

### What are MPS and ML Disorders?

Mucopolysaccharidoses (MPS) and Mucopolipidoses (ML) are genetic lysosomal storage disorders caused by the body's inability to produce specific enzymes. Normally, the body uses enzymes to break down and recycle cells after the cells die. In affected individuals, the missing or insufficient enzyme prevents the normal breakdown and recycling of cells resulting in the storage of these cell deposits in virtually every cell of the body. As a result, cells do not perform properly and may cause progressive damage throughout the body, including the heart, bones, joints, respiratory system and central nervous system. While the disease may not be apparent at birth, signs and symptoms develop with age as more cells become damaged by the accumulation of cell deposits.

<u>Syndrome</u>		<u>Enzyme Deficiency</u>
MPS I H	Hurler	$\alpha$ -L-Iduronidase
MPS I S	Scheie	$\alpha$ -L-Iduronidase
MPS I H-S	Hurler-Scheie	$\alpha$ -L-Iduronidase
MPS II Hunter		Iduronate sulfatase
MPS III A	Sanfilippo A	Heparan <i>N</i> -sulfatase
MPS III B	Sanfilippo B	$\alpha$ - <i>N</i> -Acetylglucosaminidase
MPS III C	Sanfilippo C acetyltransferase	Acetyl CoA: $\alpha$ -glycosaminide
MPS III D	Sanfilippo D	<i>N</i> -Acetylglucosamine 6-sulfatase
MPS IV A	Morquio A	Galactose 6-sulfatase
MPS IV B	Morquio B	$\beta$ Galactosidase
MPS VI	Maroteaux-Lamy (arylsulfatase B)	<i>N</i> -Acetylgalactosamine 4-sulfatase
MPS VII	Sly	$\beta$ -Glucuronidase
MPS IX		Hyaluronidase
ML II	I-Cell	<i>N</i> -acetylglucosamine-1-phosphotransferase
ML III	Pseudo-Hurler polydystrophy	<i>N</i> -acetylglucosamine-1-phosphotransferase

### What are the Major Characteristics of MPS and ML Disorders?

While the symptoms of the diseases may vary from one syndrome to another, there are similarities. Affected individuals may have mental retardation, cloudy corneas, short stature, stiff joints, incontinence, speech and hearing impairment, chronic runny nose, hernia, heart disease, hyperactivity, depression, pain and a dramatically shortened life span.

### How Are These Disorders Inherited?

MPS and ML are hereditary. In nearly all cases a child receives a recessive gene from each parent. MPS II is the only exception where the gene may be passed from a mother to her male children. A couple's chance of having another child with one of these disorders is 1 in 4 with each pregnancy. Unaffected siblings may be gene carriers of the disorder. The occurrence of MPS and ML in the general population is thought to be one in 25,000 births.

### **Is Prenatal Diagnosis or Carrier Detection Testing Available?**

Yes! For most MPS and ML conditions, amniocentesis can be performed between 14 and 17 weeks gestation to determine if the unborn child is affected. Alternatively, chorionic villus sampling (CVS) can be performed between eight and ten weeks of pregnancy. Tests also are available to determine whether individuals are carriers of an MPS or ML disorder. To learn more about these tests contact your doctor, nearest genetic center or the National MPS Society.

### **Is Research Helping Today's Families?**

Absolutely! Although there is currently no cure for MPS or ML disorders, research is making great strides. Carrier detection, the development of replacement enzymes, and the possibility of gene therapy, are among today's research themes and treatment options. Bone marrow transplantation has been considered successful for many, though relatively few individuals qualify for this high-risk procedure. We've made major advancements in research thanks to the fundraising efforts of the Society and its members.

### **How Can You Help Find the Cure?**

Since 1975 the Society has supported individuals and families affected with MPS and ML disorders. We are governed by a member-elected volunteer Board of Directors, many of whom are parents of MPS/ML children. We benefit from the expertise of a Scientific Advisory Board, comprised of world-class physicians, researchers and medical professionals throughout the world. We need your support in helping us teach our mission to others and raising the money we need to support medical research - the key to longer, happier lives with MPS and ML.

To **support research** volunteers raise money to provide student fellowships and fund research projects at a number of prestigious university centers. Contributions may be designated for general or syndrome-specific research. Technical conferences are periodically held allowing researchers to collaborate and discuss their findings. Sponsors are urgently needed to accelerate the encouraging research that is bringing therapies - and ultimately the cures - to children and adults with MPS and ML.

To **support families** the Society works in many ways. We publish an ever-growing series of resource guides dealing with specific disorders and treatments. Our newsletter, *Courage*, is distributed by and for the membership and is commonly referred to as a lifeline for families, physicians and professional care providers. *Courage* contains letters and photos submitted by families, reports on the breakthroughs in research, and information on disease management and improvements in therapies. We hold conferences every year in different parts of the country enabling families to meet and learn more about their disorder. Leadership families have been identified both by region and by syndrome to serve as valuable points of contact for newly diagnosed families. A fund has been designated to assist families to attend our conferences and to obtain

medical products that will improve their quality of life. We also provide much needed emotional support to all those affected by the tragedies of MPS and ML.

To **increase public and professional awareness** the Society sponsors public events, issues press releases, publishes syndrome and treatment materials, and maintains a website. The Society's web site, [www.mpsociety.org](http://www.mpsociety.org), is available to anyone seeking information and updates on research, legislative activities, family support and upcoming events. In keeping with our mission, the committee on federal legislation advocates enhanced MPS research in the pursuit of treatments for MPS and ML disorders. We work to cultivate working relationships with congressional offices and government agencies for the benefit of MPS and ML families as well as advocating enhancements to federal programs such as SSI, Medicaid and others important to the daily lives of MPS and ML families. The National MPS Society participates in international symposiums, which gather a global contingent of MPS and ML medical and scientific professionals, and networks with a growing number of international sister organizations.

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The National MPS Society. is a tax-exempt non-profit organization under IRC Section 501(c)3, dedicated to individuals affected with MPS and ML disorders and to their families.