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.

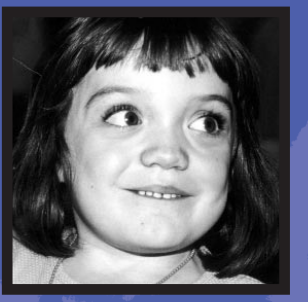
Join the National MPS Society and enjoy a variety of benefits, including:

- *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

Morquio Syndrome

Mucopolysaccharidosis (MPS) IV

MPS

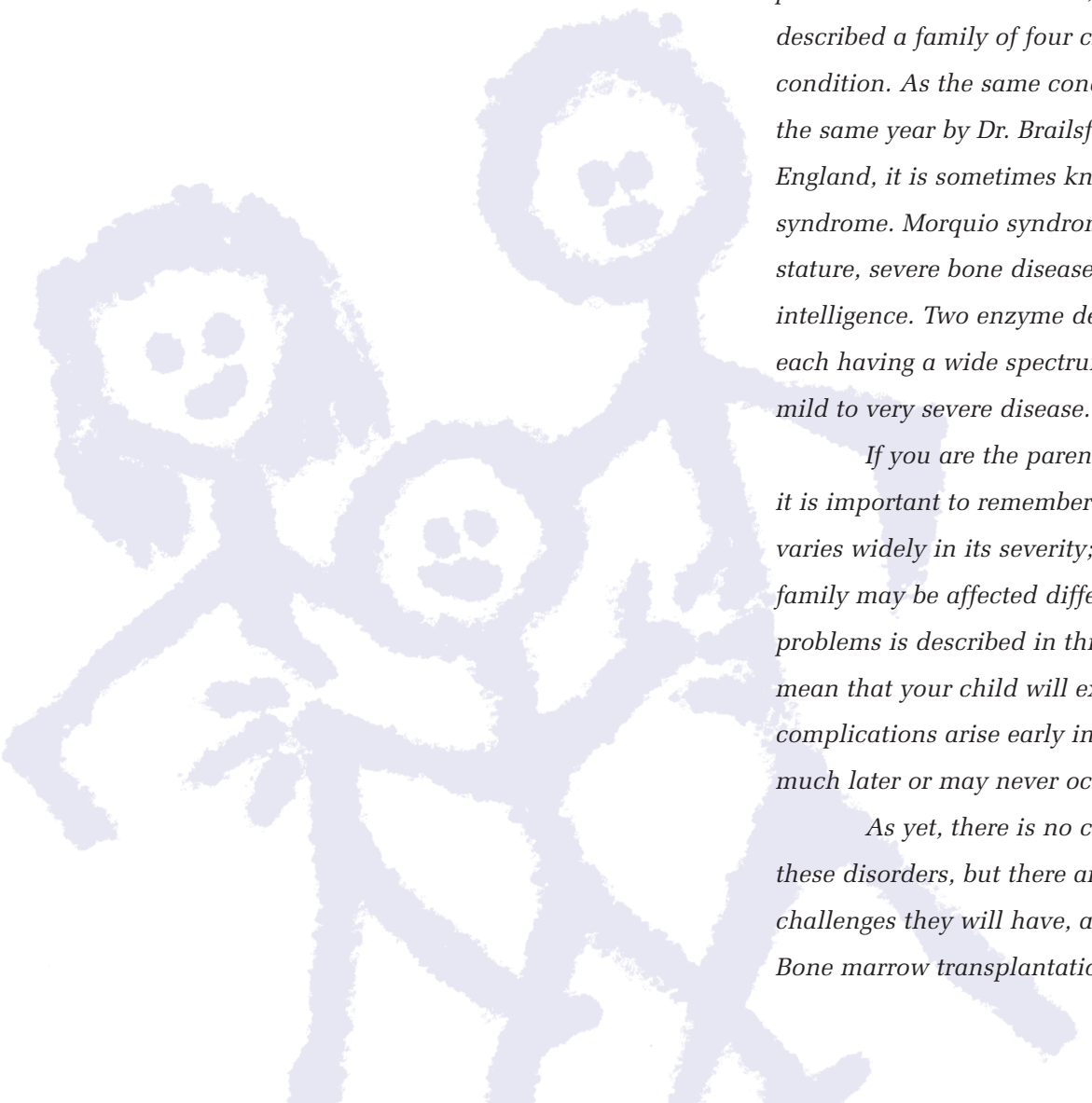
The National MPS Society, Inc.

Introduction

Morquio syndrome is a mucopolysaccharide storage disorder, also known as Mucopolysaccharidosis IV (MPS IV). It takes its name from Dr. Morquio, a pediatrician in Montevideo, Uruguay, who in 1929 described a family of four children affected by this condition. As the same condition was also described in the same year by Dr. Brailsford from Birmingham, England, it is sometimes known as Morquio-Brailsford syndrome. Morquio syndrome is characterized by short stature, severe bone disease and preservation of intelligence. Two enzyme deficiencies can cause MPS IV, each having a wide spectrum of clinical severity from mild to very severe disease.

If you are the parent of a newly diagnosed child, it is important to remember that Morquio syndrome varies widely in its severity; even children from the same family may be affected differently. A range of possible problems is described in this booklet, but this does not mean that your child will experience them all. Some complications arise early in childhood, while other arise much later or may never occur.

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 attempted



front cover
top: Allison, age 5, MPS IV A
middle: Ian, age 12, MPS IV A
bottom: David, age 4, MPS IV A

to treat MPS IV, with little success. Scientists who study MPS continue to look for better and more effective ways to treat these disorders, and it is likely that individuals 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 IV 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 is in part made of mucopoly-saccharides. 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 IV, 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 for your body to keep itself healthy.

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).

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 works on nails. Individuals with MPS IV are missing one of two specific enzymes, which are essential in the breakdown of one of the GAG called keratan sulfate. The incompletely broken down keratan 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.

The incompletely broken down keratan sulfate remains stored inside cells in the body and begins to build up, causing progressive damage.

Are there different forms of the disorder?

There are two different enzyme deficiencies that have been found to cause Morquio syndrome, and the disorder is described as type A or B. The names of the enzymes are N-acetylgalactosamine 6-sulfatase (type A), also called galactose 6-sulfatase, and beta-galactosidase (type B). MPS IV A, the most common form, was first recognized to be a severe disorder. MPS IV B was initially considered to be a mild form or late onset variant of Morquio syndrome. It is now recognized that the severe and mild forms of Morquio syndrome are not caused by a unique enzyme deficiency, but rather that each form has a wide spectrum of clinical severity. In general, individuals with MPS IV B have similar problems, but tend to be mildly affected compared to MPS IV A individuals. Both enzymes are only involved with the breakdown of keratan sulfate, a type of glycosaminoglycan. Keratan sulfate is primarily found in the bones and connective tissues. The accumulation of keratan sulfate in the bones and connective tissues is responsible for the major problems that affect patients with both types of MPS IV.

There is little clinical difference between the different types of the Morquio syndrome, since both types accumulate the same GAG, keratan sulfate. The accumulation of keratan sulfate in the bones and connective tissue is responsible for the major problems that affect patients with both types of MPS IV.

Carly, age 4
MPS IV A

How common are these disorders?

Morquio syndrome is one of the rarest kinds of mucopolysaccharide disorders in the United States. Reliable incidence figures are not available, but estimates have varied between 1 in 200,000 live births to 1 in 300,000 live births. Even though these disorders are rare, each patient needs such extensive medical care that the effect on the medical system is much larger than their numbers suggest.

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 IV is one such disease. Most families with an MPS IV child do not have a family history of any genetic problem – MPS IV 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 IV 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 IV 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 IV, 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 IV in utero.

Clinical problems in MPS IV

Growth

Children with Morquio syndrome usually grow normally at first, but growth may start to slow down around eighteen months. Those who are severely affected usually stop growing around the age of eight. Final height may be between 3 feet and 4 feet. Other Morquio individuals continue growing into their teens and can reach 5 feet. The trunk is relatively shorter than the limbs.

Intelligence

Individuals with Morquio syndrome are of normal intelligence.

Physical appearance

The face is altered to a certain extent by the disorder. The mouth tends to be wide, the jaw square, and the bridge of the nose flattened. The neck is very short, but the texture of the hair is not affected as it is in other MPS disorders.

Nose, throat, chest, and ear problems

If you look at a skeleton or a diagram of one, you will see that the breastbone (sternum) is joined to the spine by the ribs. In Morquio syndrome, the growth of the spine is affected. The breastbone continues to grow more normally, but as it is joined to the spine, it is forced to buckle outward in a rounded curve or sometimes in a prominent beak shape. The chest is bell-shaped and the ribs are held fixed in a horizontal position, causing restriction of efficient breathing. A breathing test, called a pulmonary function test, can be used by a lung specialist to assess the amount of restriction the abnormal bones are causing with breathing.

Although children with Morquio syndrome are not necessarily more prone to respiratory infections, their restricted chests mean that they are less well able to cope with an infection if it involves the lungs. Your doctor may advise you to contact him or her immediately if you think an infection might be starting so that he can prescribe an antibiotic, if necessary.

Cases have been reported of Morquio patients suffering severe breathing problems after spending several days at high altitude. It would be wise to check with your doctor if you are planning a long airplane flight or a vacation in the mountains.

Treatment of respiratory infections

Drugs may affect people with MPS IV 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 anti-histamines, 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 IV. Cough suppressants or drugs that are too sedating may cause more problems with sleep apnea by depressing muscle tone and respiration.

Carol, age 7
MPS IV A

Although most normal individuals with colds do not require antibiotics, individuals with MPS IV almost always 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 worsens the problem in overcoming infections, therefore it is common to have infections improve on antibiotics and then promptly recur after the antibiotic course is over. Chronic antibiotic therapy may 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 an Ears, Nose and Throat (ENT) specialist experienced with MPS disorders to determine which tube is best.

Many people with MPS IV 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

People with MPS IV may have a prominent chin, wide mouth, and enlarged tongue. The teeth can 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 signs of an infected tooth in a severely involved individual.

If an MPS IV individual has a heart problem, it is advised that antibiotics should be given before and sometimes 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.

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.

Heart

Heart disease can occur in people with Morquio 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. 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. Many individuals with MPS IV have some degree of heart valve leakage or blockage. Some individuals with Morquio syndrome may develop problems with the aortic or mitral valves. They may have slowly progressive valvular heart disease for years without any apparent clinical effects. If the condition worsens, an operation may be needed to replace the damaged valves.

From top left, clockwise:
Donna, age 38, MPS IV
Noel, MPS IV
Ryan, age 6, MPS IV
Ian, age 12, MPS IV
David, age 4, MPS IV

As heart problems occur in MPS IV, 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.

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

Abdomen and hernias

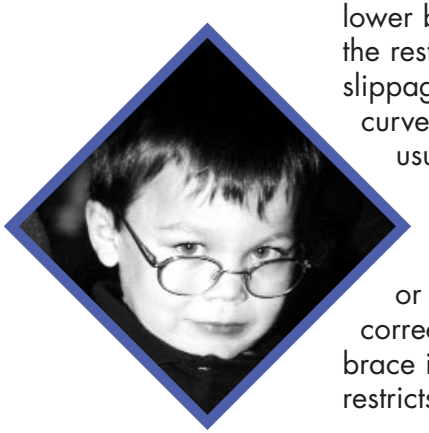
In people with MPS IV, the abdomen bulges out due to posture, weakness of the muscles and the enlarged liver and spleen. Frequently part of the abdominal contents will push out behind a weak spot in the wall of the abdomen. This is called a hernia. The hernia can come from behind the navel (umbilical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

Bones and joints

People with MPS IV tend to have significant problems with bone formation and growth. The bone disorder in MPS IV is different from that seen in other MPS disorders. The major clinical features of MPS IV are those related to the bones and their effect on the nervous system if nerves are compressed by abnormal bone movement.

Spine

The bones of the spine (vertebrae) normally line up from the neck to the buttocks. In individuals with MPS IV, the spine tends to be severely affected and its bones or vertebrae are abnormally flattened (platyspondyly). The vertebrae are often poorly formed and may not stably interact with each other. If this happens, the vertebrae can compress and injure the spinal cord. One or two of the vertebrae in the lower 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 surgical treatment. If curvature is not severe, then bracing may be suggested. There are different opinions on whether surgery is needed or whether a brace can be used either to correct a curve or to support the back. If a brace is used, it should not be the kind that restricts the movement of the chest even further.



Ryan, age 6
MPS IV

Neck

Problems in this area are perhaps the most serious facing people with Morquio syndrome. The neck problems need to be discussed at the time of diagnosis, since serious problems can occur before 5 to 6 years of age. The difficulties arise from a structural defect in the upper vertebrae of the spine, which is made worse by the very loose ligaments. The bones that stabilize the connection between the head and neck are usually malformed (odontoid dysplasia) in people with MPS IV. There should be a piece of bone called the odontoid process sticking up between the first and second vertebrae which gives support as the head moves backwards and forwards. With odontioid dysplasis, the neck can become unstable, which places the spinal cord at risk to life-threatening injury. The spinal cord is big bundle of nerves that carries messages between the brain and the rest of the body. If the cord is compressed or squeezed (cervical myelopathy), there will be gradual worsening effects on the child with paralysis or death occurring, if left untreated.

Children with Morquio syndrome should be referred to an orthopedic surgeon from an early age to monitor the condition of the cervical spine. MRI studies or X-rays will be performed with the head bent forward and with the neck straight (flexion and extension view) and will be repeated from year to year to monitor the situation. A baseline study of the neck is recommended at the time of diagnosis. 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.

Parents may be alarmed at the thought that their child has a weakness in such a vital place and may be harmed by a severe fall. The surgeon will be able to advise you on the risks. It is important to keep a balance between avoiding risks and letting the child lead as normal a life as possible, but activities such as somersaults, head stands, or diving should be avoided completely.

The problem with the cervical spine can be corrected by an operation called a cervical fusion. Small pieces of bone are taken from the legs and fixed into the neck where they eventually grow to form a firm support, joining the top two vertebrae to the base of the skull. Fusion operations are often carried out on patients with other conditions, but there are special problems with Morquio patients and common methods may fail. Most orthopedic surgeons have minimal or no experience with MPS IV individuals. One or two hospitals have taken a particular interest in Morquio patients and have performed a number of fusion operations. The National MPS Society has information about orthopedic surgeons and hospitals where successful cervical fusions have been performed.

It has been found essential to immobilize the neck in the correct position (reduced) for over four months while the grafted bone grows up to join the base of the skull. One method of achieving this successfully is known as the "halo" method.

A metal "halo" ring is attached to the skull, and bars leading from it are fixed to the body by a plaster cast. The operation usually involves a hospital stay of usually no more than one week. The halo is typically required for 3 to 4 months to allow healing and fusion of the spine. Although caring for a patient in a halo is hard work, patients adjust remarkably well.

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 MPS disorders, with the range of movement of joints becoming limited. However, in MPS IV, the joints may be more mobile than normal due to very loose ligaments.

Hips

It is common for the hips to become dislocated, but this is often not a problem and treatment may not be necessary. If there is pain later in life, surgery may be considered.

Legs and feet

The knees of most MPS IV individuals grow abnormally and develop severe "knock-knees" (genu valgum). It is possible for the legs to be straightened by an operation, but it is best to wait until the child has stopped growing. Surgery attempted earlier has had disappointing results as the legs rapidly revert to their former crooked condition. The ankles may be weak and turn inward in MPS IV. Occasionally boots or splints are worn, but firm shoes are usually adequate. The bones composing the arches in the feet are held in position by ligaments and tendons. These are likely to be weakened, resulting in flat feet. The toes may be deformed.

Shoulders

The shoulders are often partially dislocated downward (subluxed) so that the arms cannot be raised straight above the head. This rarely causes any discomfort, but it can make dressing or attracting the teacher's attention more difficult.

Donna, age 38
MPS IV

Wrists

The wrists are enlarged and curved. They may become very loose and floppy as the tiny carpal bones fail to develop properly and the ligaments are lax. This can mean considerable loss of strength in the hands. Attempts have been made to correct this problem surgically by grafting bone from another part of the body to the wrists, but unfortunately, the technique has not yet worked. Small plastic splints with wrist bands may be helpful in some cases. Writing may be difficult and some individuals may find it easier to use a typewriter or word processor.

Carpal tunnel syndrome is common in most forms of MPS disorders, but has not been a frequently reported problem in MPS IV. If your child seems to have pain or numbness in the hands, particularly at night, it would make sense 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.

Neurological problems: brain, senses and nerves

Eyes

The circular window at the front of the eye (cornea) can become cloudy due to storage of mucopolysaccharides, which disrupts the clear layers of the cornea. Corneal clouding tends to be mild in most MPS IV individuals. If corneal clouding is severe, it may reduce sight, especially in dim light. Some individuals cannot tolerate bright lights as the clouding causes uneven refraction of light. Wearing caps with visors or sunglasses can help.

Ears

Some degree of deafness is common in all types of MPS IV 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 IV 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.

General treatment and management

Diet

There is no scientific evidence that a particular diet has any helpful effect on people with MPS IV, 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 some individuals. 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 appears to increase the child's symptoms.

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.

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

Vomiting

A small number of children with MPS IV experience occasional severe episodes of vomiting to such an extent that the child becomes dehydrated. The cause of this is not yet known, but if your child should have this problem, it is advisable to call your doctor.

Physical therapy

Joint stiffness is not a feature of MPS IV, but loose joints can be a problem. Intensive physical therapy will be needed to help an individual with MPS IV start walking again after neck, spine or knee surgery. Individuals with MPS IV should be as active as possible to maintain their muscle strength and general health. Your child's doctor or physical therapist may be able to suggest ways of achieving this. It is important to keep a balance between avoiding risks and letting the child lead as normal a life as possible. Activities such as somersaults, head stands, or diving should be avoided completely because of risk to the neck.

Anesthetics

Giving an anesthetic to an MPS IV individual requires skill and should always be undertaken by an experienced anesthetist. If the cervical spine is unstable, the MPS IV individual is at risk if the neck is flexed while unconscious, and special precautions must be taken. 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 may be problems with the neck and possibly with intubation (placement of the breathing tube). The airway can be very small and may require a very small endo-tracheal 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

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

Adolescents with Morquio syndrome will go through normal pubertal development although teenage girls may have delayed menstrual periods. Morquio syndrome does not affect fertility, but a woman who is severely affected may be advised not to get pregnant because of the risks to her health. All children born to an MPS IV parent are automatically carriers but none will have the disease unless the other parent is also a carrier.

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 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 Morquio syndrome

In the past, severely affected patients did not survive beyond their twenties or thirties, although those less severely affected have lived to their fifties and sixties. It is likely that medical advances and better management of the condition will significantly increase life expectancy. It seems sensible, therefore, for families to hope for a reasonable life expectancy for their child.

First learning about the diagnosis may have been a devastating experience. This can be a severe disorder, but there is every reason to find comfort and hope. Morquio children can lead very active and full lives; many are at ordinary schools, joining in clubs and other activities. Adults have gone on to college, found satisfying jobs and learned to drive. Many enjoy the friendship and support of other Morquio families. To learn more about how to get in touch with other Morquio families, contact the National MPS Society.

Education

Some MPS IV 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 IV

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.

Enzyme replacement therapy (ERT)

A clinical trial of enzyme replacement therapy was conducted for MPS I and experimental trials are being planned for MPS II and MPS IV. Enzyme replacement therapy may be a future option for the MPS IV individual.

For up-to-date information on treatment options, contact the National MPS Society.

MPS

The National MPS Society, Inc.