The National MPS Society

Families Joining Together

Common bonds unite the lives of those affected by mucopolysaccharidoses (MPS) and mucolipidoses (ML) disorders – the need for support and the hope for a treatment.

The National MPS Society is committed to making a difference through support, research, education and advocacy. Families from around the world gain a better understanding of these rare genetically-determined disorders through the Society's help in linking them with health care professionals, researchers, and perhaps most importantly, each other.

Individuals affected with MPS and ML and their families have a resource. One that stands ready to help — one resource that takes an active role in fostering the courage necessary to confront these disorders every day.

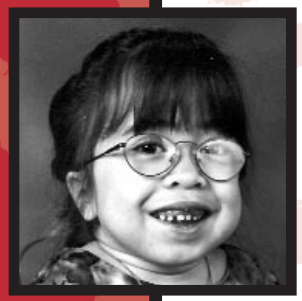
Join the National MPS Society and enjoy a variety of benefits, including:

- *Courage, our quarterly newsletter that shares stories and information about people with MPS or ML.*
- *News about various National MPS Society-sponsored conferences and gatherings, where families and leading MPS and ML scientists, physicians and researchers are brought together.*
- *Information on local events like regional picnics and fundraisers. Opportunities for families to meet each other and help raise community awareness of these rare genetic diseases.*
- *A listing in our annual directory of members, which assists families in locating each other.*

For more information or to join the National MPS Society, please visit:
www.mpssociety.org
or contact us by e-mail at
info@mpssociety.org

MPS

The National MPS Society, Inc.



A Guide to Understanding

Maroteaux-Lamy Syndrome

Mucopolysaccharidosis (MPS) VI

MPS

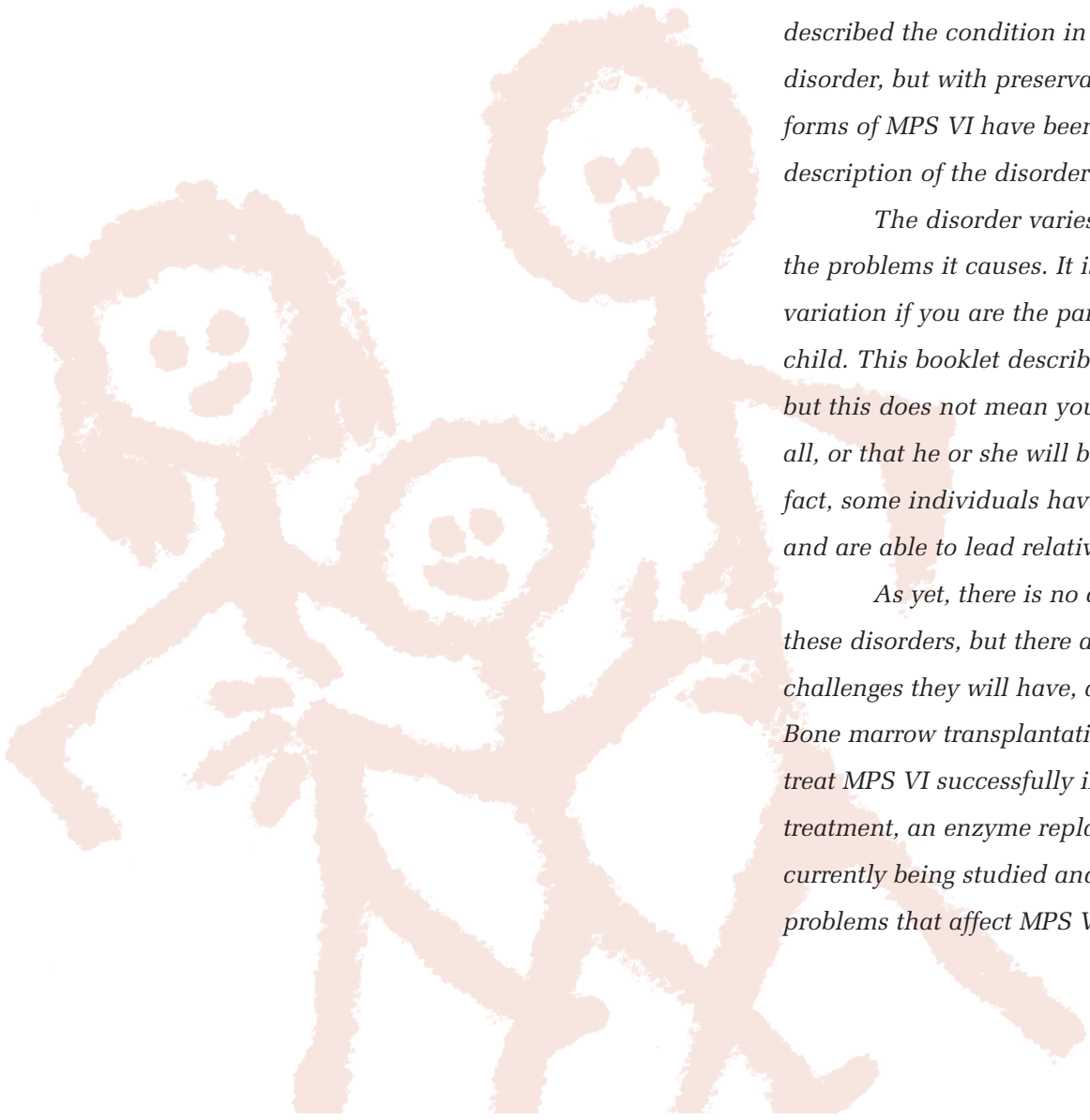
The National MPS Society, Inc.

Introduction

Maroteaux-Lamy syndrome, also known as Mucopolysaccharidosis VI (MPS VI), is one of the rarer mucopolysaccharide disorders. It takes its name from two French doctors, Dr. Maroteaux and Dr. Lamy, who first described the condition in 1963 as a Hurler-like (MPS I) disorder, but with preservation of intelligence. Milder forms of MPS VI have been recognized since the initial description of the disorder.

The disorder varies enormously in the severity of the problems it causes. It is important to remember this variation if you are the parent of a newly diagnosed child. This booklet describes all the possible problems, but this does not mean your child will experience them all, or that he or she will be severely affected by them. In fact, some individuals have very few physical problems and are able to lead relatively normal lives.

As yet, there is no cure for individuals affected by these disorders, but there are ways to manage the challenges they will have, and to help them enjoy life. Bone marrow transplantation (BMT) has been used to treat MPS VI successfully in some patients. Another treatment, an enzyme replacement therapy (ERT), is currently being studied and may help some of the problems that affect MPS VI individuals. However, ERT is



front cover
top: Kaitlyn, age 6
middle: Autumn, age 6
bottom: Melanie, age 37

not yet available to MPS VI individuals. Scientists who study MPS continue to look for better and more effective ways to treat these disorders, and it is likely that patients will have more options available to them in the future.

This booklet is intended as an introduction into the nature of the disorder, as well as to help families understand more about what is happening to those with MPS VI and what they can do to manage it. In April 2000, this booklet was updated by the National MPS Society with help from experts in the field and MPS/ML parents to provide families with the latest information.

What causes these disorders?

Mucopolysaccharides are long chains of sugar molecules used in the building of bones, cartilage, skin, tendons and many other tissues in the body. They 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 mucopolysaccharides. The rubbery resilient cartilage in your joints is another example. All tissues have some of this substance as a normal part of their structure.

The more modern word for mucopoly-saccharides is glycosaminoglycans or GAG, which stands for the sugar-amino-sugar polymer or long repeating sugar chains found in these materials. These sugar chains are submicroscopic and cannot be seen with the eye, but can be studied using special scientific instruments and analytical methods.

To understand how GAG accumulate and cause MPS VI, it is important to understand that in the course of normal life, there is a continuous process of building new mucopoly-saccharides and breaking down old ones – a recycling process. This ongoing process is required to keep your body healthy. The breakdown and recycling process requires a series of special biochemical tools called enzymes. To break down GAG, a series of enzymes or tools work in sequence one after another. The GAG chain is broken down by removing one sugar molecule at a time starting at one end of the GAG chain. 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

The word "mucopolysaccharide" can be broken down into its parts to help understand it:
muco-poly-saccharide
muco refers to the thick jellylike consistency of the molecules;
poly means many;
saccharide is a general term for a sugar molecule (think of saccharin).

works on nails. Individuals with MPS VI are missing one specific enzyme called arylsulfatase B, which is essential in the breakdown of certain GAG called dermatan sulfate. The incompletely broken down dermatan sulfate remains stored inside cells in the body and begins to build up, causing progressive damage. The GAG itself is not toxic, but the amount of it and the effect of storing it in the body lead to many physical problems. Babies may show little sign of the disorder, but as more and more GAG accumulate, symptoms start to appear. Sugar or other foods normally eaten will not affect whether there is more or less buildup of GAG.

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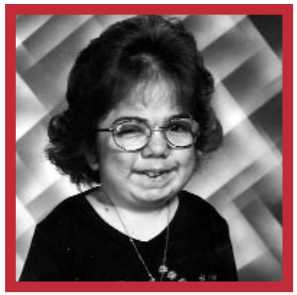
How common are the disorders?

It has been estimated that about 1 in 215,000 births are affected by MPS VI. Though this disorder is rare, each patient needs such extensive medical care that the effect on the medical system is much larger than their numbers suggest.



Jessica, age 15

How are the disorders inherited?

When most people think of genetic disease, they think of a health problem that gets passed down from father or mother to child and so on. While many genetic diseases are passed down the generations in an obvious way, some genetic diseases are "hidden," or recessive and only show up when both genes in an individual are affected. MPS VI is one such disease. Most families with an MPS VI child do not have a family history of any genetic problem – MPS VI seems to show up suddenly.

To understand this better, it is important to understand some basics of genetics. All humans are formed with two complete sets of genes, one set from each parent. So any individual has half his genes from his mother and half from his father. Together, the individual has 100% of the genes required to live.

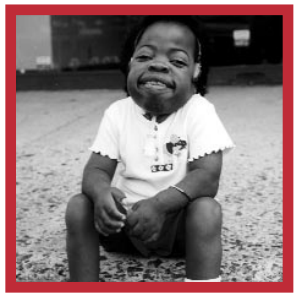
For each enzyme made in the body, there are two genes for it, one from the mother and one from the father. For most enzymes, if only one of the genes actually works, the nearly 50% level of enzyme is more than enough to keep the person healthy. Basically, half as much enzyme can do twice the usual amount of work. However, if both genes for the enzyme from mother and father are not functioning correctly, the individual will have little or no enzyme and then will suffer from the disease. Disease occurs only when both genes from mother and father are not working right, or are recessive. This means it is hidden until an individual inherits two genes for the same enzyme that are not working.

Because the parents of the child with MPS VI each have another gene that does work, there is a 3 out of 4 chance that a repeat pregnancy will result in a child with at least one normal gene and no disease. There is also a 1 in 4 chance with every pregnancy that the child will inherit the defective gene from each parent and will be affected with the disorder. There is a 2 in 3 chance that unaffected brothers and sisters of MPS VI individuals will be carriers. Carriers have one good gene and one defective gene. In general, the disorder is so rare that the likelihood of one carrier marrying another carrier is very low.

All families of affected individuals should seek further information from their medical genetics doctor or from a genetic counselor if they have questions about the risk for recurrence of the disease in their family or other questions related to inheritance of MPS disorders.

Prenatal diagnosis

If you already have a child with MPS VI, it is possible to have tests during a subsequent pregnancy to find out whether the baby you are carrying is affected. It is important to consult your doctor early in the pregnancy if you wish tests to be arranged. Both amniocentesis and chorionic villus sampling can be done to diagnose MPS VI in utero.



Karima, age 11

Clinical problems in MPS VI

Growth

Growth in height is usually significantly less than normal, but varies according to the severity of the disorder. Individuals with the most severe form of MPS VI may not grow taller than 3½ feet. Adults with a milder form of Maroteaux-Lamy syndrome will usually be smaller than normal, reaching a height of between 3 to 5 feet.

Intelligence

Intelligence is not affected by Maroteaux-Lamy syndrome – many children are of above average ability.

Physical appearance

Maroteaux-Lamy individuals look remarkably similar due to large heads, short necks, chubby cheeks, broad noses with a flat bridge, and wide nostrils. The shoulders are narrow and rounded and the stomach tends to protrude. The hair on the body is coarser and more abundant than usual, and the eyebrows are bushy. The skin may become thickened and less elastic than usual.

Nose, throat, chest and ear problems

Throat

The tonsils and adenoids often become enlarged and can partly block the airway. The neck is usually short, which contributes to problems in breathing. The windpipe (trachea) becomes narrowed by storage material and

may be more floppy, or softer that usual, due to abnormal cartilage rings in the trachea. Nodules or excess undulations of tissue can further block the airway.

Chest

The shape of the chest is abnormal and the junction between the ribs and the breastbone (sternum) is not as flexible as it should be. The chest is therefore rigid and cannot move freely to allow the lungs to take in a large volume of air. The muscle at the base of the chest (diaphragm) is pushed upwards by the enlarged liver and spleen, further reducing the space for the lungs. When the lungs are not fully cleared, there is an increased risk of infection (pneumonia).

Breathing difficulties

Many affected individuals breathe very noisily, even when there is no infection. At night they may be restless and snore. Sometimes the individual may stop breathing for short periods while asleep (sleep apnea). Pauses of up to 10-15 seconds may be considered normal. This noisy breathing, which stops and starts, can be very frightening for parents to hear. They may fear that their child is dying. If this is happening, the child's oxygen level may be low when sleeping, and can cause problems with the heart. If a parent notices significant choking or episodes of interrupted breathing, the child should be evaluated by a sleep specialist using a polysomnogram. It is important to know that many individuals may breathe like this for years. Sleep apnea can be treated in some patients by removing the tonsils and adenoids (adenoids may regrow), opening up the airway with nighttime CPAP treatment (continuous positive airway pressure), BiPAP (bilevel positive airway pressure), or tracheostomy, as discussed in the follow paragraphs.

The doctor may want the 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 monitors of the body's function. From this study, the doctors can assess how much blockage to breathing is present, how much trouble your child is having moving air into the lungs during sleep, and how much effect this has on the body.

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

Nighttime CPAPor BiPAP can open the airway up at night using air pressure, which can help the child's airway stay open. This treatment involves placing a mask on the face each night and having air pumped into the airway to keep it from collapsing. This may seem to be an extreme measure, but many people are able to accept it because it can greatly improve the quality of sleep, as well as help prevent or reduce the risk of heart failure caused by low oxygen levels at night.

In severe cases of sleep apnea with heart failure, a tracheostomy (a hole into the airway made in the front of the neck) may be needed. Most families will try to avoid a tracheostomy because it is so invasive and seemingly destructive of the child's normal function. In fact, many doctors feel that MPS individuals should receive a trach-eostomy earlier than they do, and many do much better after improving their nighttime breathing.

Chest postural drainage can be helpful in clearing secretions from the lungs. The physiotherapist will be able to teach the parents and someone at the child's school how to do this.

Treatment of respiratory infections

Drugs may affect people with MPS differently, so it is essential to consult your doctor rather than using over-the-counter medications. Drugs for controlling mucous production may not help. Drugs, such as antihistamines, may dry out the mucous, making it thicker and harder to dislodge. Decongestants usually contain stimulants that can raise blood pressure and narrow blood vessels, both undesirable for people with MPS. Cough suppressants or drugs that are too sedating may cause more problems with sleep apnea by depressing muscle tone and respiration.

Although most normal individuals with colds do not require antibiotics, individuals with MPS VI may end up with secondary bacterial infections of the sinuses or middle ear. These infections should be treated with antibiotics. Poor drainage of the sinuses and middle ear make overcoming infections difficult. Therefore, it is common to have infections improve on antibiotics and then promptly recur after the antibiotic course is over. Chronic antibiotic therapy can be used to help some individuals with recurring ear infections. Ventilation tubes can be used to improve drainage from the ear and speed resolution of infections. It is important to consult with an Ear, Nose and Throat (ENT) specialist experienced with MPS disorders to determine which tube is best.

Many people with MPS VI become allergic to antibiotics or may acquire resistant infections. Your doctor can prescribe other antibiotics to help manage this problem. While overusing antibiotics is not advised, most people with MPS will require some treatment for most infections. You will need a doctor with whom you can develop a good working relationship to manage the frequent infections.

Mouth

Individuals with MPS generally have thick lips and an enlarged tongue. Gum ridges are broad. The teeth may be widely spaced and poorly formed with fragile enamel. It is important that the teeth are well cared for, as tooth decay can be a cause of pain. Teeth should be cleaned regularly, and if the water in your area has not been treated with fluoride, the child should have daily fluoride tablets or drops. Cleaning inside the mouth with a small sponge on a stick soaked in mouthwash will help keep the mouth fresh and help avoid bad breath. Even with the best dental care, an abscess around a tooth can develop due to abnormal formation of the tooth. Irritability, crying, and restlessness can sometimes be the only sign of an infected tooth in a severely involved individual.

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If an MPS VI individual has a heart problem, it is advised that antibiotics be given before and after any dental treatment. This is because certain bacteria in the mouth may get into the bloodstream and cause an infection in the abnormal heart valve, potentially damaging it further. 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.

Heart

Heart disease can occur in people with Maroteaux-Lamy syndrome, but may not develop or cause any real problems until later in the individual's life. Medications are available to help manage the heart problems that occur in MPS. Cardiomyopathy (weak heart muscle) and endocardiofibroelastosis (stiff heart) are conditions that can occur occasionally in individuals with the severe form of MPS VI. Most people with MPS VI have some degree of heart valve leakage or blockage. Some individuals with Maroteaux-Lamy syndrome may develop heart failure and have problems with the aortic or mitral valve. They may have slowly progressive heart disease for years without any apparent clinical effects. If the condition worsens, an operation may be needed to replace the damaged valves.

Your doctor may hear heart murmurs (sounds caused by turbulence in blood flow in the heart) if the valves become damaged by stored mucopolysaccharides. The heart valves are designed to close tightly as blood passes from one chamber of the heart to another in order to stop blood from flowing back in the wrong direction. If a valve is weakened, it may not shut firmly enough and a small amount of blood may shoot backwards, leading to turbulence and a murmur. Most people with MPS VI have some degree of murmur or leakage.

As heart problems occur frequently in MPS VI, individuals should have a test known as an echocardiogram annually (or as often as your doctor thinks necessary) to show whether any problems are beginning. The test is painless, and similar to the ultrasound screening of babies in the womb. It can identify problems with the heart muscle, heart function, and heart valves, but like many tests, it cannot detect all possible problems.

In individuals who are severely affected, the muscle of the heart may be damaged by GAG storage (cardiomyopathy). The heart may also be put under strain by having to pump blood through abnormal lungs (corpulmonale or right heart failure). A number of affected individuals have high blood pressure.

Because of the unusual special problems that can occur in these disorders, you should select a cardiologist with some knowledge of MPS VI. At a minimum, you should inform the doctor about heart problems experienced by MPS VI individuals.

Liver and spleen

In most MPS VI individuals, both liver and spleen become enlarged by storage of mucopolysaccharides (hepatosplenomegaly). The enlarged liver does not usually cause liver problems or lead to liver failure, but it can interfere with eating and breathing.

Abdomen and hernias

In people with MPS VI, the abdomen bulges out due to posture, weakness of the muscles and the enlarged liver and spleen. Frequently, part of the abdominal contents will push out behind a weak spot in the wall of the abdomen. This is called a hernia. The hernia can come from behind the navel (umbilical hernia) or in the groin (inguinal hernia).

Inguinal hernias should be repaired by an operation, but hernias will sometimes recur. Umbilical hernias are not usually treated unless they are small and cause entrapment of the intestine, or are very large and are causing problems. It is very common to have a reoccurrence of an umbilical hernia after a repair has been made.

Bowel problems

Many Maroteaux-Lamy individuals suffer periodically from loose stools and diarrhea. The cause of this is not fully understood. Occasionally the problem is caused by severe constipation and leakage of loose stools from behind the solid mass of feces. More often, however, parents describe it as "coming straight through." It is thought that there may be a defect in the autonomic nervous system, the system that controls those bodily functions usually beyond voluntary control. Studies have found storage in the nerve cells of the intestine and it seems likely that abnormal motility in the bowel is the cause of the diarrhea.

An examination by your pediatrician, supplemented by an X-ray if necessary, may establish which is the cause. The problem may disappear as the child gets older, but it can be made worse by antibiotics prescribed for other problems. The episodic diarrhea in some MPS individuals appears to be affected by diet; elimination of some foods can be helpful.

If antibiotics have caused the diarrhea, eating plain live-culture yogurt is often helpful during episodes of diarrhea. This provides a source of lactobacillus to help prevent the growth of harmful organisms within the bowel wall, which can cause diarrhea or make it worse. A diet low in roughage may also be helpful.

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

Bones and joints

People with MPS VI tend to have significant problems with bone formation and growth. This leads to bone problems (called dysostosis multiplex) as well as neurological problems if nerves get compressed by bone.

Spine

The bones of the spine (vertebrae) normally line up from the neck to the buttocks. Individuals with the severe form of MPS VI often have poorly formed vertebrae that may not stably interact with each other. One or two of the vertebrae in the middle of the back are sometimes slightly smaller than the rest and set back in line. This backward slippage of the vertebrae can cause an angular curve (kyphosis or gibbus) to develop, but it usually does not need treatment. In the mild form of MPS VI spinal cord compression is common. The compression is due to accumulation of GAG in the membrane surrounding the spinal cord.

Neck

The neck is short and sometimes restricted in movement. The bones that stabilize the connection between head and neck can be

malformed (odontoid dysplasia) in people with the severe form of MPS VI, making the neck unstable. Fusion surgery is required to connect all the bones to each other so they do not slip further. If severe pain or pain associated with weakness or tremors in the lower legs occur, the child should have studies of the neck (MRI and flexion-extension X-rays) to evaluate for slippage of the neck vertebrae.

Scoliosis

Abnormal curvature of the spine, or scoliosis, can also occur and if severe, may require intervention. In general, fusion with bone is the best alternative and hardware-like rods are not well tolerated. In any case, the soft bone makes the surgery and recovery difficult. Many patients need multiple procedures.

Joints

Joint stiffness is common in all forms of MPS, and the maximum range of movement of all joints may become limited. Later in the individual's life, joint stiffness may cause pain, which may be relieved by warmth and ordinary painkillers. The limited movement in the shoulders and arms may make dressing difficult. Anti-inflammatory drugs, such as ibuprofen, can help with joint pain, but their use should be monitored closely to make sure that irritation and ulcers in the stomach do not occur.

Hands

The shape of the hands is very noticeable and has been used as the symbol of the National MPS Society. The hands are short and broad with stubby fingers. The fingers stiffen and gradually become curved due to limited joint movement. The tips of the fingers can become permanently bent over.

Legs and feet

Many people with MPS VI stand and walk with their knees and hips flexed. This, combined with a tight Achilles tendon, may cause them to walk on their toes. They sometimes have knock-knees but this is very unlikely to need treatment. Severe knock-knees can be treated by surgery on the tibia bones, but this is not common in MPS VI. The feet are broad and may be stiff with the toes curled under, rather like the hands.

Skin

People with MPS VI tend to have thickened and tough skin, making it difficult to draw blood or place intravenous catheters. Excess hair on the face and back occurs in some people with Maroteaux-Lamy syndrome. Sweating and cold hands and feet are also common problems, and are possibly related to the heart, circulation, or other mechanisms that control temperature regulation. Periodic blue or cold hands or feet should be evaluated by a cardiologist to determine if the heart or the aorta might be responsible for the problem.

Neurological problems: brain, senses and nerves

Brain

The brain and the spinal cord are protected from jolting by the cerebrospinal fluid that circulates around them. In some people with MPS VI, the circulation of the fluid can slowly (over months to years) become blocked so that it cannot be taken back into the bloodstream. The blockage (communicating hydrocephalus) causes increased pressure inside the head, which can press on the brain and cause headaches and delayed development. If hydrocephalus is

suspected, an imagining study of the brain (CT or MRI scan) should be performed. A lumbar puncture with pressure measurement is another way to assess if hydrocephalus exists. If the doctor confirms that your child has communicating hydrocephalus, it can be treated by the insertion of a thin tube (shunt), which drains fluid from the brain into the abdomen (ventriculo-peritoneal or VP shunt). The shunt has a pressure-sensitive valve, which allows spinal fluid to be drained to the abdomen when the pressure around the brain becomes too high. The lack of papilledema (swelling around the optic disk) does not rule out hydrocephalus in an MPS child. Communicating hydrocephalus is more likely to occur in a child with severe MPS VI.

Eyes

The eye problems described are common to all forms of the MPS VI disorder. The circular window at the front of the eye (cornea) becomes cloudy due to storage of GAG, which disrupts the clear layers of the cornea. If corneal clouding is severe it may reduce sight, especially in dim light. Some MPS VI individuals cannot tolerate bright lights as the clouding causes uneven refraction of the light. Wearing caps with visors or sunglasses can help. Many people with MPS VI have had a corneal transplant, usually resulting in improved vision.

There may be problems with vision caused by changes to the retina or glaucoma (increased pressure) which should be checked during an eye examination. It is often difficult to determine which combination of problems is responsible for the decrease in eyesight. An ophthalmologist can perform special studies to help determine whether the problem is due to an effect on how light gets in the eye (the cornea) or on how the eye responds to light (the retina or optic nerve disease).

Ears

Some degree of deafness is common in all types of MPS disorders. It may be conductive or nerve deafness or both (mixed deafness) and may be made worse by frequent ear infections. It is important that MPS VI individuals have their hearing monitored regularly so that problems can be treated early to maximize their ability to learn and communicate.

Conductive deafness

Correct functioning of the middle ear depends on the pressure behind the eardrum being 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 lining of the middle ear will build up and in time become thick like glue. This is called middle ear effusion.

If it is possible for the child to have a light general anesthetic, a small incision through the eardrum can be made (myringotomy) to remove the fluid by suction. 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. The tubes placed in the eardrum may quickly fall out. If this happens, the surgeon may decide to use T-tubes, which usually stay in place much longer. It is expected that, once a ventilation tube is in place, fluid should drain out and hearing should improve.

Sensorineural (nerve) deafness

In most cases, the cause of nerve deafness is damage to the tiny hair cells in the inner ear.

It may accompany conductive deafness, in which case it is referred to as mixed deafness. Nerve or conductive deafness can be managed by the fitting of a hearing aid or aids in most individuals. In general, it is felt that hearing aids are underutilized in MPS disorders.

Carpal tunnel syndrome and other nerve entrapments or compression

People with MPS VI sometimes experience pain and loss of feeling in the fingertips caused by carpal tunnel syndrome. The wrist or carpus consists of eight small bones known as the carpals, which are joined by fibrous bands of protein called ligaments. Nerves have to pass through the wrists in the space between the carpal bones and the ligaments. Thickening of the ligaments causes pressure on the nerves, and this can cause irreversible nerve damage. The nerve damage will cause the muscle at the base of the thumb to waste away and will make it hard for a child to oppose his thumb in a position for a normal grasp. Although your child may not complain of pain, the carpal tunnel syndrome may be severe. If your child seems to have pain or numbness in the hands, particularly at night, it would be sensible to have an electrical test called a nerve conduction study performed. This test will show whether carpal tunnel syndrome is the cause. If your child has any weakness at all in the hand or has decreased muscle mass at the base of the thumb, then ask for the test from your neurologist. Be persistent, as many physicians may not believe that carpal tunnel syndrome is present without the classic symptoms. Most individuals affected by MPS do not have the classic symptoms of carpal tunnel syndrome, even with severe nerve entrapment and damage.

General treatment and management

Diet

There is no scientific evidence that a particular diet has any helpful effect on people with MPS VI, and symptoms such as diarrhea tend to come and go naturally. Some parents, however, find that a change in their child's diet can ease problems such as excessive mucous, diarrhea or hyperactivity. Reducing intake of milk, dairy products and sugar, as well as avoiding foods with too many additives and coloring, have helped individual children. It would be advisable to consult your doctor or a dietician 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.

Individuals with MPS VI should be as active as possible to maintain their joint function and improve their general health. Your child's doctor or physical therapist may be able to suggest ways of achieving this.

Swallowing may become difficult as an MPS VI individual gets older and the disease progresses. If this occurs, the individual may choke or aspirate food or liquids into the lungs, which can result in recurrent pneumonia. During this time there may also be a decrease in weight and feeding can take more and more time. It is often difficult for a family to consider alternate means of feeding, such as a gastrotomy tube (G-tube), and consultation with your medical geneticist and pediatric surgeon can help with your decision making.

It is important to note that there is no diet that can prevent the storage of mucopolysaccharides because they are actually made by the body. So reducing sugar intake or other dietary components cannot reduce GAG storage.

Physical therapy

Joint stiffness is a common feature of MPS VI. Limitation of motion and joint stiffness can cause significant loss of function. Range-of-motion exercises (passive stretching and bending of the limbs) may offer some benefits in preserving joint function, and should be started early. Exercises that cause pain should be avoided. Once significant limitation has occurred, increased range of motion may not be achieved, although further limitation may be minimized. Individuals with MPS VII should be as active as possible to maintain their joint function and improve their general health. Your child's doctor or physical therapist may be able to suggest ways of achieving this through a combination of daily activities and passive range-of-motion exercises.

Anesthetics

Giving an anesthetic to an MPS VI individual requires skill and should always be undertaken by an experienced anesthetist. You should inform your child's school or any other caregivers of this in case you cannot be contacted. If you have to go to a different hospital in an emergency, you should tell the anesthetist that there might be problems with intubation (placement of the breathing tube). The airway can be very small and may require a very small endotracheal tube. Placing the tube may be difficult and require the use of a flexible bronchoscope to place it gently. In addition, the neck may be somewhat lax and repositioning the neck during anesthesia or intubation could cause injury to the spinal cord. For some individuals, it is difficult to remove the breathing tube after the surgery is completed. Please advise physicians of the critical nature of these problems and that many problems have occurred during anesthesia of MPS individuals. For any elective surgery in an MPS child, it is important to choose a pediatric anesthesiologist who has experience with difficult airways. This may require that the surgery be performed at a regional medical center, not at a local hospital.

Puberty and marriage

Teenagers will go through the normal stages of puberty, though possibly later than their peers. Maroteaux-Lamy syndrome does not affect fertility, but a woman who is severely affected may be advised not to get pregnant because of the risk to her health. Children born to an MPS VI parent are automatically carriers, but none will have the disease unless the other parent is also a carrier.

Taking a break

Caring for a severely affected child is hard work. Parents need a break to rest and enjoy activities, and this may not be possible when their MPS VI child is with them. Brothers and sisters need their share of attention, and to be taken on outings that may not be feasible for the MPS child. Many parents use some form of respite care or have someone come to help at busy times.

Mildly affected individuals may need help to become more independent from their families and may benefit from a vacation, perhaps with others who have disabilities.

Health care information

Assistance may be available from specialized agencies for the disabled and from genetic clinics. You might want to look into Social Services, Social Security, Medicaid Wavers, and the Katie Beckett Law. Investigate these options, and others, in your state or with your Department of Health. If you have a social worker assigned to you, he or she should be able to locate additional information and/or resources for your family.

Living with Maroteaux-Lamy syndrome

Maroteaux-Lamy syndrome is very varied in severity. It is not possible to give estimates of the likely life span as the disorder has been only recently described, but it is likely that medical advances and better management will increase life expectancy.

Learning the diagnosis may have been a devastating experience. MPS VI can be a severe disorder, but it is possible to help affected children and adults to get the best out of their lives. Many enjoy the friendship of other MPS families. Ask the National MPS Society to put you in touch with other individuals.

Education

Some MPS VI children may benefit from having a mainstreamed education and enjoy the social interaction with peers. It is important to work with your school system and develop the best Individualized Education Program (IEP) for your child.

Specific treatment of MPS VI

The theory behind the treatment of MPS disorders

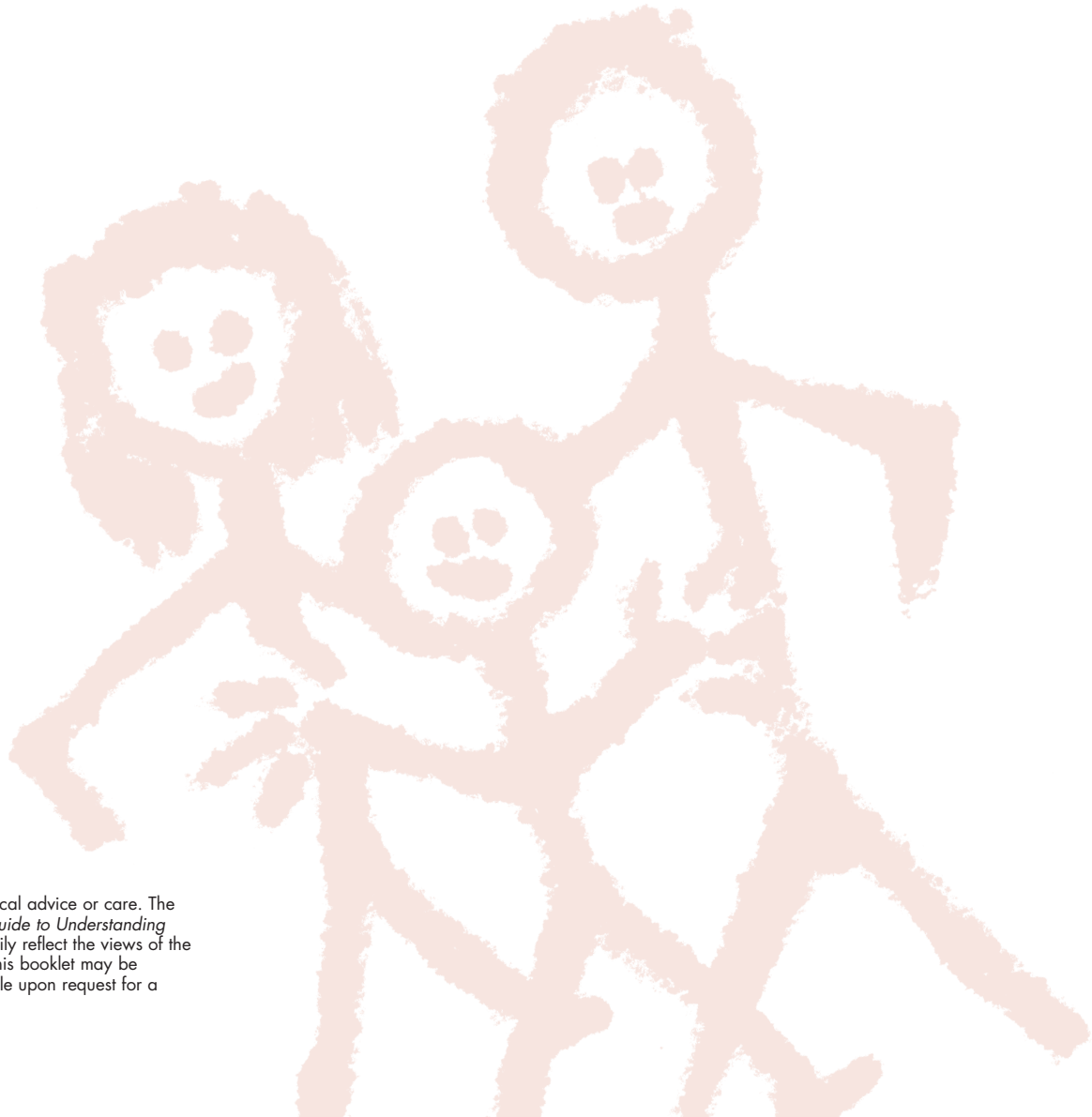
It was shown by Dr. Elizabeth Neufeld that small amounts of lysosomal enzymes, although they are intracellular in nature, could be secreted from normal cells. The secreted enzymes could then be taken up by adjacent cells and directed to the lysosome where they functioned normally. It was then shown that the biochemical defect in a cell that is deficient in a lysosomal enzyme could be corrected by taking up the small amount of enzyme secreted from an adjacent normal cell. This phenomenon, referred to as "cross section," forms the basis of all of the therapeutic strategies being developed.

Bone marrow transplant (BMT)

For some years bone marrow transplants (BMT) have been used to treat very young children with Maroteaux-Lamy syndrome. A transplant may be suggested. It is important to give yourself time to think very carefully about this and to weigh the potential risks and possible advantages of this treatment. Some children with Maroteaux-Lamy syndrome have benefited from BMT. The National MPS Society can put you in touch with parents whose children have had BMT so that you may be better informed before reaching a decision.

Enzyme replacement therapy (ERT)

An experimental trial of enzyme replacement therapy is being planned for individuals with MPS VI. Although there is reason to hope that enzyme replacement therapy given by intravenous infusion will help some of the physical problems, the blood-brain barrier may prevent enzyme therapy from directly helping the brain.



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The National MPS Society, Inc.