult to get medications and other substances into the brain because the brain is protected by the blood-brain barrier, a network of modified blood vessels that blocks many substances in the blood from entering the brain. Intrathecal therapy is a way of getting past the blood-brain barrier

and allowing the treatment to reach the brain. Intrathecal therapy is given by inserting a hollow needle in between the bones of the lower spine to gain access to the cerebrospinal fluid, then injecting the medication into the fluid.

To learn more about intrathecal therapy research, check with the Canadian MPS Society or your healthcare provider, or visit www.clinicaltrials.gov.

Substrate deprivation therapy

Substrate deprivation therapy (SDT) is a technique that aims to reduce the rate at which unwanted substances build up in the body. Usually, the unwanted substance is something that could normally be broken down by the body, but cannot be broken down by people with a

particular disease. With SDT, small molecules are introduced into the body. These molecules block the production of the unwanted substance with the goal of reducing the rate at which it builds up inside the body.

To learn more about SDT research, check with the Canadian MPS Society or your healthcare provider, or visit www.clinicaltrials.gov.

Hunter Outcome Survey

What is the Hunter Outcome Survey?

The Hunter Outcome Survey (HOS) is a global patient data survey for people with Hunter Syndrome (MPS II). The purpose of HOS is to collect data on people with Hunter syndrome in order to increase understanding of the genetic disorder, its progression, and its management. This is a global, collaborative effort involving a large number of people with Hunter syndrome, their families, and their doctors. Patients worldwide with Hunter syndrome are encouraged to participate, and participation is through your physician.

Shire Human Genetic Therapies worked with experts in the management of Hunter Syndrome (MPS II) to design the ongoing Hunter Outcome Survey (HOS). Scientific governance is provided by a global board of participating physicians.

The Hunter Outcome Survey is an ongoing program that tracks natural history and outcomes for people with MPS II.

The primary goals of HOS are:

- to support the gathering, analysis, reporting, and sharing of Hunter syndrome data from healthcare professionals worldwide
- monitor safety and effectiveness of enzyme replacement therapy
- to characterize and describe the MPS II population as a whole, including the variability, progression, and natural history of MPS II
- to help the MPS II medical community with development of recommendations for monitoring patients and reports on patient outcomes to help optimize patient care

It is up to you whether you would like to contribute information via your doctor to HOS. All people with Hunter syndrome are invited to participate, whether or not they are receiving treatment with ELAPRASE® (idursulfase). Participating means that information about your health and treatment will be added to the database in a way that does not identify you by name. HOS is maintained as confidential in accordance with applicable national privacy regulations and other state and local laws related to medical information.

You will get the same care and treatment from your doctor regardless of whether you participate in the program. There are no expenses to you or your doctor for participating.

How can I participate?

To participate in the HOS or learn more about it, talk to your doctor. Your doctor will ask you to complete a form which will tell you more about the HOS, the possible benefits to you, what is involved in participating, what personal information will be collected, and how this personal information will be used.

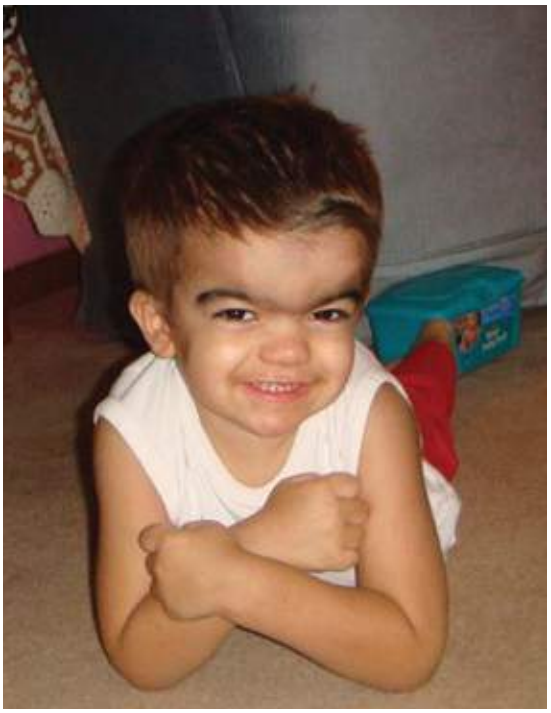
By participating in HOS, you are helping people with MPS II by adding to the information available to healthcare professionals.

You will need to give your permission to participate. Once your permission is obtained, your doctor will begin to regularly send information on your health and treatment to the HOS. However, your name will be confidential. A number

will identify you, and only your doctor will know which number is yours. If you have any questions or concerns about confidentiality, talk to your doctor.

Participating in HOS is voluntary. By participating, you are helping people with MPS II by adding to the information available to healthcare professionals. The information you provide will be grouped together with information from other people with MPS II. Once the information is put together, medical professionals can use it to learn more about MPS II, develop recommendations for care, and hopefully improve treatment for people with MPS II.

HOS provides the medical community with information about MPS II, management guidelines, and reports that are designed to help them better care for their patients with MPS II. Participating doctors can also use the database to answer specific questions they may have about MPS II or conduct research studies.



Courtesy of the Canadian MPS Society

To participate in the Hunter Outcome Survey (HOS) or learn more about it, talk to your doctor.

Anesthesia considerations

Content adapted from "Is Your Child Having an Anesthetic?" with permission from the National MPS Society.

Courtesy of Shire HGT

Overview

Various management options and surgical procedures to manage the symptoms of MPS II have been described in this resource. For many of these procedures, a person with MPS II may require an anesthetic. General anesthesia uses a medication or gas that "puts the person to sleep" before surgery. To make sure the person under anesthesia (i.e., getting an anesthetic) receives enough oxygen during the surgery, a tube is placed into the throat and connected to a machine that helps the person breathe.

Individuals with MPS II – especially those at the more rapidly progressing end of the disease spectrum – are at a particularly high risk of complications from anesthesia. One reason for this is that it can be difficult to insert the necessary breathing tube, as the airways of such individuals are often narrow as a result of the underlying disease. In addition, the spine needs to be protected during placement of the breathing tube, which can also complicate the process of inserting it. Individuals with MPS II may also take longer to recover after anesthesia.

Individuals with MPS II are at a higher risk of complications when under anesthesia.

It is important for individuals with MPS II to be properly assessed by an anesthesiologist before undergoing procedures that may require anesthesia. It is also important for such interventions to be performed at a hospital where there is knowledge of and experience with MPS II in order to reduce the risk of potentially serious complications. In addition, it is recommended that consideration be given to performing multiple procedures under a single anesthesia session in order to minimize the number of times an anesthetic needs to be given.

This section is intended to describe anesthesia and its use for individuals with MPS II in more detail, and is intended primarily for the parents of children with MPS II.

What is anesthesia?

"Anesthesia" means a loss of feeling, particularly the sensations of pain and touch. Usually, the term is used to refer to the use of medication to temporarily suppress sensations, especially before surgery.

There are 3 main types of anesthesia:

- *Local anesthesia*
  - Only the relevant area of the body is numb and experiences no pain.
  - The individual remains awake and aware of what is going on during the medical treatment.
  - No breathing assistance is required.

- *General anesthesia*
  - The entire body is numb and experiences no pain.
  - The individual remains unconscious and unable to move for the duration of the medical treatment.
  - Mechanical help is required to breathe. To assure safe oxygen levels while the individual under general anesthesia is unconscious, the airway needs to be kept open. The typical way to manage this problem is to pass a tube through the larynx (voice box) and into the trachea (airway). The tube remains in the airway during the procedure but is removed before the person fully wakes after it ends.

- *Regional anesthesia*
  - The nerve supply to a selected body area (e.g., arm or leg) is blocked to prevent the individual from feeling pain in that area.
  - The individual usually remains awake and aware of what is going on during the medical procedure, although they may receive a sedative.
  - Breathing assistance is generally not required. People who receive a sedative during a procedure will have their breathing and oxygen levels closely monitored. They may need to receive oxygen through an oxygen mask during the procedure.

Normal procedure for general anesthesia

1. A gas given by mask or a medication given by injection is used to make the patient go to sleep.
2. A muscle relaxant is introduced, which paralyzes all the muscles, including the breathing mechanism.
3. Oxygen is given by squeezing a bag linked to a face mask. This is a temporary supply until a breathing tube is inserted.
4. An instrument known as a laryngoscope is passed over the tongue and down the back of the throat so that the anesthesiologist can view the entrance to the larynx.
5. A tube (endotracheal) is put into the trachea through the larynx and the laryngoscope is removed.
6. The endotracheal tube is connected to a machine that breathes for the individual during the procedure.
7. At the end of the surgical procedure, the anesthetic is stopped. The individual should start to wake up once the anesthetic begins to wear off.
8. The endotracheal tube is usually removed before the individual is fully awake.

It is important for individuals with MPS II to be properly assessed by an anesthesiologist prior to undergoing medical interventions that may require anesthesia.

What is different for individuals with MPS II?

The impact of underlying symptoms

Anything that makes it difficult for an anesthesiologist to perform the steps required to deliver anesthesia will increase the risks associated with a medical intervention. In people with MPS II, the effect of the storage of GAGs in many soft tissues, as well as the effects on bone formation, can create such difficulties. For example:

- The storage of the GAGs narrows the nasal passages; enlarges the tonsils, adenoids, and tongue; and causes loose extra tissues and thick secretions to form around the larynx. These problems have the collective effect of severely limiting an anesthesiologist's ability

Courtesy of the Canadian MPS Society

to view the larynx in order to insert a breathing tube.
- The muscle relaxation produced by pre-medications and/or by general anesthesia leads to further obstruction of the airway.
- A stiff cervical spine (the neck region of the spine) and the possibility of an unstable spine may prevent an anesthesiologist from placing the head and neck of the person with MPS II in the best position to view the larynx. The limited jaw movement and short neck of individuals with MPS II make it even harder for even a very skilled anesthesiologist to see the larynx.

Individuals with MPS II have other symptoms that can also contribute to the increased risks associated with general anesthesia. For example:

- The storage of GAGs causes joint stiffness and thick skin, which can make it hard to give intravenous (IV) injections, especially in an emergency situation.
- The storage of GAGs in the heart and blood vessels of the heart can add to the overall burden to the cardiovascular system.

Potential risks and complications

As a result of the burden of symptoms described earlier, individuals with MPS II may experience some of the following complications while under general anesthesia:

- Airway (breathing) problems:
  - There may be difficulty in placing the breathing tube into the trachea.
  - There may be difficulty in keeping the airway open after the breathing tube has been removed.
  - The breathing tube may have to remain in place after the surgery if the initial placement was very difficult or traumatic.
  - Emergency tracheostomy (making an incision in the neck and inserting a tube directly into the trachea) may be necessary if the airway becomes compromised during intubation (putting the tube in) or extubation (taking the tube out).

- Cardiac (heart) problems:
  - Heart failure may occur.
  - Heart rhythm may become irregular.
  - There may be large changes (up or down) in blood pressure.

It is important to remember that this list does not imply that all patients with MPS II will experience these problems. It should be noted that the risk of heart-related problems is much less than that of airway problems in individuals with MPS II under general anesthesia.

What can be done to reduce the risks?

Assessing the risks prior to a procedure

If you are worried about the proposed surgery, discuss it with your primary care doctor, surgeon, or medical geneticist. He or she may suggest seeking a second opinion. The risks of general anesthesia should be weighed against the advantage to be gained from the surgery or procedure.

For many individuals with MPS II, it is possible to determine before they undergo anesthesia whether they have a significantly increased risk associated with such a procedure. Though anesthetic complications may not be entirely preventable, a pre-operative consultation with an anesthesiologist experienced in managing individuals with MPS and/or managing difficult airways could help in planning for the management of the unusual or unexpected and may help reduce the risk of anesthetic-related complications.

The risks of anesthesia should be weighed against the advantage to be gained from the surgery or procedure.

An evaluation by a pulmonologist (lung doctor) and cardiologist (heart doctor) can be helpful in determining pre-operative risks. The pulmonologist may recommend a sleep study and a pulmonary (lung) function test. He or she may also want to look at the structure of the airway using a flexible bronchoscope, which is a small tube that can be used to view the airway.

Picking and meeting the anesthesiologist

Many experts believe that for both children and adults with MPS II, it is safer to have medical interventions at a major hospital experienced with treating people with MPS. This usually entails traveling to a regional medical center or university hospital.

As there are no minor anesthetics for most individuals with MPS II, planned procedures should always involve careful assessment by a pediatric or general anesthesiologist with appropriate skills and training. In fact, for many surgical procedures it may be important to identify the anesthesiologist even *before* choosing the surgeon. Anesthesiologists should be consulted during the planning process for a surgical intervention as well as for examination just before the intervention.

The anesthesiologist is responsible for deciding the best method of anesthetizing the patient who is undergoing medical intervention. There are several aspects to this:

- Because people with MPS II present the anesthesiologist with difficult airways and often an impossible intubation using usual methods, other methods need to be used. For example, the flexible bronchoscope can be used to pass the breathing tube into the trachea at the start of the procedure. Some anesthesiologists use a laryngeal mask airway in combination with a flexible bronchoscope. While such techniques have been found to be beneficial for individuals with MPS II, they require an anesthesiologist who is skilled with these newer methods.
- For some procedures, a local anesthetic may be an option. The anesthesiologist or a member of the team may visit the individual before the procedure and prescribe the medication needed to prepare for the anesthetic.
- There are steps that can be taken to try to make the overall process safer, such as avoiding particular anesthetic drugs or stabilizing the neck of an individual who has problems with the cervical spine.

The anesthesiologist should be aware of your or your child's condition and possible problems. Inform the medical team of any disabilities or hearing or vision problems, and bring hearing aids or glasses so that it will be easier for the medical team to communicate with the individual with MPS II. It may be useful to provide the anesthesiologist with information and experience from prior procedures the individual has undergone. For example, some people are afraid of injections or hate the smell of the gas.

What does the overall process look like?

Preparation

Below are some of the steps involved in preparing the patient for general anesthesia.

Consent

The surgeon or members of the medical team will explain what is planned for the operation or procedure. The parent or individual will be asked to sign an official form of consent. The individual and/or parent should continue to ask questions until completely comfortable with what is being agreed to. If the person having anesthesia is a child and is able to understand, he or she could also be asked to sign the consent form.

"Nothing by mouth (NPO)"

The individual undergoing surgical intervention will be asked to not eat or drink anything for usually 6–8 hours before receiving the anesthetic (the abbreviation "NPO" comes from the Latin for "nothing by mouth").

Pre-medication

Pre-medication is the medication that is given to people before the general anesthetic. This can vary with the age of the individual and the type of procedure. Medication is usually given to help people relax, to dry up moisture in the mouth and throat, and to make it easier for the anesthetic to be given. Such pre-medication may be consumed by mouth (for example, via a drink) or it may be given by intravenous (IV) injection or by an intramuscular (IM) injection in the thigh or buttocks. Some individuals may be irritable until the medication has had a chance to work properly. People can also become suddenly wobbly on their feet. It is safer for them to rest on their bed or sit with a caregiver. If the individual falls asleep, it is recommended that they lie on the bed or be held in a horizontal position, as blood pressure may drop while they are standing upright. Occasionally, some of the pre-medication given to people with MPS II will have a reverse effect and make them more energetic rather than drowsy. If this has happened previously, be sure to tell the anesthesiologist about it, as well as the type of medication that produced this effect.

Courtesy of the National MPS Society

Anesthetic cream

If the anesthetic is to be given by IV injection, an anesthetic cream may be applied to the site of injection an hour before the operation. This may numb the area so that the person may not feel the needle going in. Unfortunately, the cream numbs only the skin and does not help with an IM injection given as part of the pre-medication.

In the operating room

A nurse always accompanies the individual into the operating room. For a child, a favourite teddy bear, doll, or blanket could accompany the individual too.

Sometimes parents accompany their child to the operating room and remain there until he or she is asleep; this should be discussed and agreed

with the anesthesiologist ahead of time. The parent may be asked to put on a gown and shoe coverings before going into the sterile operating area.

The anesthesiologist will explain the procedure being used. As mentioned earlier, sometimes an IV injection is used to initially put the individual to sleep, or the person is asked to breathe in an anesthetic through a mask.

Occasionally, small children can be anesthetized on their parent's knee while the gas tube is held near their face. Once the child is asleep, parents are asked to leave. It is important to go as soon as asked, as the anesthesiologist has many things to do very quickly to ensure the safety of the child being anesthetized. The nurse will estimate how long a person is likely to be in the operating room. Many parents or caregivers like to go for a walk or have a meal. If the individual is going to intensive care afterwards, you could be taken to see the ward beforehand. Many operations take longer than planned, and people usually spend a period of time in the recovery room before going back to their room or being discharged. If you are worried, you can ask the nurse to check how the individual is doing. In most children's hospitals, parents will be able to join their child once he or she has been taken to the recovery room.

Back in the recovery room

After having an anesthetic, individuals may be drowsy and unaware. For a child, hearing a familiar voice will help him or her relax and sleep more deeply. The nurse will indicate when it is safe for the person to drink something.

It may be necessary for the individual to remain intubated (with a breathing tube in place) and on a ventilator (breathing machine) for a period of time following surgery, so in the case of a young person, choosing a hospital with a pediatric intensive care unit is essential. For both children and adults, even minor procedures may require a stay in the intensive care unit so that breathing may be monitored.

Outpatient surgery (where the person returns home on the same day of the surgery) may not be suitable for severely affected individuals, even when having routine operations.

Conclusion

This section lists some of the risks and complications that are associated with general anesthesia in individuals with MPS II, as well steps to potentially reduce them. This material is intended to help individuals and caregivers better prepare for anesthesia. With that comes the hope that these individuals can safely receive medical interventions intended to improve long-term health outcomes.

Living with MPS II

Overview

The objective of this section is to provide recommendations, guidance, and support resources on a number of different topics. Many sections are most appropriate for the caregivers of individuals with MPS II, but some are also specifically included for individuals with MPS II.

Getting organized

MPS II journal

Because you will be seeing multiple doctors, it's vital to maintain a thorough record of your medical team, medical visits, medical procedures and interventions, appointments, and impressions over time. You can use the MPS II journal included in this resource to help you keep all of this information in one convenient place.

Emotional support

For parents of a child with MPS II

As a parent of a child with MPS II, you devote a great deal of time and energy to helping your child. But what happens when you need help yourself? When a child is first diagnosed, parents may have a variety of feelings, including denial, anger, fear, grief, guilt, uncertainty, and "information overload." They may also feel relieved that there is finally a diagnosis for the problems their child has been having. As time goes on, parents may also feel frustrated, isolated, stressed, and worn out.

Many services are available to help parents cope, including respite care, counselling, funding, and support groups.

If you feel this way, you are not alone. Many services are available to help parents cope, including respite care, counselling, funding, and support groups. There is no single "best choice" for everyone, as everyone's needs and personality are different. Here are a few ways to get emotional support:

- **Talk to other families affected by MPS II.** The Canadian Society for Mucopolysaccharide & Related Diseases (Canadian MPS Society) may be able to match you up with families who have volunteered to share their experiences and offer emotional support to other families affected by MPS II. The Canadian MPS Society publishes an annual Family Referral Directory, enabling affected families to contact other affected families in the same region or in the same circumstances. Some families have created websites to share their experiences with others. Visit www.mpssociety.ca for more information. You may also enjoy attending regional MPS meetings and family events or national conferences. Attending these events can help parents connect with other families affected by MPS II and get a new sense of hope from ongoing research.
- **Contact the Canadian MPS Society (1-800-667-1846) to find out more.** Many people find that talking to someone who understands may help them feel less alone and eases their fears.
- **Draw strength from your family, friends, or religious community.** Although they may not have experience with MPS II, these are the people who care about you and would like to help. Keep in touch with your "support network" on a regular basis. Think of a few specific things they could do to help you, such as just coming over to talk, running some errands for you, babysitting, or bringing over some food. Getting a break, even on the small things, might really help.
- **Be good to yourself.** Although your role as a caregiver is very important, try to keep your own needs in mind too. Arrange for respite care or other help so that you are able to take some time for yourself on a regular basis. Continuing to do the activities you enjoy can help you hold onto your sense of self.

- **Take a break.** Caring for any child is hard work. Parents need a break to rest and enjoy activities, and this may not be possible when their child with MPS II is with them. Brothers and sisters also need their share of attention, and need to be taken on outings that may not be feasible for a child with rapidly progressing MPS II. Many parents use some form of respite care or have someone come in regularly to help at busy times. Slowly progressing individuals may need help to become more independent from their families and may benefit from a vacation, perhaps with others who have disabilities.

Talking with your family

Your immediate family

For parents of a child with MPS II

Receiving a diagnosis of MPS II can be a life-altering event for families. After the initial feelings of shock, despair, sorrow, hopelessness, and anger somewhat subside, parents generally regain their footing and consider how to deal with this new aspect of their lives. They realize that though they cannot control having to accept the diagnosis of MPS II, they do control how the diagnosis affects their child and the rest of the family. Please consider the following suggestions as you create your family's plan for coping with MPS II.

Though parents cannot control having to accept the diagnosis of MPS II, they do control how the diagnosis affects their child and the rest of the family.

It's not always easy to ask for help. But help is out there – try one of these forms of emotional support if you are feeling overwhelmed.

For adults with MPS II

As an adult with MPS II, you are faced with another set of challenges. MPS II is a family affair in many ways. While you may be the one with the disease, it ultimately affects everyone in the family. Your spouse may need to pick up more of the household responsibilities. It is important to talk to one another and learn how to communicate about how these changes make you feel. Children might not understand why you tire so easily. Children can also think that you are dying or that they did something to cause you to get sick. Talk openly with your children about your disease, how it will change your family, and what they can do to help.

Talk about MPS II with both your affected child and their unaffected siblings. Parents of children with MPS II may have a difficult time deciding how much information to give their affected and unaffected children about the disease. Although there may be a tendency to conceal information from children to avoid causing unnecessary anxiety, it is often best to be as open and honest as possible. Keep in mind that your child with MPS II and his or her brothers and sisters can be very perceptive. They will likely know if their parents are not being completely honest with them, and they may develop feelings of confusion and mistrust. Age-appropriate information can be delivered in small doses and should be geared toward the children's level of understanding. Parents should make sure their affected child and his or her brothers and sisters know that the parents are available to answer any questions that may result from these discussions. Answers to questions should be honest and straightforward, yet age-appropriate.

Help your children deal with their feelings about the disease. Parents of children with MPS II are faced with the difficult task

Courtesy of Shire HGT

of helping both their affected and unaffected children deal with the wide array of emotions associated with living with MPS II. Providing support by listening and discussing these feelings is essential. Younger children may believe that MPS II is a punishment for something they have done. Older children may resist discussing their concerns or feelings in order to protect their parents from becoming upset. It is important to reassure affected children and their siblings that they did not cause MPS II. They must also know that they can talk to their parents about any concerns or feelings without fear of being judged negatively or causing parents to become overly upset. Remember that children's thoughts and feelings about MPS II and their impact may change over time. As such, keeping the lines of communication open should be an ongoing task.

Prepare your children for medical procedures. Children need to know what to expect in their lives. Although parents may think they are protecting their child with MPS II by withholding information about procedures that may be painful or uncomfortable, this approach may increase anxiety. It is usually a good idea for parents to take the time to prepare both the affected child and their brothers and sisters for upcoming procedures. Parents should explain why the procedure is being done, who will be performing the procedure, what equipment will be used, whether or not there will be pain or discomfort, and what type of recovery period to expect.

While most pediatric hospitals have staff experienced in helping to prepare children for hospitalization, surgery, and various medical procedures, the responsibility of preparing the siblings often falls on the parents. Siblings may experience anxiety related to parents spending increased time away from home, getting less attention from parents, fear of outcome of the procedure being performed, and missing planned events in their own lives due to parental time and resource constraints. Keeping the child's brothers and sisters informed makes them feel important and involved, can minimize resentment, and can help facilitate healthy parental and sibling relationships. Keep in mind that the information provided does need to be geared toward the age level of the children, and that the children should be encouraged to ask questions until they understand.

Keeping the child's brothers and sisters informed makes them feel important and involved, can minimize resentment, and can help facilitate healthy parental and sibling relationships.

Give your children some choices. At times, children with MPS II and their siblings may feel they have little control over their lives. Therefore, it is important for parents to help foster a greater sense of control. This can be accomplished by offering children choices whenever possible. When it is appropriate, children with MPS II may enjoy being given the choice as to which arm the IV will go into, what they will eat, or when they will do homework or play. Likewise, siblings may enjoy having a choice over which care-giving tasks they will perform and the timing of these tasks.

Help your children lead as normal a life as possible. Parents should try as much as possible to treat their child with MPS II – and his or her siblings – like any other child. While recognizing that children with MPS II do have special needs, it is important that parents encourage them and their siblings to participate in activities that involve other children of the same age. The parents should make sure siblings and playmates understand what types of activities are considered appropriate for the child with MPS II.

It is important to consider the child's developmental age when dealing with difficult behaviour.

Children with MPS II without significant developmental delays need discipline from parents according to their ability. When the parents maintain structure and consistency, it helps the children to feel safe and secure. Adequate discipline also helps children learn to control their own behaviour. When possible, parents should make sure that discipline is consistent – no matter what the day or time, who is disciplining (parents and caregivers), or who is being disciplined. Consistent discipline among children with MPS II and their brothers and sisters in the household helps to foster healthy sibling relationships. Recommended discipline techniques include praising appropriate behaviour, using time-outs for young children, and restricting privileges of older children for inappropriate behaviour. However, it is important to keep in mind that strategies for dealing with difficult behaviour must be tailored to the developmental age of the child. The discipline suggestions above may not work for children with severe developmental delays, and not all children with MPS II can be disciplined. In this case, you many need to explore other options for managing your child's behaviour, such as avoiding over-stimulation, encouraging physical activity, setting aside an area of your home where the child can play safely without continuous supervision, and arranging for respite care (a caregiver who can come to our home to give you a break).

It is important to consider the child's developmental age when dealing with difficult behavior.

Courtesy of Shire HGT

Give your children responsibilities. Just as children need discipline, they also need to be given responsibilities. Parents must use judgment in assigning tasks to affected children that can be carried out with success, and must also consider the developmental age of the child when deciding whether to give the child responsibilities. The requirements for the tasks should be clear and consistent. Parents should also remember to acknowledge and offer praise for tasks that have been done well. Siblings often enjoy helping with the care of their sibling with MPS II, although this should not be an expectation.

Develop and maintain family routines as much as possible. Children typically prefer daily routines that are predictable and consistent. Although this is not always possible in an MPS II family, an effort should be made to maintain regular routines and schedules for all family members.

Be mindful of what your children can overhear. Parents should be mindful about what is said within earshot of their affected and unaffected children. It is important that children receive consistent and age-appropriate information from their parents. They also should not be exposed to conflicts related to MPS II and its management that may take place in the home or medical environment. This can lead to feelings of insecurity and mistrust.

Prepare your children for the reactions of others. Children with MPS II and their siblings often don't know how or what to tell others about the disease. Parents can help their children by suggesting various age-appropriate explanations of what MPS II is and how it affects people. The issue of how to handle any teasing should also be discussed with children. Role-playing may be useful in helping children to craft responses to questions or teasing that may come from those unfamiliar with MPS II.

Courtesy of the Canadian MPS Society

Talking about MPS II can help both you and your family.

Your extended family

For parents of a child with MPS II

Speaking with your extended family

Talking about MPS II can help both you and your family. Because MPS II is genetic, your other family members may be carriers. Making them aware that MPS II may run in your family will give them the chance to have genetic testing. Telling your family can help them understand what you are going through. The more they know about MPS II, the more likely they will be able to help. For more information on genetic testing, see "How do people inherit MPS II?" in the "Mucopolysaccharidosis II (MPS II) disease" section.

Getting ready to talk to your extended family

Telling your family about MPS II isn't easy. It may help to plan a few things in advance. First, decide when and where to tell your family. Make an outline of things you would like to say. Think of what reactions they may have and how you may deal with them. The information below will help you with each of these steps.

When is the best time to tell them?

Give yourself some time after the diagnosis to take in the new information and deal with it within your immediate family first. Then you can begin telling other relatives. There is no "best time," but try to choose a time when you can meet face-to-face if possible, and a time when they are not stressed about issues of their own.

What should you tell them?

There are no hard-and-fast rules for what to say to your family. Before talking to them, develop an outline of what you would like to say. This will help you decide which points to focus on and which things you would like to keep private. It's OK if you are uncomfortable talking about certain things and would rather not discuss them.

Here are some points that you may want to consider including in your outline:

- What is MPS II? Explain that there is a variable range of disease progression in MPS II, and not all affected individuals have all the medical problems seen in MPS II.
- When did the symptoms start? How did they affect your life?
- Where did you go for help and how did you finally get a diagnosis?
- How is your or your child's health now?
- What can be done to manage MPS II? What management options have you and your doctor decided to use?
- How are you and your family keeping a positive attitude?
- Where can relatives find out more information on MPS II?
- How can your family help?
- What does this mean to your relatives? Should they be tested for MPS II?

This list is just a guide to help you put together your outline. You do not have to talk about any of these topics if you are not comfortable. The answers to these questions depend on your individual situation. You can turn to your doctor, this learning guide, or the Canadian MPS Society for more information.

Possible reactions and tips for dealing with them

It's impossible to predict the reactions that your family will have to your news. However, there are a few common reactions that people may have:

- They may want to know how they can help you. To be prepared for this, think in advance about a few things they could do to help (such as house-sitting or running the occasional errand).
- They may want to learn more about MPS II. You may wish to plan ahead by making a list of a few resources (see "Source of support and information," later in this section) and bringing information materials with you.
- They may be concerned about whether they or their children are at risk of MPS II. To help, you may want to explain how MPS II is inherited (see "How do people inherit MPS II?" in the "Mucopolysaccharidosis II (MPS II) disease" section).

Moving forward as a family

Telling your family about MPS II can be difficult, but it may help both you and your family. Your family may want to learn more about MPS II so that they can understand what is happening and to help. If other people in the family have undiagnosed MPS II, they may finally have an explanation for their symptoms. Family members may choose to be tested to see if they are carriers for MPS II. These things will all help you to cope with the diagnosis of MPS II as a family.

Talking to doctors

Why it's important to talk to your doctor

Getting involved in your health care means having a good partnership with your doctor. This section suggests some steps you may take to build this partnership and increase your understanding of MPS II.

MPS II can cause many challenges, including difficulties with mobility and speech. Parents of a child with MPS II may feel helpless and uncertain about their child's future. Older individuals with MPS II may feel frustrated when unable to participate in certain activities or unable to make certain lifestyle choices. You may want to consider becoming an active participant in your care and working with your doctor to make the choices that are right for you or your child.

Finding the right doctor

It's important to choose a doctor that you (or you and your child) feel comfortable with. Consider the qualities that are important to you. Make a list of those qualities. For example, do you prefer a doctor who uses clinical language or one who speaks in layperson's (everyday) terms? MPS II is a rare condition, so many doctors may not have extensive experience with the condition. Finding a doctor who is knowledgeable about MPS or is willing to work through the issues with you is very important.

Once you've thought these things through, it's time to look for a doctor in your area who meets your criteria. You may want to meet with several doctors before making a final decision on one that you feel communicates in a manner appropriate for you and understands your needs and concerns.

Preparing for your visit to the doctor

Plan ahead

You may find it helpful to plan ahead for your visit to the doctor. Consider preparing a list of questions and concerns before every visit. Your list might include your goals for the visit, any new information about your or your child's condition, such as new symptoms, and any questions or concerns you may have.

For your first visit to a new doctor, you might also want to include:

- all symptoms, when they began, how often they occur, and how they have changed over time
- any medications taken
- your or your child's medical history, including a list of your other doctors and previous medical procedures
- any problems with daily activities

MPS II is a lifelong condition. Staying well informed can help you play a role in your or your child's health. Your doctor may be a source of information on MPS II and current management options. During the visit, write down the information your doctor gives you so that you'll have a record of it, and ask for a letter from the doctor summarizing the visit.

Ask your doctor for any brochures or videos about the condition. If your doctor makes any recommendations for treatment or therapy, be sure to write them down or ask your doctor to write down the instructions for you.

Keep up to date

To stay informed in between your visits to the doctor, you can find more information by reading books on MPS II, searching websites, or attending support groups. For a list of relevant websites, see "Websites" at the end of this section. If you think of a question you'd like to ask or information to share on your next visit, keep a running list so it will be available when you're preparing for the next visit.

When in doubt, ask

Don't be embarrassed to ask your doctor questions if you need more information or if there's anything you don't understand. If something doesn't make sense, ask your doctor to explain it again differently and to define any new words. You may want to try repeating what your doctor has told you in your own words so you can be sure you've understood.

Getting involved in treatment means having a good partnership with your doctor.

Keep an open line of communication

Open and honest communication will help create a partnership with your doctor. Some symptoms or problems may be hard to talk about, or it may be difficult to admit that you have not understood something. However, knowing about these things helps your doctor give you better care. You'll both benefit in the long run: your doctor will understand your needs, and you will gain more control over your care.

Know your management options

It's important to understand the options that are available. Here are a few questions to ask your doctor about managing MPS II:

- What management options are available?
- Of these, which may be appropriate for me (or my child)?
- How and when is the intervention usually given (for example, is it a one-time procedure, or are repeated interventions required on a regular basis)?
- What are the side effects?
- How will it interact with my medications?
- How much will it cost?
- How long until it starts to work?
- For symptoms that cannot be treated, how else might I be able to manage them?

The doctor is your partner in managing MPS II. Good communication will help you get the most out of your visits to the doctor.

Talking to employers

For parents of children with MPS II

The information in this section is meant to be informative only and does not constitute legal advice. Individual situations should be discussed with a legal professional to determine the extent to which these laws may apply to you.

If your child has MPS II, you may find yourself struggling to meet the demands of the workplace while still giving your child the care and attention they need. You may need time off to take your child for medical appointments. You may also need to send them to a special daycare or school. Fortunately, there are programs to help working parents cope.

You may be able to take a leave of absence in order to care for an individual with MPS II under one of various family/sick/personal emergency leaves set out in the provincial/territorial and federal employment standards legislation in Canada.

Compassionate care leave may also be available at the provincial/territorial or federal level. This leave is generally intended for an employee who needs to care for a person with a serious medical condition who has a significant risk of death occurring within a period of 26 weeks. To review the legislation that applies to your particular area in more detail, please refer to the appropriate website listed below:

- **Alberta:**
  employment.alberta.ca/documents/WRR/WRR-ES-FI_esfs28.pdf
- **British Columbia:**
  www.labour.gov.bc.ca/esb/facshts/ccl.htm
- **Manitoba:**
  www.gov.mb.ca/labour/standards/doc,compassionate-leave,factsheet.html
- **New Brunswick:**
  www.gnb.ca/0308/FactSheets/17.pdf
- **Newfoundland and Labrador:**
  www.hrle.gov.nl.ca/lra/labourstandards/faq.htm#Compassionate_Care_Leave
- **Nova Scotia:**
  www.gov.ns.ca/lwd/employmentrights/compassionatecarefaq.asp
- **Nunavut:**
  www.gov.nu.ca/hr/site/HR_Manual/1300_Leave/1317-Compassionate_Care_Leave/1317%20-%20Compassionate%20Care%20Leave%20-%20March%2031%202009.pdf
- **Northwest Territories:**
  www.mross.com/law/Publications/In+The+News?contentId=1286
- **Ontario:**
  www.labour.gov.on.ca/english/es/pubs/guide/family.php
- **Prince Edward Island:**
  www.gov.pe.ca/cca/index.php3?number=1022358&lang=E

- **Quebec:**
  www.servicecanada.gc.ca/eng/lp/spila/clli/eslc/compass.pdf (see Quebec section)

- **Saskatchewan:**
  www.gov.sk.ca/news?newsId=abbdb324-3988-4044-ab59-bfeb34e1de3a

- **Yukon:**
  www.community.gov.yk.ca/labour/compare.html

Some companies may have employee assistance programs that can connect employees to a qualified professional who can help parents deal with the stress of having a child who is ill, and can direct them to other services that can help, such as respite care. The Canadian MPS Society can connect you to other families who have experienced similar problems and who can provide helpful information.

Other companies may offer daycare facilities, but these may not be equipped to assist children with special health needs. Check with your employer to see whether they offer any daycare services and, if so, whether they could accommodate your child.

For individuals with MPS II

Choosing a career

Please note that this section is most applicable to individuals with attenuated MPS II.

Many individuals with MPS II may do well at a variety of different jobs. Begin your search for the right job by assessing your capabilities. Try to be realistic about what you can and cannot do. Ask yourself these questions:

- How much standing, walking, and sitting are you comfortable doing?
- How much lifting and reaching can you do?
- What is your fatigue level?
- Can you hold objects or open car doors easily?

Consider your abilities on both "good" and "bad" days in making these judgments.

If the job you are interested in requires you to do activities you find difficult, you have two choices:

Courtesy of Shire HGT

try to find ways to do the job comfortably, or make the decision to find a job that is better suited to you.

For most people – with or without MPS II – choosing a career is not easy. It is usually a complex process that involves matching your skills and desires with the realities of the business world.

You know your own abilities and limitations. It is important to be practical – but not unrealistic – about what you can and cannot do. Instead of using your limitations as a restriction, use them as a guide to finding the right career.

If you are interested in an occupation but feel it is not suited to you, consider other opportunities

Courtesy of Keith Beaty

in the same field. For example, if you've always wanted to be a nurse but feel you don't have the strength it requires, other careers in the allied health fields such as nutrition or health education might give you as much satisfaction.

A career counsellor can help you explore a type of work that you might enjoy and that is well suited to your individual strengths and interests.

A career counsellor can help you explore a type of work that you might enjoy and that is well suited to your individual strengths and interests.

Talking to your employer about MPS II

The information in this section is meant to be informative only and does not constitute legal advice. Individual situations should be discussed with a legal professional to determine the extent to which these laws may apply to you.

You may be concerned that having MPS II may raise questions in an employer's mind about your ability to do your job. You may be reluctant to talk about your condition for fear you will not be hired or considered for a promotion. You should know that Section 15 of the Canadian Charter of Rights and Freedom guarantees equality rights plus freedom from discrimination for people who have a physical or mental disability. This charter applies to the Parliament of the Government of Canada and the legislature and government of each of the provinces only. For private business and organizations, federal and provincial/territorial human rights legislation protects an individual from discrimination based on certain prohibited grounds, including disability. The Employment Equity Act (EEA) of 1995 ensures that persons with disabilities are granted full and equal access to employment and opportunity. An employer must accommodate the disabilities of employees, prospective employees, and clients or customers.

In order to avoid particular actions in this regard, the employer must prove that accommodation of the needs of an individual or a particular class of individuals would impose undue hardship on the organization or on the people who would have to accommodate those needs, considering factors such as health, safety, and cost. In addition, the employer may show that a legitimate occupational requirement prevents them from hiring or promoting an employee with a particular disability or from accommodating this employee.

Protection from discrimination occurring at the private employment level would be provided by provincial or territorial legislation. To learn more about disability-related policies in Canada, see www.disabilitypolicy.ca/index_english.php. If, when, or how you decide to talk with your employer about your condition is a personal decision.

You may find that you need special accommodations to do your job. If so, your employer needs to know about your limitations or restrictions in order to make the necessary accommodations. Open communication is the key to a healthy work relationship. You should also know that accommodation is a two-way street. The employee must work with the employer in developing means for accommodating a disability.

You should ask your employer whether you are entitled to sick leave or disability benefits before applying for EI sickness benefits. In the case that your employer does not offer such

benefits and you are unable to work for a period of time because of illness, you may be entitled to EI sickness benefits. To learn more, visit www.servicecanada.gc.ca/eng/ei/types/special.shtml#Sickness3.

Working successfully

Working successfully depends greatly on having proper management of your disease. This includes care from an experienced doctor, working with your healthcare team to plan a treatment program of proper medication and exercise, and self-management skills.

Be flexible and creative. Flexibility can help you balance work responsibilities with the demands of MPS II. By figuring out your energy patterns during the day and what kind of activities hurt or help you, you can arrange your work schedule accordingly.

Create an efficient work environment. Arrange your area to limit the amount of lifting, reaching, carrying, holding, or walking necessary.

Talking to educators

Educators can play a critical role in your development and the development of your child. They need to provide you with accommodations, but first need to understand the special issues that could affect your educational needs. Please see "Educational strategies" and "An overview of MPS II for teachers" for information on this very important issue.

Sources of support and information

Resources from the Canadian MPS Society

Founded in 1984, the Canadian MPS Society is committed to providing support to individuals and families affected with MPS and related diseases, educating medical professionals and the general public about MPS, and raising funds for research so that one day there will be cures for all types of MPS and related diseases.

The Society supports individuals and families affected with MPS from their diagnoses through all stages of their journeys. Information is available through our website (www.mpssociety.ca), our 1-800 number (1-800-667-1846), and our booklets, including our Lysosomal Storage Disease Booklet, which provides general information on LSDs, and our disease-specific booklets, which contain valuable information on disease symptoms, management, and care.

Courtesy of the Canadian MPS Society

Together with the National MPS Society (USA), we published a booklet on "Daily Living with MPS & Related Diseases," which provides helpful tips for parents by parents, and our annually published Family Referral Directory helps families affected with the same syndrome or living in the same region of the country get in touch with each other. Our quarterly newsletter, *the Connection*, is a valuable resource that helps members stay on top of MPS-related news and events and stay in touch with each other. The Society has been actively involved in providing advocacy support to ensure our members receive the treatment and care they need. It also provides bereavement support to those dealing with the devastating loss of a child or family member to MPS.

In addition to informational and emotional support, the Society offers a Family Assistance Program to our member families, which provides financial aid to help with the costs of living with a chronic health condition. We also provide travel stipends to member families to attend regional

family meetings, national family conferences, and international MPS symposia.

The Society holds regional meetings as well as biennial family conferences, providing families an opportunity to learn more about new research, treatments, and care strategies, and to meet with other families, share experiences, and form lifelong friendships. For some people, these meetings and conferences present their first opportunity to meet others who are affected with or by the same disease. We were thrilled to host almost 1,000 delegates from 37 countries at the 10th International Symposium on MPS & Related Diseases in Vancouver in June 2008. All family sessions from that world-class conference are available for download on our website, and all scientific sessions are available for viewing on the website of Global Organisation for Lysosomal Disorders (GOLD). Please visit our website for directions on how to access these presentations.

Due to increased fundraising over the past several years, the Society has been able to fund excellent research into MPS, providing affected families with hope for a brighter future. Information on our funded research is available on our website. We have also begun collaborating actively with international MPS patient organizations through the International MPS Network, and with various Canadian rare disease patient organizations through Canadian Organisation for Rare Disorders (CORD). Together, we are stronger.

Living with a chronic, progressive illness can cause emotional and physical stress, as the demands can be overwhelming. To find out more about how we can help, please visit www.mpssociety.ca or call our office at 1-800-667-1846 with any questions about the resources, programs, and services we offer for individuals and families affected by MPS II.

Resources from Shire HGT

Overview of resources at Shire HGT

Many individuals and their families have placed calls to Shire HGT for a variety of reasons. The Shire Medical Information group can provide information about MPS II, management options, or to check the status of clinical trials. To reach Shire HGT medical information, call Paladin Labs Inc. at 1-888-550-6060.

The Canadian MPS Society provides support for individuals and families living with MPS II.

Reimbursement Support Program

Shire HGT (Canada) Reimbursement Support program is a private, confidential, and free patient support service for patients who have been prescribed ELAPRASE® (idursulfase) and have access to private health insurance coverage.

The reimbursement support program assigns you to a personal case worker who can help you determine your drug plan coverage, complete paperwork required by your insurance company, and research funding assistance and copayment options.

If you have any questions or wish to enrol in the Reimbursement Support Program, call our toll-free support line at 1-866-773-6302. You will reach a reimbursement specialist, who can help you deal confidentially with insurance companies or brokers. Assistance is provided Monday through Friday 8:00 am to 8:00 pm, eastern time.

Shire HGT Medical Information

Shire HGT Medical Information offers access to healthcare professionals who can answer a wide range of questions related to MPS II disease and their products.

In addition, Shire HGT Medical Information professionals can help healthcare professionals treating MPS II patients by supplying them with medical literature, information from recent medical meetings, and other doctor-oriented resources as requested.

Courtesy of Shire HGT

To reach Shire HGT medical information call, Paladin Labs Inc. at 1-888-550-6060.

Hunter outcome survey (HOS)

Disease outcome surveys are programs that study people with a particular disease to learn more about it. Shire Human Genetic Therapies is working with experts in the management of MPS II (Hunter syndrome) to enrol individuals in the Hunter Outcome Survey (HOS).

The HOS program is designed to support the gathering, analysis, reporting, and sharing of data about MPS II from individuals and healthcare professionals around the world. All people with MPS II are encouraged to participate, as this will help provide information and insight about the progression of MPS II, disease management, and medical outcomes.

Resources for adaptive living

There are many resources that may help you overcome physical limitations due to MPS II. Seek advice and recommendations from professionals such as your doctor, nurse, physical therapist, occupational therapist, or other healthcare provider.

An occupational therapist is trained to make a complete evaluation of the impact of MPS II has on your home and work activities. The therapist will review your situation by conducting an interview, asking questions about hygiene, grooming, eating, drinking, dressing, getting in and out of bed, driving, cleaning, cooking, shopping, and working. The therapist will also conduct a physical examination. After a thorough assessment, the therapist will develop a plan, which may include physical therapy and adaptive equipment.

You may also benefit from using adaptive equipment. These tools and devices help you perform your daily tasks, such as getting dressed and eating, more easily. They also help you maintain a more active lifestyle.

Some adaptive tools that might be helpful for MPS II include:

- **thick grips or handles:** These handles can make it easier for you to grasp small objects. They can be used on a variety of objects including utensils, toothbrushes, hairbrushes, pens and pencils, or combs.
- **extended handles and other tools to give you extra leverage:** These tools help you with tasks like opening jars, turning doorknobs, or twisting dials on your stove.
- **graspers and reachers:** These aids can help you pick up objects without bending or reaching. Extended shoehorns called donners can be used to help you pull up your socks.
- **slip-on shoes or Velcro closures:** These shoe styles are easier to take on and off than the lace-up kind.
- **zipper pullers and button grippers:** These devices can help you get dressed by making it easier to pull your zippers and fasten buttons. Sewing on buttons with elastic thread makes them easier to use and less likely

to break off while you are fastening them. You can also opt for clothing with an elastic waist and Velcro tabs to make dressing less painful or difficult.

Websites

The Canadian MPS Society, www.mpssociety.ca, has a listing of websites that include general information, general health, government agencies, advocacy and education, support, and helpful products.

Resources that can help you:

- www.mpsforum.com (owned and operated by a family affected by MPS)
- www.raredisorders.ca (Canadian Organization for Rare Disorders)
- www.lhsc.on.ca/programs/medgenet (Canadian Directory of Genetic Support Groups)
- www.goldinfo.org (Global Organization for Lysosomal Diseases)
- www.wsfc.ca/family.html (Western Society for Children with Birth Disorders)
- www.siblingsupport.org/ (Sibling Support Project)
- www.cacl.ca (Canadian Association for Community Living)
- www.neads.ca (National Education Association of Disabled Students)
- www.starlight.org (Starlight Children's Foundation
- www.pwd-online.ca (Persons with Disabilities online – Government of Canada)
- www.inclusiveeducation.ca/english/index.asp (Inclusive Education Canada)
- www.ldac-taac.ca (Learning Disabilities Association of Canada)
- www.cea-ace.ca (Canadian Education Association)
- www.vracanada.com (Vocational Rehabilitation Association of Canada)
- www.rhdcc-hrsdc.gc.ca (Human Resources and Skills Development Canada)
- www.geneticalliance.org (Alliance of Genetic Support Groups)
- www.climb.org.uk (Children Living with Inherited Metabolic Diseases)
- www.lsdsonline.com (Lysosomal Storage Disorders Support)
- www.lysosomallearning.com (Lysosomal Learning)
- www.abilities.ca/agc/disability_organization/view_disability_organizations.php?pid=&cid=&show=all&subid=169 (Abilities.ca guide to disability organizations)
- www.respitelocator.org (National Respite Locator Service)
- www.ablelink.org/public/new/index.html (Ability Online support network)
- www.cra-arc.gc.ca/tx/ndvdls/tpcs/rdsp-reei/menu-eng.html (Canada Revenue Agency: Registered Disability Savings Plan)
- www.cra-arc.gc.ca/cdb/ (Canada Revenue Agency: Child Disability Benefit)
- www.lysosomalstorageresearch.ca (The Lysosomal Storage Research Group)

Educational strategies

For parents of children with MPS II

Courtesy of Shire HGT

Introduction

Both parents and educators want the best for their children. Understanding how to work together with the education system can help parents of children with MPS II ensure that their child has the best possible education.

This section is designed to provide a source of information for parents and educators. It is not meant to be an exhaustive resource, but it attempts to pull together some information on strategies and resources that can be used to help devise an appropriate educational program for children with MPS II. There has been very little research on the educational aspects of MPS II. This section brings together the experiences of parents and educators who have developed useful techniques and successful educational programs (programs that provide the child with the best possible education) and have addressed difficulties commonly experienced by children with MPS II.

Understanding how to work together with the education system can help parents of children with MPS II ensure that their child has the best possible education.

How schools work

Understanding how schools are organized can help parents work together with educators. Parents of children with MPS II must often ask school personnel to do something different from the norm. Give them time to adjust to your requests. See how you can support them as they adjust to the new expectations you are requesting. Compliment them on what they do well.

Schools as organizations

All organizations have a particular way they operate. Schools have a division of labour based on specialization. Education is organized to meet the needs of the group, and education programs are conducted by regular or general education teachers. Special education was developed for those students who do not do well in general education classes. General education teachers have been told for years to refer children to special education when their educational needs cannot be met in a general classroom.

The classroom teacher and others cannot make school operational changes without getting approval from their superiors. Change generally occurs from the top down, meaning that school policy changes usually result from directives from central office administrative staff. This limits how easily changes can occur in normal school operations, and teachers may not be free to independently make necessary changes in the material they teach and how they teach it. Difficulties developing an Individual Educational

Program (IEP) often result from problems with school policies or from getting permission from superiors to make changes.

A teacher's life

Your child's teacher is there to help. Like you, their goal is to provide the best possible education for your child. By working together, parents and teachers can provide valuable resources for each other. Parents can share information with teachers regarding MPS II (you may wish to give your child's teacher a copy of "An overview of MPS II for teachers," available in the "Information Handouts" section of this binder), their child's strengths and preferences, and their goals for their child's education. Teachers can talk to the parents about the child's experiences at school and help the child meet his or her educational goals. Having a strong relationship with your child's teacher can help your child benefit as much as possible from education.

Parents of children with MPS II must often ask school personnel to do something different from the norm.

The role of parents

Parents may be in a position of asking teachers to do things that differ from the norm of the school. Without approval from their supervisors, teachers may not be able to make changes that both parents and teachers are recommending. Teachers' superiors may not always be knowledgeable about special needs children. The job of parents is to understand and then help the school system plan for developing an individualized education plan (IEP).

Relevant laws

The information in this section is meant to be informative only and does not constitute legal advice. Individual situations should be discussed with a legal professional to determine the extent to which these laws may apply to a particular child.

Canadian federal and provincial/territorial human rights laws specify a "right to reasonable accommodation for a disability subject to the employer's right to demonstrate that such accommodation would cause the employer undue hardship or that the discriminatory policy or practice is a legitimate occupational requirement." Educational institutions have a duty not to discriminate directly against a student or other individual on the basis of his/her disability as well as not to engage in conduct or have policies that result in an adverse effect of discrimination against individuals with disabilities. The duty to accommodate is in addition to this prohibition on discrimination. However, like an employer, an educational institution is not required to accommodate an individual's disability if doing so would cause undue hardship to the institution.

Planning for educational programs and supports

Educational needs: the big picture

Parents can help their school plan for their child's education by providing resources (such as "A teacher's guide to MPS II," available in the "Information Handouts" section of this binder) to help school personnel become more familiar with MPS II. This section covers some of the "big picture" issues schools should consider when developing an individual educational program (IEP) for a child with MPS II. It is difficult to write specific guidance that covers every child with MPS II, as the spectrum of severity varies widely between rapidly and slowly progressing forms of the disease. Generally, children are entitled to free and appropriate public education, regardless of any disabilities they may have, and you and your school can work together to help your child have a positive educational experience.

Overall planning and monitoring considerations

Schools should monitor changes in the needs of children with MPS II so the IEP can be adapted as needed to support a child whose physical limitations are affecting learning. The IEP should be developed to encourage social and academic

participation and development. Teachers may need additional support to understand the child's physical capabilities and to cope with any limitations due to the child's disease progression or missed school. This will assist them in tailoring their teaching and expectations to the child. Planning and goal development may be difficult for school personnel who do not have experience with children whose disorders are progressive. The traditional focus is on improvement in skills and lessened support as improvement occurs rather than maintaining skills.

For children with MPS II, it may be more suitable to aim for success toward individual goals that are appropriate to their level of development, rather than improvement of skills.

Medical care needs

Mobility problems, hearing loss, and vision difficulties may need the special attention of school personnel in program planning. All schools should have teachers who specialize in working with children with vision or hearing impairments. These teachers help the IEP team develop alterations to deal with these problems. Mobility problems and physical limitations caused by the disorder can be addressed by consulting physical and occupational therapists and adaptive physical educators. Class assignments and projects (such as art projects) can be modified to allow children with physical limitations to participate in similar projects with their peers.

Behaviour problems

Some of the physical effects of MPS II, such as poor hearing and vision, may lead to difficulty hearing or understanding a teacher's questions or instructions, difficulty interacting with peers, trouble participating in group activities, or failure to complete assigned work. For severely affected children, behaviour problems may occur due to lack of understanding, difficulty with communication, or sensory limitations. It is important for school personnel to understand MPS II so that they recognize these issues as complications of the medical condition. The learning environment should be supported with an emphasis on modifying the classroom environment and using reinforcements.

Courtesy of Shire HGT

Teacher education and support

Teachers should be educated about the child's disorder, abilities, and special needs. Teachers may be unsure of their ability to teach children with MPS II, but many skills they use in teaching non-disabled students will enable them to work well with children with MPS II. Teachers and classmates may also need support in dealing with feelings of loss if the child's condition worsens.

Academic and career expectations

Teachers need appropriate expectations for learning that are balanced by an awareness of the child's physical and mental limitations. It's important for teachers to be aware that the outward physical manifestations of the disorder do not mean that the child has a delay in intellectual development. While some people with MPS II may have severe developmental delays and mental impairment, others may have normal

or near-normal intelligence. Appropriately high expectations of academic achievement (adjusted to the individual's mental capabilities) will foster realistic self-appraisal and enhanced academic achievement. Academic and vocational programming for the child should foster independence, and career goals should be set realistically high. Planning for the transition from school to post-secondary education or work should focus on helping children with MPS II pursue vocations in a manner similar to that of their peers.

Socialization

School attendance and socialization should be encouraged and fostered through integrating the child into the classroom and through interventions for specific social skills. Independence should be supported. Teachers can do much to improve the acceptance of the child through instructional activities such as cooperative learning and encouraging support for all children in the classroom. Additional support and education is necessary during adolescence.

Special services

Overview

Provincial education laws require that school boards make education available to all school-aged persons in a district. In addition, there are individual provincial/territorial statutes with requirements specific to that province/territory.

One of the most vexing issues in devising an educational plan for children is deciding how any needed special education services will be delivered.

It should first be determined what types of adaptive equipment and services would enable the child to be educated in the regular classroom. This list could include devices to assist in writing and hearing, special desks or seats, or extra time to finish computer and writing projects. An occupational therapist should be included in determining what tools will help integrate your child into a normal classroom setting. See "Assistive technology," later in this section, for more information and adaptive resources that support the school setting.

Dr. Darlene Perner has written a book designed to help teachers carry out instructional strategies that aid in inclusion of students with disabilities and special needs. The book is accessible online at www.inclusiveeducation.ca/documents/Changing%20Teaching%20Practices%20Perner%20136583e.pdf.

Courtesy of Shire HGT

Socialization

In determining the proper placement, parents should consider their child's socialization needs. To aid in this, they should do the following:

- Ensure the IEP team understands that all children benefit from social interaction at some level.
- Decide on overall socialization needs and goals for the child. Remember that for some children, increasing the number of friends and playmates is the goal, while for others it may be reducing interaction to a tolerable level.
- Form a social network in an inclusive setting. Meet with small groups of classmates and have them develop a list of ideas on how to involve the special needs student in the school. Students who volunteer to get involved with these activities can form a peer network. Although the peers will first consider themselves to be advocates, this role may evolve into a friendship between the special needs student and other students.
- Match your child with another child according to their preference for certain activities that improve the interaction between your child and others. These activities may take place in the school cafeteria, library, computer lab, or gym.

Behaviour issues and special services

Some parents whose children have disruptive or challenging behaviour may be told their child must be placed in a special class because the

behaviour is too disruptive to other students. For this to have occurred, "reasonable" attempts at adaptation and integration of the child must have been conducted. In addition, there are individual provincial/territorial statutes with requirements specific to that province/territory.

Courtesy of the National MPS Society

Inclusive education is legally required. Therefore, schools must have a means of identifying those students who are not completely able to adjust to a standard classroom situation as a result of a disability. An assessment and identification process is used to determine if the student with disabilities qualifies for teaching assistance. In these cases a modified Individualized Education Plan (IEP) is developed. From time to time, there are no specifically trained special education assistants or teachers available (for example, in a more isolated rural setting). In these circumstances the school or school board may authorize and provide training for the teaching personnel at a relevant special needs workshop.

The individualized education plan (IEP)

If you feel that your MPS II child needs an IEP plan, the following information will help you work with your school to set an appropriate plan.

IEP goal setting

The first step in creating an IEP is to develop some broad goals for the child. While broad goals will vary from child to child, here are some examples:

- Develop relationships with adults and children in school.
- Achieve as much self-help skill as possible.
- Be as self-directed as possible.
- Be happy with himself/herself and his/her school.
- Develop the desire to be independent.
- Behave acceptably at school.
- Be accepted by others, both students and adults.

The next step is to look at the child's current performance and needs in a variety of educational areas, including:

- **academic/cognitive skills:** Aiming at appropriate academic learning goals such as reading, math, social studies, etc. – these will vary according to the child's level of academic skill and potential.
- **emotional development:** Developing satisfaction with school, life, and self, improving self-control, and enhancing personal efficacy.
- **social development:** Developing friendships, interacting with peers, feeling part of a group, contributing to the good of the school and classroom, and having models of appropriate social behaviour.
- **communication skills:** Building skills that develop understanding and communication with others – for example, improving language skills, learning compensatory communication skills, and being exposed to and practicing appropriate language skills.
- **sensory skills:** Dealing with the effects of vision and hearing loss, providing a satisfactory and stimulating environment, and protecting the child from an environment that is too stimulating.

- mobility/physical development: Working towards individual goals for mobility, coordination, and physical skills – this includes regular and adaptive physical education activities and activities with other children.
- medical/health needs: Finding and maintaining supports that meet the medical and health needs of the child to enable him or her to benefit from his or her educational program.

Strength-based planning – child

Considering your child's strengths provides a new focus on the child that can be built into the IEP program. Strengths and likes can be used as learning tools. The focus on strengths and likes enhances motivation and allows everyone to enjoy the experience.

Parents should ask the IEP team to focus on the child's strengths and likes. The team can refer to these strengths and likes when composing the IEP. Below are questions to ask about the child. Be open to new ideas as the meeting progresses.

- What is the child's favourite thing to do?
- What areas have had the most success?
- Who does the child like best in school (teachers, peers, other school staff)?
- What was the best day the child had this year? What activities and events occurred on that day?
- What activities do the parent and child enjoy doing together?
- What does the child do well?
- What are the child's strongest physical and motor skills?
- If the child could do anything, what would it be?
- What are the child's favourite foods?

There are many more examples of questions parents can ask to help the IEP team identify the child's strengths and likes. Encourage other team members to brainstorm and think about specific times and events in the past few months that remind them of strengths and likes.

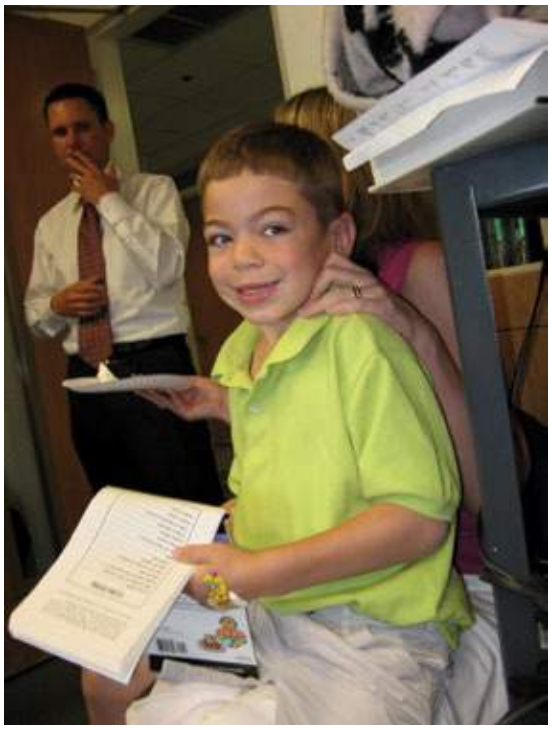
Strength-based planning – school

This approach also can be used when planning the classroom setting. Some professionals who work with children often refer to the "wrap-around" approach to developing plans. The idea is to think in terms of what supports the teacher needs in order to help this student be successful (meet his or her educational goals). Parents can do a strengths and likes assessment of the teacher and classroom, too! Here are some ideas to get started on a school strength assessment:

- What are the best aspects of the classroom?
- What does the teacher do for fun in the classroom?
- With what types of children with special needs is the school most successful (success means providing the child with the best possible education)?
- What are some things children enjoy doing in the classroom?
- What aspects of teaching does the teacher do best?
- What types of students respond well to the teacher?
- What is most exciting for the teacher on the first day of school each year?
- Who are the most supportive individuals in the school?
- What are the teacher's favourite subject areas?

Many people don't consider the fact that children with serious disabilities have strengths.

This also can be an illuminating process for the teacher. It gets people thinking about how to use their strengths to educate children. It may identify things that people haven't thought of in a long time. It also gets people to think about developing supports to better serve the children. It helps if parents have a few strengths of the teacher and school that have been identified before the IEP meeting.



Courtesy of Shire HGT

Preparing for the IEP meeting

Be an advocate for your child. Prepare ahead of time for the IEP meeting and keep the focus on what your child's needs and goals are. Review the child's records. Read over the last IEP. Make notes on areas where the child has improved and areas where there needs to be more work. Review any classroom work or progress notes received since the last review. Review reports or evaluations from outside professionals. Bring these reports to the meeting. Make a list of the child's strengths and a list of his/her needs. Think of academic, social, emotional, and physical strengths. Involve the child and other family members. Make a list of the things that need to be done to meet the child's needs. Think in terms of classroom size, peers, accommodations, curriculum, modifications, related services, assistive technologies, and transition. Make a list of the main points to be discussed at the meeting. Be sure to specifically discuss assistive technologies and adaptive physical education (discussed later in this section). Read materials on IEPs and work with the school so you are prepared. Parents may want another person to attend the meeting for support or to have another professional attend who can better explain the child's needs.

Having a successful IEP meeting

- Remember to bring all the information to the meeting.
- If something is difficult to understand, ask to have it explained. Ask questions.
- Keep emotions in check. It may be difficult, but it is best to remain calm. It is helpful to have another person present to provide support. If the meeting seems to be too emotional, ask for a break or reschedule another meeting.
- Keep in mind that the parent is a full member of the IEP team, and by working together the team can help the child have the best possible education.
- Listen to what others have to say and agree with what is reasonable.
- Make sure all of what is decided is written down on the IEP and get a copy.
- If you disagree with the school, attach a written statement of the disagreement to the IEP and don't forget your right to appeal.

Managing disagreement with the IEP

If the parent and school personnel don't agree on the IEP, focus on areas of agreement and work from there.

- Try to agree on as much of the IEP as possible so the school can begin implementing the plan.
- Look for others to help (other parents, special education law centers, advocates for the disabled, and organizations for children with disabilities). Take someone else with you to meetings who can observe and can assist you.
- Work hard at keeping cool during meetings.
- Write down goals and plans before going to meetings and provide evidence that backs the claims.

If the experience is difficult, hang in there. Remember, most of the time the local school district has the burden of proof that its programs are meeting the child's needs. And it's also important to keep in mind that you and the

school share the same goals – to help your child have the best possible education.

If parents cannot come to an agreement through discussion, negotiation, or mediation, and they feel that their child is being discriminated against by the school board, a complaint may be lodged with their provincial or territorial human rights commission or agency. After the investigation is complete, a commission or agency employee writes a report. The staff usually tries to facilitate a negotiated settlement. If a settlement is not reached, the case may be referred to a specialized tribunal, board of inquiry, board of adjudicators, or human rights court. A decision will then be made as to whether or not there has been discrimination in violation of the law. If there is a hearing, the human rights commission or agency will supply a lawyer to the complainant. It may take months, however, before the hearing occurs.

Monitoring progress with the IEP

It is important to monitor the progress of your child's education:

- Keep in touch with the child's teachers, principal, and other related personnel. Visit the school as often as possible.
- Keep a record of the child's progress on the IEP goals.
- Ask for a review of the IEP as the child's needs change.
- Make sure the IEP is being followed.
- Make sure that school personnel provide documentation that goals have been met or needs have changed.

Consider the following areas:

Academic achievement

- Is the child meeting their academic goals?
- Is he or she meeting the goals of the IEP?
- Does the teacher believe that academic goals are important for the child?
- Does the teacher encourage the child's best performance?

Courtesy of the Canadian MPS Society

- Does the child have appropriate expectations of his or her academic performance?
- Does he or she try hard to reach the goals?

Social development

- Does the child have a variety of friends in the class (a close friend, some acquaintances)? How many friends and how often the child plays with them may be related to the child's basic temperament and ability level.
- Does the teacher promote social interaction between all students in the classroom?
- Does the child get to work cooperatively with other students during learning activities?
- Does the child participate in non-academic activities (sports, socials, etc.)?

Emotional development

- Does the child like school? Is this a change?
- Does the child feel that he or she can master his or her environment at school?
- Does the child have strong negative feelings at school (anger, sadness, anxiety)?
- Does the child manage his or her feelings satisfactorily at school (anger, sadness, etc.)?

School environment

- Is the child getting an appropriate amount of assistance or support to reach his or her emotional, social, and academic goals?
- Does the teacher understand the child's needs?
- Does the teacher want to teach exceptional children in the classroom?
- Are the classroom and other areas easily accessible for the child?

- Are school-related tasks modified where appropriate but still similar to tasks other children do?

Behaviour intervention plan

Overview

All children who receive special education services and have behaviours that get in the way of their learning or the learning of other children must have a behaviour intervention plan as part of their individualized education plan (IEP). This should include an analysis of the problem behaviours, along with an assessment of the factors that might cause these behaviours.

The program also should specify strategies to help reduce the problem behaviour. These strategies should include positive behavioural interventions, such as reinforcement (reward) for appropriate behaviour, and teaching the skills necessary to perform the appropriate behaviour. There also should be attention directed to arranging the classroom environment to reduce stress on the child and to provide support for appropriate behaviour. An example of a situation where a classroom support is needed would be for a child who cannot communicate his needs clearly and has angry outbursts as a result. One possible solution would be to provide a communication board with words or pictures that the child can point to as a way of communicating with the teacher or other children. If the communication need is met, there is less need for the child to become angry.

If their child's behaviour problems interfere with learning, parents should make sure the IEP includes psychological services. This would allow the school psychologist to consult with the teacher in implementing behavioural interventions.

It is also important for the behaviour intervention plan to be consistent with the discipline the child receives at home.

Keep in mind that not all children with MPS II can be disciplined, and these behavioural interventions may not work for children with significant developmental delays. For these children, other strategies such as redirecting the child to another activity away from other children may be helpful.

Dealing with difficult behaviour

It is important to consider the child's developmental age when dealing with difficult behaviour. The discipline suggestions below may not work for children with severe developmental delays, and not all children with MPS II can be disciplined. In this case, you and the school may need to explore other options for dealing with the child's behaviour, such as avoiding over-stimulation, encouraging physical activity, and redirecting the child to another activity away from other children when the problem behaviour is occurring. You may also need to enlist the help of a special education expert or school psychologist.

When dealing with difficult behaviour, look closely at the environment and use consistent strategies to manage the behaviour. Try to identify what the function of the behaviour is. Possible functions include:

- to communicate anger, boredom, pain, or hunger
- to avoid a task the child finds unpleasant
- to get something the child wants
- to discharge pent-up energy

Look closely at the environment. Environmental change can often make a difference.

Simplify the environment. Frequent negative behaviour results from an environment that is too complex, is difficult to understand, or has too high expectations. In these cases, try to simplify the environmental demands on the child.

Make the environment more attractive or stimulating. Other times, difficult behaviour is the result of an environment that is boring or unattractive. Improving appropriate room decorations, activities, and levels of stimulation can improve the learning environment.

Use positive interventions first. Identify positive behaviour and reinforce it as much as possible. Try to arrange it so the child is able to be successful at things and has more positive interactions with people than negative interactions. Attention, praise, and smiles are all examples of reinforcements.

Courtesy of Shire HGT

Use mild penalties. Don't over-punish negative behaviour. Usually mild penalties will result in behaviour changes. Removing the child from the situation, taking away a privilege, and turning your back are all examples of mild penalties.

Don't take it personally. Much of the behaviour of children with MPS II is not directed personally at caregivers. Taking it personally makes you angry and liable to punish more harshly.

Be consistent. Consistency is often reassuring for children, especially those who have limitations in understanding or communication. Changing the limits of what you are willing to tolerate can make difficult behaviour less likely to resolve itself. Consistently reinforce positive behaviour and penalize negative behaviour.

Find people to help. School psychologists and special education teachers with experience in working with autistic children or children with emotional problems often are the best people to consult about behaviour management.

Consider what others have done to assist with these specific behaviours:

Fears

- Consult/train teachers to intervene empathically.
- Use behaviour management and medication.

Overactivity/restlessness

- Provide a quiet place with little stimulation and few choices until the child is calmer.
- Designate an activity table to include favourite books or toys for a one- to five-minute break.
- Have the child take a walk around school, then go back and continue the activity.

Aggressive/destructive

- Physically protect and block the child. Put your hand on top of the child's hand to tell him or her to "stop" or "let go."
- Be a role model for how to deal with anger.
- Label appropriate touching (e.g., no hitting) and use positive reinforcement of appropriate behaviour.
- Teach other children to remind the child to use nice touching.
- Have other children leave a space around the child so he or she doesn't feel crowded.
- Teach others to approach the child from the front so as not to startle him or her.
- Arrange the environment to reduce the child's frustration.

Sensory stimulation

- Assess undesirable behaviours that may be adopted because the child is trying to manipulate the environment to increase or decrease stimulation to a desired level. The function of sensory stimulation and automatic reinforcement is to increase or reduce stimulation.
- Check for possible health problems (such as an ear infection) that may be uncomfortable.
- Provide an enriched environment, such as stimuli that match the behaviour. For example, provide textured items to replace behaviour that seems to provide tactile stimulation.
- Reduce sensory stimulation when over-stimulation may be causing the negative behaviour. Move to a quieter area, and pad the area if necessary to prevent tactile stimulation.
- Change noise, crowding, and room temperature to provide a more optimal environment based on the child's preferences.

- Consult with an occupational therapist for additional strategies.

The discipline strategies used at school should be consistent with those used at home by the child's parents.

Adaptive physical education

Children with MPS II, like all children, must have a physical education program. The individualized education plan (IEP) should include the services of an adaptive physical educator. Adaptive physical educators learn how to adapt existing physical education activities and develop innovative activities to meet the physical, motor, personal, social, and learning needs of children with disabilities.

The adaptive physical education teacher should assess the child's physical education needs and develop an adaptive physical education program as part of the child's IEP. Adaptations can be made in existing games to include children with disabilities. Some possibilities include the following:

- using a batting tee instead of pitching in a softball game
- having designated runners
- decreasing the distances in games
- using real teams where children assist each other in parts of activities
- allowing children in wheelchairs to hold the ball on their lap while being pushed by another child
- changing rules of games
- modifying equipment
- adapting the layout of the game space
- developing new games that emphasize interaction rather than competition

Adaptive physical educators may work directly with children with disabilities and/or may provide consultation to teachers and parents on how to provide physical education activities that meet a specific child's needs.

Every school district should have at least one adaptive physical educator. If not, one can be hired as a consultant. Evaluate the effectiveness of the goals and activities at each IEP update, and have the adaptive physical educator present at the IEP meeting.

Parents can ask for an IEP team meeting if they aren't sure how things are going or if they aren't happy with the current status of their child's physical education plan.

Assistive technology

It is important to consider whether a child needs assistive technology devices and services to accomplish the educational goals in the IEP. An assistive technology device is a piece of equipment or system used to maintain or improve the functional capabilities of a child with a disability. These devices can be homemade or commercially available. Some devices may be specifically designed for persons with a disability; others may be commonly available.

Assistive technology helps compensate for limitations in functional skills caused by a disability. These devices can be used to help communicate, control the environment, get around, and do other activities of daily living. They have been commonly used for children with a variety of disabilities. Few devices are likely to be available off the shelf for children with complex physical disabilities, so it may take some creativity to modify already existing devices, or create homemade devices, to fit the particular situation. Each school district should have an assistive technology specialist on staff or available as a consultant to help you.

The following are examples of assistive technology devices for different areas of activity:

Communication

- Augmentative communication devices use computerized devices to provide a "voice" for communication.

Daily activities

- Devices can make it easier for a person to turn something on and off. "The Clapper" is one

such device that allows a child with mobility difficulties to turn a light on and off without getting up.
- Mobility aids can help a person get around or participate in an activity that otherwise requires a motor skill the child finds difficult because of mobility or motor control difficulties.
- Specially designed recreational equipment can allow a child to participate in games or sports. For example, a ball ramp can allow children to bowl who cannot pick up and move a bowling ball.

Educational activities
- Large-button calculators can assist with math.
- Speech recognition software and other computer software can enable a child to enter and read text.

To successfully use assistive technology for your child's IEP, make sure:

- the assistive technology device doesn't inhibit the child's development or reduce his or her skill level, but extends his or her capabilities
- the parent, child, and school personnel receive adequate training on the use of the device
- service and maintenance are available for the device
- back-up plans exist if crucial devices break down
- the child's assistive technology needs are monitored regularly
- all assistive technology devices are written into the IEP
- the IEP team considers the child's assistive technology needs only after determining his/her educational goals
- a person knowledgeable about assistive technology is on the IEP team
- the device is sent home if the child needs the assistive technology at home
- the child has a monitored trial period with the device to ensure it is functioning properly

Resources for more help

Websites
- Canadian Education Association: www.cea-ace.ca/foc.cfm?subsection=pol&page=lan&subpage=int
- Persons with Disabilities Online: www.pwd-online.ca/pwdhome.jsp?&lang=en
- The National Educational Association of Disabled Students: www.neads.ca/en/
- Inclusive Education Canada: www.inclusive-education.ca/english/index.asp
- Learning Disabilities Association of Quebec: www.ldac-taac.ca/Chapters/qc-e.asp
- School-Aged Therapy Program www.mcf.gov.bc.ca/spec_needs/school_aged_therapies.htm
- Adult Special Education www.aved.gov.bc.ca/adultspecialed/welcome.htm
- A Parent's Handbook on Inclusive Education (British Columbia Association for Community Living): www.bcacl.org/documents/Final_Handbook_144_pp.pdf
- Wrightslaw Special Education Advice and Resources: www.wrightslaw.com
- Project INSPIRE at Texas Women's University: www.twu.edu/inspire
- Ericec.org: www.ericec.org
- PE Central at Virginia Tech (a variety of lesson plans and activities): www.pecentral.org
- The Adapted Physical Education homepage at the University of Virginia (see what adapted physical education teachers learn in their program of study): www.teach.virginia.edu/overview-ape-324
- RESNA (Rehabilitation Engineering and Assistive Technology Society of North America): www.resna.org
- ABLEDATA: U.S. Dept. of Education Assistive Technology Clearinghouse: www.abledata.com
- National Association of School Psychologists: www.nasponline.org
- Little People of America: www.lpaonline.org

MPS II journal

Introduction

How can this journal help?

This journal has been designed for you, to help you organize your schedule and records for medications, supportive care, and medical tests. There is also a section that helps you keep track of the members of your medical team, and a section to help you prepare for medical visits. There's even a diary where you can record thoughts, observations, and questions. Using the journal helps you keep all important medical information in one convenient place for easy reference. And it's simple to use!

How do I use the journal?

Just fill in each section as you have a new medical test, visit, or medical intervention or procedure, or whenever you add a new member to your medical team (e.g., doctor, physical therapist, speech therapist). You can write in the diary as often as you wish. See the "What this section is for, and how to use it" box in each section for more details on how you can get the most out of using the journal.

You may wish to make extra copies of the journal pages so that you don't run out of space.

The journal contains the following sections:

- Glossary of terms used in the journal
- My medical team
- Getting the most out of your visit
- Medication log
- Care log
- Test results/medical reports log
- Observation diary
- Notes

Why are regular doctor's visits and medical tests so important for individuals with MPS II?

The effects of MPS II and the management of MPS II vary from person to person. All individuals with MPS II should have certain medical tests on a regular basis. Regular medical appointments help the doctor keep track of your health and the management of your MPS II. You can play a role in monitoring progress as well by keeping track of tests your doctor has ordered. Open communication with your team of healthcare specialists is important to ensure appropriate care. Different medical tests are recommended at different time intervals. Speak to your doctor about which medical tests you should have, and how often you should have them.

Canadian MPS Society
for Mucopolysaccharide & Related Diseases

Glossary of terms used in the journal*

BiPAP: Bilevel positive airway pressure, often used for people with sleep apnea to open the airway during sleep by blowing in air through a special mask. It uses two pressure settings, one that is used while the person is breathing in and the other that is used while the person is breathing out.

cardiologist: A medical doctor who specializes in disorders of the heart and blood vessels.

cerebrospinal fluid (CSF): The fluid that surrounds the brain and spinal cord and that is produced in the ventricles of the brain.

CPAP: Continuous positive airway pressure, often used for people with sleep apnea to open the airway during sleep. A special mask blows air into the airway at a constant pressure to keep it open.

echocardiogram: Ultrasound of the heart to evaluate for heart valve and heart muscle function.

G-tube: A tube inserted into the stomach using a surgical procedure in which an opening is made into the stomach from the outside. It is usually performed to allow nutrition and/or medications to be given directly into the stomach when swallowing is difficult because of disease or blockage of the esophagus (the tube from the mouth to the stomach).

hydrocephalus: An abnormally high level of cerebrospinal fluid within the brain.

J-tube: A tube inserted into the small intestine using a surgical procedure in which an opening is made into the small intestine from the outside. It is usually performed to allow nutrition and/or medications to be given directly into the small intestine when swallowing is difficult because of disease or blockage of the esophagus (the tube from the mouth to the stomach).

I2S: Iduronate-2-sulfatase, the enzyme that is deficient in MPS II.

MPS II: Mucopolysaccharidosis II (Hunter syndrome), an inherited genetic disorder that affects many body systems and that may lead to damage of different body organs (e.g., the heart). MPS II is caused by a defect in the gene that instructs the body to make an enzyme called iduronate-2-sulfatase, also called I2S.

shunt (ventriculoperitoneal shunt): A thin tube that drains fluid from the brain into the abdominal cavity. Used in the treatment and management of hydrocephalus.

tracheostomy: A surgical procedure in which a hole is made into the trachea through the neck to relieve obstructions to breathing. A curved breathing tube is usually inserted through the hole.

**For further information on medical terms used in the journal and the binder, including the terms underlined above, please consult the main Glossary.*

My medical team

What this section is for, and how to use it:

This section helps you keep track of the different health professionals that are part of your, or your child's, medical team. Because MPS II affects many different aspects of the body, a number of different health professionals may be involved in your care. For example, your medical team may include specialists such as anesthetists, cardiologists, and ophthalmologists. It may also include other health professionals such as physical therapists or speech therapists. Keeping track of all of these different professionals can be a challenge, but this section helps you keep all of your information organized in one convenient place.

To use this section, simply fill in the information on each medical professional on your team. Use the business card holders provided to store the business cards from your health professionals.

You may wish to make extra copies of the medical team log pages so that you don't run out of space.

Name/Profession	Address	Phone/Email	Notes
Example: Dr. John Doe Cardiologist	123 Main Street, Anytown BC V1V 1V1	111-123-4567 jdoe@cardiocare.org	No appointments available on Fridays. Best way to reach him is by phone.

Name/Profession	Address	Phone/Email	Notes

Getting the most out of your visit

What this section is for, and how to use it:
This section helps you prepare for your medical visits to ensure that you get the most out of each visit. Review this section before each medical visit. This section refers to either your visit and medications if you are an adult with MPS II, or to your child's visit and medications if you are a parent of a child with MPS II.

You or your child will get more out of your visit if you are prepared ahead of time. Here are a few tips:

Before your medical visit

- Write down any questions you may have for your health professional (e.g., doctor, nurse, physical therapist). Put your most important questions first. See the list below for some ideas to get you started.
- Make a note of any concerns you would like to discuss at the meeting (e.g., new symptoms).
- Think about what you would like to achieve from the visit (e.g., "During this visit, I would like to gather the information I will need to decide whether to start tube feeding for my child.")
- If you don't already have one, put together an up-to-date list of your or your child's medical conditions, medications, recent procedures, and medication allergies. You can use the journal to keep track of this information, then bring the journal to your medical appointment.
- Confirm the time, date, and location of your appointment. If you have not been there before, make sure you have directions on how to get to your appointment.

Going to your visit

- Bring your journal with you so that you will have all of your medical information in one place. The journal will also help the various medical professionals involved in your care share information, which can save you time and ensure that everyone has the complete picture.
- Bring a pen or pencil to take notes (or a tape recorder if you would like to record the visit, with your medical professional's permission).
- Leave plenty of extra time to get to the appointment so you don't have to worry about being late. Plan to arrive a bit early to give yourself time to park, find the office, and check in at the desk.
- You may wish to bring a friend or family member with you to the visit for support, and to take notes and remind you to ask your questions.

At your visit

- If the health professional says anything you don't understand, ask them to explain it to you again. Sometimes having it explained in a different way can help.
- Ask the health professional the questions from your list. To make the most of your time during the visit, make sure you ask your most important questions first.
- Take notes, have a family member or friend take notes, or tape-record the visit (first, get the health professional's permission). Ask if there are any written materials you can take with you.
- Find out if and when you should follow up with your health professional after the visit, and ask about the best way to contact your health professional to follow up.

After your visit

In some cases, you may need to follow up with your health professional after your visit. Reasons to follow up may include:

- to get the results of medical tests done during the visit
- to ask questions that came up after the visit
- to report on how a new medication is working
- to get help if symptoms get worse or if medication side effects occur

Questions to ask your health professional

The questions you will want to ask during your visit will depend on your individual situation, the type of health professional you are seeing, and the nature of your visit. Use this list as a guide to develop your own personal list of questions.

1. What is causing my symptoms? Are these symptoms related to MPS II or to another condition?
2. Do I need to have any medical tests?
3. How can I manage MPS II?
4. Which management options do you recommend and why?
5. For a specific medication:
   a. What are the benefits?
   b. What are the risks?
   c. How will it affect my daily life?
   d. Are there any possible interactions with my other medications or medical conditions?
   e. What will the medication help with, and how much of an improvement can I expect?
   f. When will the medication start to work?
   g. How long will I need to use the medication?
   h. When should I seek medical attention (e.g., if you experience certain side effects, if your symptoms get worse)?
   i. Will my insurance cover this medication?
6. Are there any modifications I can make to my home or work to make my daily activities easier?
7. What else can I do to improve my health?
8. What other resources are available to help me learn about MPS II?
9. When can I expect to get the results of any medical tests that were done?
10. When should I come back for another visit?
11. Other: _____
12. Other: _____
13. Other: _____
14. Other: _____

These questions may not apply to everyone. When putting together your list of questions, select the ones that apply to you or your child, and add any extra questions you may have.

Medication log

What this section is for, and how to use it:

This section helps you keep a record of medications and medication allergies. Keeping a current medication list is important for your health and safety. The list will ensure that all medical professionals are aware of any medication allergies and can avoid potentially dangerous drug interactions. In the "details" space, you can write the dosage and any observations you may have about the effects of the medication. Remember to contact your doctor if there are any side effects. To learn more about medication safety, visit the Health Canada website at www.hc-sc.gc.ca.

You may wish to make extra copies of the log pages so that you don't run out of space.

Current medications (include name, dose, how it is given, and how often it is taken)	**Purpose of medication** (what medication is being used for)	**Date started**	**Date stopped**	**Details**
Example: Acetaminophen 80 mg by mouth every 6 hours	For pain and fever	April 2, 2010	April 4, 2010	Helped relieve pain and fever.

Current medications (include name, dose, how it is given, and how often it is taken)	**Purpose of medication** (what medication is being used for)	**Date started**	**Date stopped**	**Details**

Medication allergies (include a brief description of the allergy)

Example:
penicillin (hives, swelling of face and throat)

Additional comments

Supportive care log

What this section is for, and how to use it:

"Care" refers to important medical procedures and interventions that are used to manage the symptoms of MPS II, but are not medications. Examples of supportive care include physical therapy, speech therapy, respiratory therapy, insertion of a feeding tube (such as a G-tube or J-tube), CPAP or BiPAP machines for sleep apnea, tracheostomies, or shunts for hydrocephalus. This section helps you keep a record of your medical interventions and procedures. It will also inform medical professionals of the medical interventions and procedures that have been received. You may also record the results of other medical visits (e.g., specialist visits) here.

You may wish to make extra copies of the log pages so that you don't run out of space.

**Please note that individuals should always consult their physicians before starting any form of supportive therapy. Persons requiring diagnosis or management or with questions specific to a single individual are urged to contact their local healthcare providers.*

Type of supportive care	Date	Health professional's name and location	Results/Health professional's observations
Example: G-tube placement	April 7, 2010	Dr. Anna Sample, Memorial Hospital, Anytown	G-tube successfully placed. For questions about G-tube care, contact Frieda Smith, RN, at 444-123-1234.

Type of supportive care	Date	Health professional's name and location	Results/Health professional's observations

Test results/medical reports log

What this section is for, and how to use it:

The test result/medical reports log was developed to help you keep track of medical test results and reports that you receive over time. MPS II can affect many different areas of the body, so you may be referred to different specialists. These specialists may order regular medical tests to get a complete picture of how the condition is affecting your body over time. That's why you may have tests even for body areas that aren't giving you any problems right now. For example, your cardiologist (heart doctor) may recommend an echocardiogram, a test that checks for heart problems, even if you're not having any trouble with your heart.

To use this section, simply write down the test name, date, and results (if you have a printout, you can insert it in the folder provided) in the spaces of the table. This way all of your test results will be in one convenient location.

Period: MM/DD/YY ***to*** MM/DD/YY

Test/assessment	Date	Comments/observations
Example: Hearing test	April 1, 2010	Moderate hearing loss (see full report in folder)

Test/assessment	Date	Comments/observations

Observation diary

What this section is for, and how to use it:

The diary is your place to write down any thoughts, feelings, or questions you may have. Not only can it help you keep track of progress over time (e.g., improvements in symptoms or abilities), it can also give you somewhere to write about feelings and concerns as you go through the care journey.

To use this section, simply write down your feelings, concerns, and observations in the pages provided. You may wish to make extra copies of these pages so that you will have plenty of space to write in. If you only have a short amount to write for a particular day, you can start the next day's entry on the same page (just add the date).

DATE

DATE

Notes

What this section is for, and how to use it:

The notes section is a place to write down any extra information that doesn't fit under the other sections. For example, you could use this section to write down notes that you take during a doctor's visit.

To use this section, simply write your notes in the pages provided. You may wish to make extra copies of the pages so that you will have plenty of space for writing notes.

An overview of MPS II for doctors

Information handout

You may wish to make additional copies of this handout so that you will not run out.

Definition, causes and incidence

Mucopolysaccharidosis II (MPS II), also known as Hunter syndrome, is a rare X-linked recessive disease that has progressive, pathologic manifestations in many organ systems. The disease is caused by a defect in the gene coding for the lysosomal enzyme iduronate-2-sulfatase (I2S). As a result of this genetic defect, cells either are unable to produce the enzyme or produce it in low amounts. This results in an inability of the lysosome to catabolize the glycosaminoglycans heparan sulfate and dermatan sulfate – a process essential for normal growth and homeostasis of tissues. These glycosaminoglycans, commonly referred to as GAGs, progressively accumulate in the lysosome, ultimately causing irreversible damage to cells, tissues, and organs. It has been estimated that MPS II occurs in at least 1 of every 170,000 live male births.

Clinical presentation and prognosis

Because of its X-linked recessive inheritance, MPS II mainly affects males. It is a progressive disease with a broad clinical spectrum, with the most rapidly progressing individuals on one end, the most slowly progressing individuals on the other end, and a whole range of different severities in between. Disease severity varies significantly for people with MPS II and it is not possible to predict the expected lifespan for a given individual. Those on the more slowly progressing end of the disease spectrum may have

Mucopolysaccharidosis II (MPS II) is a rare autosomal recessive disease that has progressive, pathologic manifestations in many organ systems. The disease is caused by a defect in the gene coding for the lysosomal enzyme iduronate-2-sulfatase.

Spectrum of disease severity in MPS II:

Rapidly progressing ← → *Slowly progressing*

Courtesy of the National MPS Society and Shire HGT

Pull out and copy this section

Table 1: Clinical manifestations of MPS II

General symptoms
- reduced endurance

Physical appearance
- abnormal facial features (such as broad, flat-bridged noses; thick lips; flat faces; eyes that stick out slightly; and an enlarged tongue that may stick out)
- macrocephaly
- short stature

Mouth and teeth
- enlarged tongue
- abnormal teeth (widely spaced and poorly formed teeth with fragile enamel)

Eyes, ears, nose, and throat
- reduced visual field
- optic nerve disease
- glaucoma (not common)
- impaired hearing
- recurrent otitis media
- recurrent sinusitis

Respiratory system
- obstructive airway disease
- restrictive airway disease
- sleep apnea
- reduced pulmonary function
- recurrent pulmonary infections

Cardiovascular
- valvular disease
- cardiomyopathy
- cardiac arrhythmia
- pulmonary hypertension

Gastrointestinal system
- hepatosplenomegaly
- umbilical and inguinal hernias

Musculoskeletal system
- joint stiffness and contractures
- skeletal abnormalities (dysostosis multiplex)
- hip dysplasia

Brain, nerves
- cervical spinal cord compression
- communicating hydrocephalus
- carpal tunnel syndrome
- developmental delays (although slowly progressing individuals may have normal intelligence)
- slowing of mental development by 2 to 4 years of age, followed by regression in skills (in rapidly progressing individuals)

a reasonably normal lifespan. Individuals who are more rapidly progressing are likely to have a shortened lifespan.

The signs and symptoms of MPS II involve most organ systems. Table 1 describes the clinical manifestations of MPS II.

Diagnosis

More common childhood conditions are usually ruled out before considering a diagnosis of MPS II. The first step in diagnosis is an analysis of urinary GAG levels. If high levels suggesting MPS II are found, then iduronate-2-sulfatase activity levels are measured in a blood or skin sample. Enzyme activity tests are essential for a definitive diagnosis. If iduronate-2-sulfatase activity levels are abnormally low, the diagnosis of MPS II is confirmed. To rule out multiple sulfatase deficiency, another sulfatase should be measured at the same time. DNA testing may be considered to determine the specific genetic mutations, which is helpful information if others in the family are to be tested. Testing may also be considered for others in the family. Families are encouraged to visit a genetic counsellor.

Available/emerging management options for MPS

Multidisciplinary medical management of disease is an ongoing component of all MPS disorders. This disease management includes procedures such as surgery to deal with specific disease manifestations as well as physical therapy or speech therapy on an ongoing basis. For some MPS disorders, treatments that target the underlying enzyme deficiency are also available. Please call The Canadian Society for Mucopolysaccharide & Related Diseases (Canadian MPS Society) at 1-800-667-1846 for additional information.

An overview of MPS II for teachers

Information handout

You may wish to make additional copies of this handout so that you will not run out.

What is MPS II?

Mucopolysaccharidosis II (MPS II; pronounced **mew**-ko-**pol**-ee-**sak**-ah-ri-**doh**-sis **two**) is a rare genetic disorder.

MPS II is also known as Hunter syndrome. Because of the way the disease is inherited, it mainly affects males. The gene for MPS II is found on the X chromosome. Males have an X and a Y chromosome. The Y chromosome is smaller and does not contain a backup copy of the gene that is defective in MPS II. This means that if a male inherits an X chromosome with a faulty copy of the MPS II gene from his mother, he will have the disease. But since a female has two X chromosomes, she will inherit a healthy "backup" copy of the gene, so she will usually not have the disease. However, she will become a "carrier" who can pass on the gene to her children.

Mucopolysaccharidosis II (MPS II) is a rare genetic disorder. It is caused by a deficiency of an enzyme called iduronate-2-sulfatase.

MPS II is caused by a deficiency of an enzyme called iduronate-2-sulfatase (pronounced eye-**dur**-o-nate **two sul**-fa-tace). This enzyme is needed to break up substances called glycosaminoglycans (GAGs; pronounced **gly**-cose-a-**mee**-no-**gly**-cans), which are long chains of sugar molecules. Without the enzyme, GAGs build up in cells throughout the body, leading to damage in many body systems and organs.

MPS II signs and symptoms may include:

- "thickened" facial features, a large head, and a large tongue
- hearing loss and vision problems
- possible developmental delays
- problems with speech and language
- breathing problems
- an enlarged liver and spleen, which can look like a large "belly"
- abnormal bone structure
- carpal tunnel syndrome
- joint stiffness and movement problems
- heart damage

The severity of the condition varies widely between affected individuals. It is now perhaps more appropriate to view MPS II as a continuous spectrum of disease, with the most rapidly progressing individuals on one end and the more slowly progressing individuals on the other end, with a whole range of different severities in between.

See Table 1 (next page) for a more detailed list of MPS II signs and symptoms. Not all people with MPS will necessarily show all of these signs and symptoms, and not all people with MPS will have the same degree of severity of symptoms.

How can MPS II affect a child's school performance?

Children whose MPS II is slowly progressing may have normal intelligence. However, children with more rapidly progressing disease usually have developmental delays. In rapidly progressing cases, mental development begins to slow by 18 to 24 months of age. After this point, individuals may make slow progress until the age of 3 to 5 years, when most individuals reach a plateau

Pull out and copy this section

Table 1: MPS II signs and symptoms

General symptoms
- reduced endurance

Physical appearance
- abnormal facial features (such as broad, flat-bridged noses; thick lips; flat faces; eyes that stick out slightly; and an enlarged tongue that may stick out)
- large head (macrocephaly)
- short stature

Mouth and teeth
- enlarged tongue
- abnormal teeth (widely spaced and poorly formed, with fragile enamel)

Eyes, ears, nose, and throat
- vision problems, such as optic nerve damage or a reduced field of vision (some people have glaucoma, but this is not common in MPS II)
- hearing loss
- frequent ear infections (otitis media)
- frequent sinus infections (sinusitis)

Respiratory system (lungs and breathing)
- lung problems and reduced lung function
- sleep apnea
- frequent lung infections

Heart and blood vessels
- heart valve problems
- damage to the heart muscle (cardiomyopathy)
- irregular heartbeat (cardiac arrhythmia)
- high blood pressure in the blood vessels supplying the lungs (pulmonary hypertension)

Gastrointestinal system (stomach and intestines)
- enlarged liver and spleen (hepatosplenomegaly)
- umbilical and inguinal hernias*

Musculoskeletal system (bones and joints)
- joint stiffness
- skeletal abnormalities (dysostosis multiplex)
- abnormal hip formation (hip dysplasia)

Brain and nerves
- pressure on the neck area of the spinal cord from abnormal tissue growth nearby (cervical spinal cord compression)
- fluid buildup in the brain (hydrocephalus)
- carpal tunnel syndrome
- • possible developmental delays (i.e., the child takes longer to reach milestones such as sitting up, walking, and speaking) – however, individuals with slowly progressing disease may have normal intelligence.
- slowing of mental development by 2 to 4 years of age, followed by regression in skills (in rapidly progressing individuals)

**Hernias: When part of an organ (such as the intestine) protrudes from a weak spot in the muscular wall surrounding the abdomen, producing a bulge in the skin, this is called a hernia. With an umbilical hernia, the bulge is in the belly button area. With an inguinal hernia, the bulge is in the groin area.*

in their development. Skills and development then regress and people with rapidly progressing MPS II are usually severely mentally handicapped by their second decade of life. Learning difficulties can be compounded by hearing loss and vision problems.

Changes in physical appearance (such as the bone problems, the "belly" from an enlarged liver and spleen, and thickened facial features) make children with MPS II look different from their peers, which could lead to feelings of isolation or teasing.

Physical symptoms such as claw hands and carpal tunnel syndrome, limited mobility, heart problems, and breathing problems may make it hard for children with MPS II to do the physical tasks and activities that their classmates find easy, such as handwriting, artwork, putting on a coat, or attending gym class. If the physical problems are severe, they may require a personal care worker or nurse.

Relevant legislation

Inclusive education is legally required in Canada; therefore, schools must have a means of identifying those students who are not completely able to adjust to a standard classroom situation as a result of a disability. Canadian human rights laws imply that schools and other education authorities have a legal obligation or "duty to accommodate," which ensures they take appro-

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priate steps to eliminate discrimination resulting from a rule, practice, or barrier that has, or can have, an adverse impact on individuals with disabilities.

How teachers can help children with MPS II

Teachers are part of the educational team for the child with MPS II. Here are a few ways that teachers may be able to help children with MPS II:

Canadian legislation stipulates that educational facilities have a "duty to accommodate."

- Teachers may want to encourage children with MPS II to socialize with unaffected children by organizing group activities where students work together and support each other.

- Teachers may want to monitor changes in school behaviour and performance so the IEP can be adjusted if necessary.

- Class assignments and projects (such as art projects) can be modified to allow children with physical limitations to participate in similar projects with their peers.

- Keep in mind that while some children with MPS II have developmental delays, others may have normal intelligence, even though they may appear to be affected physically. Academic and vocational programming should be adjusted accordingly.

- For severely affected children, behaviour problems may occur. It is important to consider the child's developmental age when dealing with difficult behaviour. Teachers may wish to request a consultation with a professional experienced in dealing with these behaviour issues.

- It is important to consider the child's developmental age when dealing with difficult behaviour. Children with MPS II without significant developmental delays should be disciplined consistently and appropriately according to their age and abilities. Teachers and parents should arrange to coordinate discipline strategies in advance to maintain consistency between home and school. However, not all children with MPS II can be disciplined. Discipline techniques may not work for children with severe developmental delays. In this case, teachers and parents may need the help of a special education expert or school psychologist.

- Teachers need support too! The child with MPS II may have a personal care worker or nursing assistant if required. Your school may have access to physical and occupational therapists as well as specialists in developmental disorders and speech therapists. Having a teacher's aide for the student may also be an option.

- For more information on IEP, placement issues, and support for children with MPS II, visit www.mpssociety.org/content/4064/MPS__ML_Booklets/ and download "Educational Strategies & Resources."

Resources

- Canadian Education Association: www.cea-ace.ca/foc.cfm?subsection=pol&page=lan&subpage=int
- Persons with Disabilities Online: www.pwd-online.ca/pwdhome.jsp?&lang=en
- The National Educational Association of Disabled Students: www.neads.ca/en/
- Inclusive Education: www.inclusiveeducation.ca/english/index.asp
- Learning Disabilities Association of Canada: www.ldac-taac.ca/
- School-Aged Therapy Program: www.mcf.gov.bc.ca/spec_needs/school_aged_therapies.htm

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- Adult Special Education: www.aved.gov.bc.ca/adultspecialed/welcome.htm
- A Parent's Handbook on Inclusive Education (British Columbia Association for Community Living): www.bcacl.org/documents/Final_Handbook_144_pp.pdf
- Wrightslaw Special Education Advice and Resources: www.wrightslaw.com
- Project INSPIRE at Texas Women's University: www.twu.edu/INSPIRE/
- PE Central at Virginia Tech (a variety of lesson plans and activities): www.pecentral.org
- RESNA (Rehabilitation Engineering and Assistive Technology Society of North America): www.resna.org
- Parents, Let's Unite for Kids Family Guide to Assistive Technology: www.pluk.org/AT1.html
- National Association of School Psychologists: www.naspweb.org

An overview of MPS II for case managers and support workers

Information handout

You may wish to make additional copies of this handout so that you will not run out.

What is MPS II?

Mucopolysaccharidosis II (MPS II; pronounced **mew**-ko-**pol**-ee-**sak**-ah-ri-**doh**-sis **two**) is a rare genetic disorder caused by a deficiency of an enzyme called iduronate-2-sulfatase (pronounced eye-**dur**-o-nate **two sul**-fa-tace). It can damage many systems and organs of the body.

Mucopolysaccharidosis II (MPS II) is a rare genetic disorder caused by a deficiency of an enzyme called iduronate-2-sulfatase.

MPS II is also known as Hunter syndrome. Because of the way the disease is inherited, it mainly affects males. The gene for MPS II is found on the X chromosome. Males have an X and a Y chromosome. The Y chromosome is smaller and does not contain a backup copy of the gene that is defective in MPS II. This means that if a male inherits an X chromosome with a faulty copy of the MPS II gene from his mother, he will have the disease. But since a female has two X chromosomes, she will inherit a healthy "backup" copy of the gene, so she will usually not have the disease. However, she will become a "carrier" who can pass on the gene to her children.

MPS II signs and symptoms include:

- "thickened" facial features, a large head, and a large tongue
- hearing loss and vision problems
- possible developmental delays
- problems with speech and language
- breathing problems
- an enlarged liver and spleen, which can look like a large "belly"
- abnormal bone structure
- carpal tunnel syndrome
- joint stiffness and movement problems
- heart damage

The severity of the condition varies widely between affected individuals. You can view MPS II as a continuous spectrum of disease, with the most rapidly progressing individuals on one end and the more slowly progressing individuals on the other end, with a whole range of different severities in between.

See Table 1 for a more detailed list of possible MPS II signs and symptoms. Not all people with MPS will necessarily show all of these signs and symptoms, and not all people with MPS will have the same degree of severity of symptoms.

Services that may help MPS II families

Individuals with MPS II and their families may need help from case managers and support workers to access a variety of services, including physical supportive care, emotional support, and financial assistance.

Physical supportive care

Depending on the severity of the condition, the doctor may request access to physical therapy, occupational therapy, respiratory therapy, and devices such as CPAP or BiPAP machines; hearing aids, speech therapy, or sign language tutoring; and home nursing care, personal care workers, or special education experts.

Emotional support

Parents and family members may need emotional support to help them cope. Families may need access to respite care, individual counselling, and support groups.

Table 1: MPS II signs and symptoms

General symptoms
- reduced endurance

Physical appearance
- abnormal facial features (such as broad, flat-bridged noses; thick lips; flat faces; eyes that stick out slightly; and an enlarged tongue that may stick out)
- large head (macrocephaly)
- short stature

Mouth and teeth
- enlarged tongue
- abnormal teeth (widely spaced and poorly formed, with fragile enamel)

Eyes, ears, nose, and throat
- vision problems, such as optic nerve damage or a reduced field of vision (some people have glaucoma, but this is not common in MPS II)
- hearing loss
- frequent ear infections (otitis media)
- frequent sinus infections (sinusitis)

Respiratory system (lungs and breathing)
- lung problems and reduced lung function
- sleep apnea
- frequent lung infections

Heart and blood vessels
- heart valve problems
- damage to the heart muscle (cardiomyopathy)
- irregular heartbeat (cardiac arrhythmia)
- high blood pressure in the blood vessels supplying the lungs (pulmonary hypertension)

Gastrointestinal system (stomach and intestines)
- enlarged liver and spleen (hepatosplenomegaly)
- umbilical and inguinal hernias*

Musculoskeletal system (bones and joints)
- joint stiffness
- skeletal abnormalities (dysostosis multiplex)
- abnormal hip formation (hip dysplasia)

Brain and nerves
- pressure on the neck area of the spinal cord from abnormal tissue growth nearby (cervical spinal cord compression)
- fluid buildup in the brain (hydrocephalus)
- carpal tunnel syndrome
- possible developmental delays (i.e., the child takes longer to reach milestones such as sitting up, walking, and speaking) – however, slowly progressing individuals may have normal intelligence.
- slowing of mental development by 2 to 4 years of age, followed by regression in skills (in rapidly progressing individuals)

**Hernias: When part of an organ (such as the intestine) protrudes from a weak spot in the muscular wall surrounding the abdomen, producing a bulge in the skin, this is called a hernia. With an umbilical hernia, the bulge is in the belly button area. With an inguinal hernia, the bulge is in the groin area.*

Financial assistance

Many families may benefit from financial assistance from health insurance or government programs to help cover the costs of medical treatment and devices for MPS II. To help with expenses that are not covered by public or private insurance plans, the Canadian Society for Mucopolysaccharide & Related Diseases (Canadian MPS Society) offers a Family Assistance Program and Bone Marrow Transplant Family Assistance Program to help MPS member families in need. Applications can be downloaded from the Canadian MPS Society website at www.mpssociety.ca.

Relevant legislation

There are a number of federal and provincial legislations that may affect access to health care, education, and services for families affected by MPS II:

Workplace legislation

- Section 15 of the Canadian Charter of Rights and Freedom guarantees equality rights plus freedom from discrimination for people who have a physical or mental disability. The Employment Equity Act (EEA) of 1995 ensures that persons with disabilities are

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granted full and equal access to employment and opportunity. An employer must accommodate the disabilities of employees, prospective employees, and clients or customers. Compassionate leave legislation is overseen in Canada at the provincial level. A summary of the elements of the compassionate care leave provisions in employment standards in legislation published by Human Resources and Skills Development Canada can be found online at www.hrsdc.gc.ca/eng/labour/labour_law/esl/compass.shtml.

Educational legislation

- Inclusive education is legally required in Canada. Therefore schools must have a means of identifying those students who are not completely able to adjust to a standard classroom situation as a result of a disability.
- Canadian human rights laws imply that schools and other education authorities have a legal obligation or "duty to accommodate," which ensures they take appropriate steps to eliminate discrimination resulting from a rule, practice, or barrier that has, or can have, an adverse impact on individuals with disabilities.

Social support

The Service Canada website of Health Canada links with a number of programs for financial support of people with disabilities. The website can be found online at www.servicecanada.gc.ca/eng/audiences/disabilities/index.shtml.

Here are a number of links to programs that provide financial assistance for people with disabilities and/or their families:

- Child Disability Benefit: www.cra-arc.gc.ca/cdb/
- Grants for Students with Disabilities (includes grants/accommodation/tuition/books etc): www.rhdcc-hrsdc.gc.ca/eng/learning/canada_student_loan/grant2.shtml
- Residential Rehabilitation Assistance Program for Persons with Disabilities (RRAP – Disabilities): www.cmhc-schl.gc.ca/en/co/prfinas/prfinas_003.cfm
- Children's Medical Equipment Recycling Loan Service: www.cnchl-cncdh.ca/article.asp?id=30163&tid=078
- Disabled Income Assistance: www.google.ca/search?hl=en&q=disabled+income+assistance&btnG=Search&meta=
- Fuel Tax Refund for Persons with Disabilities (British Columbia): www.sbr.gov.bc.ca/documents_library/bulletins/mft_004.pdf
- Canadian Bar Association: Financial help for people with disabilities: www.cba.org/bc/public_media/employment/289.aspx
- Canada Revenue Agency: Registered Disability Savings Plan: www.cra-arc.gc.ca/E/pub/tg/rc4460/rc4460-e.html
- Planned Lifetime Advocacy Network: www.plan.ca
- President's Choice Children's Charity: www.presidentschoice.ca/LCLOnline/aboutUsCharity.jsp

MPS II resources for case managers and support workers

The following resources may help you assist families affected by MPS II:

- The Canadian MPS Society, 1-800-667-1846, provides a number of programs to assist MPS II families:
  - A Family Assistance Program to help offset costs associated with caring for oneself or a child with MPS II. Grants may be used for home renovations, durable medical goods, or other costs associated with management and care.
  - A Respite Program to provide funding to parents who need a break from caring for their child(ren) with MPS II.
  - Scholarships to family conferences.
  - A Family Referral Directory service to connect families with other MPS II families who have volunteered to share their experiences.

- The Vocational Rehabilitation Association (VRA) of Canada is a national organization that supports and advocates for members who work with other professionals to deliver timely and effective vocational rehabilitation services.

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The VRA has provincial chapters which can be located online at vracanada.com/societies.php.

Individuals with disabilities should notify local emergency medical services about their unique circumstances and any special medical situations that would affect their care in case of an emergency. This will help healthcare providers offer the best possible care in an emergency situation. The form provided below can be filled out and faxed to your local medical emergency services agency.

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Emergency medical services notification

Patient's name: _____

Patient's age: _____

Patient's weight: _____

Patient's address: _____

_____

Patient's phone #: _____

_____ has Mucopolysaccharidosis II.
This is notification that the following special situations exist with this patient.

- ☐ Restricted airway
- ☐ Tracheostomy
- ☐ Gastrostomy tube
- ☐ Jejunostomy tube
- ☐ Supplemental oxygen: Amount _____
- ☐ Wheelchair assistance
- ☐ Limited ability to move head/neck
- ☐ Difficult to intubate
- ☐ Other medical assistance required:

_____

_____

Please fax this to your local emergency medical services agency.

If you need assistance finding who to send this to, contact your local emergency, police or fire department for the fax number of your local emergency medical agencies.

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Glossary

This content has been re-used in significant part with permission from the National MPS Society.

Words that are underlined in definitions also have their own entries in this glossary.

acetyl CoA: alpha-glucosaminide acetyltransferase (acetyl CoA: α-glucosaminide acetyltransferase): Lysosomal enzyme deficient in MPS III-C.

adenoids: The collection of lymphatic tissue at the rear of the nose. Enlargement of the adenoids may cause obstruction of breathing through the nose.

adenoidectomy: A surgical procedure to remove adenoid growth.

airway patency: The openness of the airways (parts of the respiratory tract through which air passes).

alpha-L-iduronidase (α-L-iduronidase): Lysosomal enzyme deficient in MPS I.

alpha-N-acetylglucosaminidase (α-N-acetylglucosaminidase): Lysosomal enzyme deficient in MPS III-B.

amino acid: A class of chemical compounds that can be built up to form larger polymers called proteins. In most biological systems there are 20 common amino acids that can be linked in various combinations to generate larger molecules containing 100–10,000 amino acids. These larger molecules, or proteins, carry out most of the active functions within a cell or an organism.

amniocentesis: Procedure involving withdrawal of amniotic fluid, the fluid that surrounds the growing fetus in the uterus, generally performed between the 15th and 20th weeks of pregnancy by inserting a needle through the abdominal wall into the uterus. Cells that are contained in the fluid can be isolated and used for prenatal diagnosis of gender and for particular genetic conditions.

anesthesia: A loss of feeling, particularly the sensations of pain and touch. Usually, the term is used to refer to the use of medication to temporarily suppress sensations, especially before surgery.

angioneurotic edema: Swelling under the skin of the face, throat, hands, or feet.

anterior: Front.

antibody: A type of protein that is produced by the immune system to attack foreign substances that enter the body.

aorta: A large blood vessel that transports blood from the heart to other parts of the body.

apneic episodes: Periods of time where breathing stops.

arrhythmia: An abnormal heartbeat.

arthralgia: Joint pain.

arylsulfatase B (also called ASB)**:** Lysosomal enzyme deficient in MPS VI.

aspiration: The accidental inhalation of food or liquids into the lungs. Aspiration may lead to a lung infection.

atrophy: A wasting of tissues, organs, or the entire body, as from death and reabsorption of cells, diminished cellular proliferation, decreased cellular volume, pressure, ischemia (blockage of blood flow), malnutrition, lessened function, or hormonal changes.

attenuated: Diminished, not as severe.

audiometry: Hearing tests.

autosomal recessive disease: A disease which follows an autosomal recessive pattern of inheritance requiring the presence of 2 copies of a particular gene mutation in order to express clinical signs and symptoms of a condition. A pattern of inheritance seen in all MPS disorders with the exception of MPS II.

autosomal recessive inheritance: A pattern of inheritance in which a nondominant (recessive) gene on a non-sex-determining chromosome (autosome) results in a person either being a carrier of a trait or being affected. Males and females are affected with equal frequency. There is usually no family history of the trait. Instead, it is revealed when 2 unaffected parents who are both carriers of a particular recessive gene have a child who receives 2 copies of the recessive gene. A pattern of inheritance seen in all MPS disorders with the exception of MPS II.

beta-galactosidase (β-galactosidase): Lysosomal enzyme deficient in MPS IV-B.

beta-glucuronidase (β-glucuronidase): Lysosomal enzyme deficient in MPS VII.

BiPAP: Bilevel positive airway pressure, often used for people with sleep apnea to open the airway during sleep. It uses 2 pressure settings, one for while the person is breathing in and the other for while the person is breathing out.

blood-brain barrier: The walls of the blood vessels of the brain (and the retina) are much more difficult for large molecules to pass through than are blood vessels elsewhere in the body. This has important implications for the ability of the body to mount an immune response and to provide protection to these tissues, although the reason for the difference is not well understood. The implications for human genetic disease are that it is far more difficult to provide therapeutic treatment to neural (brain) tissues than to other tissues in the body. Since many lysosomal storage diseases have a specific involvement in the neural tissues, it is critical to provide access to these tissues during treatment.

bone marrow transplant: See stem cell transplant.

bone marrow: Tissue found in the center of most bones. It is the site in which most blood cells are made, including red blood cells, which are involved in transport of oxygen in the blood, and white blood cells, which are involved in immune response.

cardiac arrhythmia: See arrhythmia.

cardiologist: A medical doctor who specializes in disorders of the heart and blood vessels.

cardiomyopathy: A general term referring to disease of, or damage to, the heart muscle.

carpal tunnel: The space between the carpal bones of the wrist and the connective tissue over the flexor tendons. The carpus (wrist) consists of 8 small bones known as carpals, which are joined by a band of fibrous proteins called ligaments. Nerves have to pass through the wrists in the space between the carpal bones and the ligaments.

carpal tunnel syndrome: Thickening of the ligaments in the carpal tunnel causing pressure on the nerves. This can cause irreversible nerve damage if not surgically corrected. In individuals with MPS VI, carpal tunnel syndrome often occurs because of accumulation of glycosaminoglycan (GAG) deposits.

carrier: An individual who has a recessive, disease-causing version of a gene at a particular site on one chromosome of a pair and a normal version of a gene at the same location on the other chromosome. By definition, carriers of a recessive condition do not have clinical signs and symptoms of the condition.

cell therapy: A therapy currently under investigation for MPS. This therapy involves implanting enzyme-producing muscle cells from another individual into an individual with MPS.

cerebrospinal fluid (CSF): The fluid that surrounds the brain and spinal cord and that is produced in the ventricles of the brain.

cervical spinal cord compression: Pressure on the neck area of the spinal cord from abnormal tissue growth nearby.

chorionic villus sampling (CVS): Prenatal diagnostic procedure involving sampling the chorionic villi (part of the amniotic sac, which surrounds the growing fetus), generally performed between the 10th and 12th weeks of pregnancy. The test can reveal many, but not all, genetic abnormalities. The decision to have prenatal testing, and the appropriate method of prenatal diagnosis, should be discussed with your healthcare provider.

chromosome: The linear, double-stranded structural unit of genetic material consisting of DNA and supporting proteins called chromatin. Human cells are expected to contain 46 chromosomes identified as 23 pairs; 22 pairs are autosomes and one pair are the sex chromosomes.

cognitive function: The ability to think, reason, remember, pay attention, use judgment, and have insight (understanding one's self and situation).

conductive deafness: A type of hearing loss that occurs when the transmission of sound through the outer and middle ear is blocked.

contracture: Muscle shortening resulting in loss of motion of the joint.

cord blood transplant: See stem cell transplant.

cornea: The transparent circular part of the front of the eye.

corneal clouding: Disruption of the clear layers of the cornea in individuals with certain types of MPS due to storage of glycosaminoglycans (GAG), causing a milky appearance of the eye, decreased vision, and sensitivity to light. Cloudy corneas can be replaced with a corneal transplant.

corneal transplant: Surgical procedure to remove a cloudy cornea (see corneal clouding) and replace with a healthy, donated cornea.

coronary arteries: A pair of blood vessels that supply the heart muscle with blood.

CPAP: Continuous positive airway pressure, often used for people with sleep apnea to open the airway during sleep using a constant pressure setting.

cranium: The part of the skeleton that encloses the brain.

deposits: See glycosaminoglycans (GAG).

dermatan sulfate: A type of glycosaminoglycan (GAG) that is not properly broken down in people with MPS II.

DNA: The molecule that encodes the genes responsible for the structure and function of an organism and allows for transmission of genetic information to the next generation.

dyspnea: Shortness of breath.

dysostosis: The abnormal formation of bone caused by the lack of proper ossification (conversion of cartilage or tissues into bone).

dysostosis multiplex: The abnormal formation of many bones caused by the lack of proper ossification.

echocardiogram: Ultrasound of the heart to evaluate for heart valve and heart muscle function.

electroencephalogram (EEG): A record of the electric potentials in the brain recorded by attaching electrodes on the scalp. Often this procedure is used to look for seizure activity.

electrocardiogram (EKG or ECG): A study of the currents in the heart that control its contraction.

electromyography (EMG): Continuous recording of the electrical activity of a muscle by means of electrodes inserted into the muscle fibres. Used, although not required, to diagnose carpal tunnel syndrome (which can be diagnosed by nerve conduction studies).

enamel: The hard outer covering of the crown of a tooth.

enzyme: A protein that facilitates a biological reaction without itself being used up in the reaction (i.e., it acts as a catalyst). An enzyme acts by binding with the substance involved in the reaction (the substrate) and converting it into another substance (the product of the reaction).

Eustachian tube: The tube from the middle ear to the throat that equalizes pressure in the ear, drains secretions from the ear, and protects the ear from mucus in the nose and throat.

fontanelle: A soft spot on a baby's head.

galactose 6-sulfatase: Lysosomal enzyme deficient in MPS IV-A.

gastroenterologist: A medical doctor who specializes in disorders affecting the stomach and intestines.

gastrostomy: A surgical procedure in which an opening is made into the stomach from the outside. It is usually performed to allow nutrition and/or medications to be given directly into the stomach through a small flexible tube (the **G-tube**). This may be done when swallowing is difficult because of disease or obstruction of the esophagus (the tube from the mouth to the stomach).

gene: Basic unit of heredity that codes for a specific protein leading to a particular characteristic or function – for example, details of physical appearance or organ function.

gene therapy: A therapeutic approach to a genetic disorder whereby a corrected copy of the gene or a new gene is inserted to replace the incorrect version.

genetic code: Information carried by the DNA molecules that decides the physical traits of an offspring. The code fixes the pattern of amino acids that build body tissue proteins within a cell.

genetic skeletal survey: See skeletal survey.

genu valgum: Knock-knees (knees curving inward in relation to the thigh).

gibbus: Abnormal angular curve of the vertebrae of the spine (synonym: kyphosis).

glaucoma: A condition in which loss of vision occurs because of an abnormally high pressure in the eye.

glycosaminoglycan (GAG): A long repeating chain of complex carbohydrate (sugar) molecules that is a common constituent of secretions and the connective tissue between cells. Formerly called mucopolysaccharide.

G-tube: See gastrostomy.

heart failure: A condition where the heart cannot pump enough blood to meet the needs of the body.

hematopoietic stem cell transplant: Also called HSCT. See stem cell transplant.

heparan sulfate: A type of glycosaminoglycan (GAG) that is not properly broken down in people with MPS II.

heparan N-sulfatase: Lysosomal enzyme deficient in MPS III-A.

hepatomegaly: Enlargement of the liver.

hepatosplenomegaly: Enlargement of the liver and the spleen. (Hepatomegaly: enlargement of the liver; splenomegaly: enlargement of the spleen.)

hernia: Protrusion of a part or structure (e.g., a loop of the small intestine) through the tissues normally holding it in.

heterogeneity: Variations in clinical features (characteristics) within a specific disease.

heterozygote: An individual possessing a variant gene and a normal gene at identical sites of homologous chromosomes (adjective: heterozygous).

hip dysplasia: Abnormal hip formation.

homologous chromosomes: A pair of chromosomes, one from each parent, having the same gene loci (locations) in the same order.

homozygote: An individual possessing a pair of identical genes, either both normal or both variant, at identical sites on homologous chromosomes.

Hurler-Scheie syndrome: Historical term for less-severe MPS I, the part of the clinical spectrum that is intermediate between Hurler and Scheie syndromes.

Hurler syndrome: Historical term for the severe end of the clinical spectrum of MPS I.

Hunter syndrome: See MPS II.

hyaluronidase: Lysosomal enzyme deficient in MPS IX.

hydrocephalus: An abnormal increase in the amount of cerebrospinal fluid within the ventricles of the brain. Communicating hydrocephalus may be caused by failure to resorb cerebrospinal fluid into the cerebral sinuses. It can be treated using a ventriculoperitoneal shunt.

hypoxia: A deficiency of oxygen in the tissue or blood.

inguinal hernia: Hernia occurring in the lower abdomen and groin.

I-cell disease: See ML II.

iduronate-2-sulfatase (I2S): Lysosomal enzyme deficient in MPS II.

individual educational program (IEP): A program designed for each child within the public school system who receives special educational services. Its goals are to improve teaching, learning, and appropriate goal setting for each individual. Often, a team including members from the school system and the family are involved in designing the IEP.

intubation: The placement of a breathing tube during anesthesia.

jejunostomy: A surgical procedure in which an opening is made into the small intestine from the outside. It is usually performed to allow nutrition and/or medications to be given directly into the small intestine through a small flexible tube (the **J-tube**). This may be done if swallowing is difficult because of disease or obstruction of the esophagus (the tube from the mouth to the stomach).

joint contracture: Fibrosis of a muscle tissue producing shrinkage and shortening of the muscle without generating any strength. It is usually a consequence of pain in or disuse of a muscle or limb.

J-tube: See jejunostomy.

kyphosis: Abnormal angular curve of the vertebrae of the spine (synonym: gibbus).

left ventricular aneurysm: A weakened outpouching of the wall of the heart's main pumping chamber.

lumbar puncture: A procedure in which cerebrospinal fluid is withdrawn by means of a needle inserted into the membrane space in the region of the lower back. This procedure may be performed to measure intracranial pressure (pressure inside the head) to aid in diagnosing hydrocephalus.

lysosomal enzyme: A protein found within the cytoplasm of most cells, especially leukocytes (white blood cells), kidney cells, and liver cells. It is a key component in the function of digestive processes within the cell.

lysosomal storage disorder (LSD): An inborn error of metabolism resulting in a particular lysosomal enzyme deficiency. At this time there are more than 40 identifiable lysosomal storage disorders.

lysosome: A specialized compartment (organelle) in the cytoplasm of cells that contains enzymes responsible for breaking down substances in the cell.

macrocephaly: An abnormally large head.

melatonin: A compound involved in circadian rhythms (biological variations during a 24-hour period). It is sometimes used as a sleep aid for those with MPS disorders.

Maroteaux-Lamy syndrome: See MPS VI.

menarche: A woman's first menstrual period.

mitral valve prolapse: A condition where flaps between 2 parts of the heart, the left atrium and the left ventricle, don't close evenly, allowing a small amount of blood to leak back into the left atrium.

mixed deafness: A type of hearing loss that is due to a combination of conductive deafness and sensorineural deafness.

ML II: Also called **I-cell disease**. Caused by a deficiency of the lysosomal enzyme N-acetylglucosaminyl-1-phosphotransferase. An autosomal recessive disease characterized by severe psychomotor developmental delay and by many of the clinical features seen in severe MPS I.

ML III: Also called **pseudo-Hurler polydystrophy**. Caused by a deficiency of the lysosomal enzyme N-acetylglucosaminyl-1-phosphotransferase. An autosomal recessive disease with a less severe disease course than ML II, becoming apparent later in life, with survival into adulthood.

Morquio syndrome: See MPS IV.

MPS I: Historically called **Hurler, Hurler-Scheie, and Scheie syndromes**. Caused by a deficiency of the lysosomal enzyme alpha-L-iduronidase. An autosomal recessive, heterogeneous disease characterized by a wide range of clinical involvement, including corneal clouding, bone changes, stiff joints, large liver and spleen, and heart disease.

MPS II: Also called **Hunter syndrome**. Caused by a deficiency of the lysosomal enzyme iduronate sulfatase. An X-linked recessive, heterogeneous disease characterized by a wide range of clinical involvement, including large liver and spleen, stiff joints, bone changes, and heart disease. There is a broad spectrum of disease severity.

MPS III: Also called **Sanfilippo syndrome**. An autosomal recessive disease classified into 4 types based on the enzyme deficiency. The features in each type are similar and characterized by severe central nervous system degeneration but only mild somatic (body-related) problems.

MPS III-A: Caused by a deficiency of the lysosomal enzyme heparan N-sulfatase.

MPS III-B: Caused by a deficiency of the lysosomal enzyme alpha-N-acetylglucosaminidase.

MPS III-C: Caused by a deficiency of the lysosomal enzyme acetyl CoA: alpha-glucosaminide acetyltransferase.

MPS III-D: Caused by a deficiency of the lysosomal enzyme N-acetyl glucosamine 6-sulfatase.

MPS IV: Also called **Morquio syndrome**. An autosomal recessive disease classified into 2 types based on the enzyme deficiency, each with a wide range of clinical manifestations. Both types are characterized by short-trunk dwarfism, fine corneal deposits, and preservation of cognitive function.

MPS IV-A: Caused by a deficiency of the lysosomal enzyme galactose 6-sulfatase.

MPS IV-B: Caused by a deficiency of the lysosomal enzyme beta-galactosidase.

MPS VI: Also called **Maroteaux-Lamy syndrome**. Caused by a deficiency of the lysosomal enzyme arylsulfatase B. An autosomal recessive heterogeneous disease characterized by a wide range of clinical involvement, including corneal clouding, bone abnormalities, stiff joints, large liver, heart disease, and normal cognitive function.

MPS VII: Also called **Sly syndrome**. Caused by a deficiency of the lysosomal enzyme beta-glucuronidase. An autosomal recessive disease characterized by large liver and spleen, bone abnormalities, and a wide spectrum of severity.

MPS IX: An autosomal recessive disease caused by a deficiency of the lysosomal enzyme hyaluronidase, characterized by short stature, soft-tissue masses, normal joint movement, and normal cognitive function.

mucolipidosis: Term coined to denote diseases that combined clinical features common to both the mucopolysaccharidoses and the sphingolipidoses (diseases characterized by abnormal lipid or fat metabolism, affecting nerve tissue). See ML II and ML III.

mucopolysaccharide: A complex carbohydrate molecule that is a common constituent of secretions and the connective tissue between cells. Although the molecules were originally called "mucopolysaccharides" because of their ability to form thick, mucous-like solutions, the terminology was revised to "proteoglycans" and subsequently to "glycosaminoglycans" or GAGs in recent decades.

mutation: A change in the genetic material (DNA) of a cell that alters expected genetic processes.

N-acetyl-α-glucosamine 6-sulfatase: Lysosomal enzyme deficient in MPS III-D.

N-acetylglucosaminyl-1-phosphotransferase: Lysosomal enzyme deficient in ML II/III.

odontoid dysplasia: Malformation in the bones that stabilize the connection between head and neck.

odontoid process: A bone found in the neck that is important for keeping the neck stable.

ophthalmologist: A medical doctor who specializes in disorders affecting the eye.

otitis media: Inflammation of the middle ear occurring commonly in children as a result of an infection and often causing pain and temporary hearing loss.

otolaryngologist: A medical doctor who specializes in disorders affecting the ear, nose, and throat.

papilledema: Swelling around the optic disc (the "blind spot" where the optical nerve joins the eye).

PEG tube: Also known as a *percutaneous endoscopic gastrostomy tube.* This is a type of G-tube that is inserted into the stomach by using an endoscopic procedure (which uses a small camera on the end of a flexible tube to see inside the body).

polysomnogram: Also called a sleep study. This test is done to check for sleep problems such as sleep apnea. During the test, the person sleeps overnight at a sleep centre or hospital while his or her sleep patterns are observed and recorded using special monitors and video cameras.

Port-a-Cath®: Brand name for a long-term indwelling catheter into a central vein with access through the skin.

posterior: Back.

precocious puberty: The early onset of sexual maturation.

preimplantation genetic diagnosis (PGD): Also known as preimplantation testing. A procedure used to decrease the chance of a particular genetic condition for which a fetus is specifically at risk by testing one cell from embryos from in vitro fertilization for the DNA mutation known in the family. Only embryos found not to carry the DNA mutation are transferred to the mother's uterus.

pseudo-Hurler polydystrophy: See ML III.

psychomotor performance: The ability to coordinate mental activity with physical activity. For example, catching a ball, driving, and dancing are all examples of activities requiring good psychomotor performance."

pulmonary: Of the lung, or relating to the lung.

pulmonary hypertension: High blood pressure in the blood vessels supplying the lungs.

pulmonologist: A medical doctor who specializes in disorders affecting the lungs.

recessive disease: See autosomal recessive disease and X-linked recessive disease.

recombinant DNA: DNA that contains genes from different sources that have been combined by the techniques of genetic engineering.

retina: A part of the eye that turns light into nerve signals for the brain, allowing your brain to process the images that you see.

retinal: Of the retina, or relating to the retina.

retrosternal pain: Pain behind the chest area.

rhinorrhea: Thick, chronic discharge of mucus from the nose.

scaphocephalic: Having a long, narrow head shape.

Sanfilippo syndrome: See MPS III.

Scheie syndrome: Historical term for the less severe (attenuated) end of the clinical spectrum of MPS I.

scoliosis: Lateral (sideways) deviation of the spine.

seizures: Disruption of electrical signals in the brain. Seizures may cause brief changes in a person's body movements, awareness, emotions, or senses such as taste, smell, vision, or hearing.

sensorineural deafness: A type of hearing loss that occurs from damage to the sensory hair cells in the inner ear or to the nerves that transmit sound to the brain. In MPS, a common cause of sensorineural deafness is damage to the tiny hair cells in the inner ear.

sinusitis: A sinus infection. Sinus infections are common in people with MPS disorders.

skeletal survey: An X-ray to examine the skeleton (the bones of the body) for problems or abnormalities.

sleep apnea: A temporary cessation of breathing during sleep, generally caused by obstruction of the airway.

Sly syndrome: See MPS VII.

spinal cord compression: See cervical spinal cord compression.

spinal fusion: Surgery to connect the spinal bones to each other to prevent slippage.

spleen: A large organ situated on the left side of the body below and behind the stomach.

splenomegaly: Enlargement of the spleen.

stature: A person's height. People with MPS disorders often have short stature.

stem cell transplant: A therapeutic treatment where stem cells from bone marrow or umbilical cord blood are infused into the bloodstream after the original bone marrow cells have been ablated (destroyed) by chemotherapy and/or radiation therapy. The purpose is to allow the donor stem cells to re-populate the bone marrow and various other tissues of the recipient. If the cells can also provide the missing gene and function to the recipient, then clinical symptoms can sometimes improve. It is important to note that the process of destroying the recipient's bone marrow cells is extremely invasive and leaves the individual with a compromised immune system and susceptible to life-threatening infections. Also, it is critical to have donor cells come from an individual with compatible tissue types in order to avoid rejection of the donor cells after the transplant.

stem cell: A cell whose "daughter" cells have the potential to develop into a variety of specialized cell types.

sternum: A long, flat bone, jointed with the cartilages of the first 7 ribs and with the clavicle (collar bone), forming the middle part of the anterior (front) wall of the thorax (chest area). It is the vertical bone that can be felt in the middle of the chest between the ribs.

stoma: The surgical opening for a G-tube or J-tube.

substrate deprivation therapy: A potential treatment method for MPS that works by slowing down the production of glycosaminoglycans in order to reduce the rate at which they build up in lysosomes.

swallowing study (modified barium swallow study): Videotaped X-ray of a individual's oral (mouth) and pharyngeal (throat) mechanism during eating or drinking. This procedure is often ordered to evaluate for obstruction or aspiration (inhalation of foods). The results from this procedure may allow a therapist to better identify ways to safely feed the individual and ways to help the family make appropriate modifications.

trachea: The air tube from the mouth to the lungs. Around the level of the middle of the chest, it divides into the right and left main bronchi.

tracheostomy: A surgical procedure in which a hole is made into the trachea through the neck to relieve obstruction to breathing. A curved breathing tube is usually inserted through the hole.

transverse atlantal ligament: A strong tissue band that helps hold the odontoid process, an important bone for stabilizing the neck, in place.

trigger finger: A condition caused by a thickening of the tendon that bends the fingers, often experienced as swelling in the palm of the hand as the finger is moved.

tympanostomy: A surgical procedure in which a small incision (cut) is made in the eardrum. Tiny tubes (called tympanostomy tubes or ear tubes) can be inserted into the incision to allow fluid in the middle ear to drain.

umbilical hernia: A hernia in which bowel or connecting tissue protrudes through the abdominal wall under the skin at the umbilicus (navel).

urinary: Of the urine, or found in the urine.

urticaria: Hives.

ventriculoperitoneal shunt: A thin tube that drains fluid from the brain into the abdominal cavity. Used in the treatment and management of hydrocephalus.

X-linked recessive disease: A disease that follows a pattern of inheritance in which a mutation in a gene on the X chromosome causes males to have clinical features of a particular condition, as they only have one X chromosome. A pattern of inheritance seen in MPS II.

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Updates and new information

Medical professionals and researchers are constantly learning new things about MPS II disease and its management. Some of the support resources, websites, and laws mentioned in this guide may change over time.

Contact one of the following organizations to keep up to date on the latest information on MPS II and its management:

The Canadian Society for Mucopolysaccharide & Related Diseases (The Canadian MPS Society):

- 1-800-667-1846
- www.mpssociety.ca

Shire HGT:

- Paladin Labs Inc. at 1-888-550-6060
- Shire HGT Reimbursement Support Program at 1-866-773-6302