

## MPS Disorders (Hunter / Hurler / Morquio) at a Glance

The Mucopolysaccharidoses disorders (MPS) belong to a group of disorders known as lysosomal storage disorders (LSDs), which are a group of diseases caused by a defect in lysosomal enzymes. Lysosomes act as the "recycling center" of each cell, breaking down unwanted materials into simple waste products. The lack of certain lysosomal enzymes causes a buildup of waste products in many cells of the body.

The clinical symptoms of each type of LSD depend on which cells and tissues use the deficient enzyme and if any working amount of enzyme is present. The different types are named for the substance that builds up in the cells.



Meet Travis on page 12!

MPS disorders are the most common (1 in 25,000 births) of the LSDs, but there are more than 40 known types. Below are some of the more common types\*, in addition to MPS:

- **Fabry** disease - causes kidney and heart problems, pain, and a skin rash
- **Gaucher** disease - causes the spleen to enlarge, anemia, and bone lesions if untreated; can also cause progressive neurologic problems
- **Niemann-Pick B** disease - leads to enlargement of the spleen and liver, as well as lung disease
- **Pompe** disease - a storage disease in which glycogen builds up in the liver, heart and muscle, and can be fatal if it develops in infancy (also known as acid maltase deficiency)
- **Tay-Sachs** disease - a lysosome storage disease that occurs more commonly in people of Eastern European Ashkenazi descent. It causes degeneration of the brain in infants with early death.

*\*Links to support organizations for these conditions are found in the Resource section below.*

### Things to think about

#### 1. Medical / Dietary Needs

#### What you need to know

Changes to the diet will not prevent disease progression, but limiting milk, sugar, and dairy products has helped some individuals experiencing excessive mucus. It is important to discuss dietary needs of the child with the parents to learn if there are restrictions. If there isn't a special diet required for an individual with an MPS, a well-balanced diet is important.

If the child is experiencing sleep apnea or obstructive airway disorder, surgery to remove tonsils and adenoids may be done to improve breathing. Sleep studies can assess airway status and the possible need for nighttime oxygen.

Surgery may also be done to correct hernias, help drain excessive cerebrospinal fluid from the brain, and free nerves and nerve roots compressed by skeletal and other abnormalities.

Mobility problems, hearing loss and vision difficulties are the major medical complications in MPS that may need the special attention of school personnel in program planning. It is important to discuss the nature of the student's condition and implications for school activities. The child's physicians will determine the student's permitted activity levels.

School age children with MPS may have multiple doctors and specialist visits to monitor medical conditions.

### *Physical characteristics and/or symptoms of MPS disorders:*

Mucopolysaccharides are long chains of sugar molecules used in the building of the connective tissues in the body. The types of MPS are listed below with the most common types in bold. The bolded types will be discussed further:

- **MPS Type I – Hurler, Hurler-Scheie, and Scheie syndromes**
- **MPS Type II – Hunter syndrome**
- MPS Type III – San Filippo syndromes
- **MPS Type IV – Morquio syndrome**
- MPS Type VI – Maroteaux-Lamy syndrome
- MPS Type VII – Sly syndrome

### **Mucopolysaccharidosis Type I (MPS Type I):**

- MPS I is seen in all populations at a frequency of approximately 1:100,000 for the severe form and 1: 500,000 for the attenuated form
- Types based on age of onset and severity

- **Hurler** (This is the most severe type of MPS Type I.) Possible findings:
  - Progressive skeletal dysplasia
  - Coarsening facial features
  - Corneal clouding
  - Can lead to significant vision disability
  - Hearing loss is common
  - Death usually by year 10
  - Cardiac involvement
  - Gastrointestinal; diarrhea and constipation
  - Intellectual disability is progressive and profound
  - Limited language skills
  
- **Attenuated MPS Type I (Hurler-Scheie and Scheie)** (These are less severe than Hurler syndrome.) Possible findings:
  - Skeletal and joint are significant sources of disability and discomfort
  - Scoliosis
  - Back pain
  - Many have normal or near normal intellect
  - Some have learning disabilities
  - Sleep apnea
  - GI: hernias
  - Respiratory: progressive pulmonary disease
  - Hearing loss (moderate to severe)
  - Cardiac valvular disease
  - Short stature
  - Enlarged spleen (hepatomegaly)
  - Corneal clouding can lead to significant vision disability
  
- **MPS Type II (Hunter syndrome)**
  - Incidence is approximately 1 in 100,000 births
  - Primarily affects males with females being carriers, but rarely a female can be affected
  - Varies from severe to attenuated

- Symptoms of the severe form resemble the features seen in Hurler syndrome (MPS Type 1)
  - Symptoms of the attenuated type resemble the features seen in the more attenuated MPS Type 1 (Hurler-Scheie and Scheie syndromes) although the facial features may be coarser in Hunter
  - Intelligence can be normal or can be affected in the more severe form
- **MPS Type IV (Morquio syndrome)**
- Coarsening of the facial features
  - Skeletal deformities
    - Carpal tunnel syndrome
    - Joint stiffness
  - Abnormal walk
  - Breathing problems
  - Eye problems
  - Hearing loss
  - Dental problems
  - Enlarged liver and spleen
  - Juvenile form (early-onset, severe form):
    - Mental function declines
    - Severe intellectual disability is typical
    - Aggressive behavior and hyperactivity
    - Spasticity
  - Late (mild) form
    - Mild to no mental deficiency

*Treatment for MPS – all types:*

- Enzyme replacement therapies can help increase the missing enzymes
- Occupational therapy
- Speech therapy
- Physical therapy
  - Range of motion exercises help preserve joint function
  - Carpal tunnel exercises
- Eye shades can help reduce glare from corneal clouding
- Hearing aids may be appropriate

- Gastrointestinal
  - Constipation and diarrhea can often be controlled by diet and possibly laxatives.

### **What you can do**

- A yearly check-up and studies as needed should occur in the child's Medical Home.
- Be aware of any changes in behavior or mood that seem out of the ordinary and notify the parents.
- Be aware of changes in vision as the corneal clouding can be progressive
- Note any excessive sleepiness during the day and if napping takes place at school, report any changes in snoring
- Be aware of any progression in the joint disease that may make their schoolwork more difficult
- It is important to be aware of any academic changes. Contact parents when any differences are noticed.
- Seizure emergency plan should be in place if seizures are part of the child's condition
- Students may need to visit the nurse to take medications during school
- If there are heart issues, consider AED devices and staff education on their use, as well as CPR training for staff.

## **2. Education Supports**

### **What you need to know**

Mobility problems, hearing loss and vision difficulties are the major medical complications in MPS that may need the special attention of school personnel in program planning. Each child with MPS may need specific education supports, some are physical and some are learning. It is important to discuss the child's individual's needs. Many of these conditions are progressive. It is important to have high but realistic expectations for each child.

Consider therapists and specialists to consult and support classroom teacher.

- Physical therapy
- Occupational therapy
- Speech therapy
- Vision therapy
- Musculoskeletal support
- Treatment for prevention of scoliosis related complications

- Alternative communication and supports
- Paraprofessional support may be needed
- Nursing support as needed

## What you can do

Vary support to meet the needs of the individual child who has an MPS disorder.

Communication /Language:

- Incorporate typical language skills in social, work, and life skill areas.
- Children with speech and motor difficulties often benefit from speech and occupational therapy.
- Teach learning strategies for non-verbal communications.
  - Consider new technology, computers, and sign language.
  - Incorporate early use of augmentative communication aids such as picture cards or communication boards.
  - Communication should work with child's desire to socially interact with others in natural settings.
  - Make sure children have opportunities for choice and control in their lives (choose colors, clothing, play, work partner, etc.)
  - Consider multiple means of communication paired with the knowledge of when to use one method vs. another.
  - Find AAC system that allows for maximal social reciprocal communication.
  - Encourage finger pointing early to help with device use as they age.
  - All persons interacting with the child should have education and training on how to encourage reciprocal communication with the device.
  - Model, model, model use of the AAC device to encourage its use
  - Continue with strategies that improve oral control to maximize their potential as oral speakers.
  - Consider a team approach, such as an OT or PT to consultant to work with the SLP for optimal positioning to get the most benefit from hand use for communication.
  - Consider vision specialist to gain insight into best visual field for communication devices.

Vision:

- Seat child close to board
- Use a larger font

Hearing:

- Appropriate amplification and/or assistive technology
  - Hearing aids
  - Cochlear implants
  - FM systems (<http://www.asha.org/public/hearing/FM-Systems/>)
- A favorable acoustic environment where noise is eliminated or reduced.

Physical differences are often due to skeletal differences:

- It is important to find the balance between providing help and fostering autonomy.
- Individuals should be encouraged to the maximum extent possible to live independent productive lives with adaptive or adjustments assistance.
- An IEP or 504 plan may be in place for safety and comfort in the class and school.
- Adaptive aids in school may be required for
  - Heavy doors
  - High doorknobs
  - Reaching for blackboard
    - Extenders
    - Stools (if safe)
  - Desk size
  - Bathroom
    - Encourage use of regular bathroom with adaptations as necessary
    - Use nurse's office with a permanent step only if necessary
  - Carrying books may be challenge
    - Two sets of books; one for home one for school
    - Friend helper
    - Low locker
    - Rolling back pack
  - Allow extra time to travel between classes/use elevators
  - May need stool to rest legs on
    - Ensure best fit of desk and chair
    - Legs may fall asleep if left to dangle
    - Upper legs are too short to allow back support pillow for back support
- Occupational therapy and/or accommodations for writing

- Individuals may have small fingers and joint hypermobility due to stiffness
- May not be able to write at a quick enough speed
  - Consider tape recorder for class
  - Use computers/iPads
  - Additional time for tests or provide oral exams
- Patients who begin to lose mobility due to weakened muscles may also benefit from physical therapy.
  - A customized exercise and/or physical therapy program may help to preserve range of motion and strength.
  - Use of assistive devices such as orthotics, canes, or walkers may help with ambulation.
  - A wheelchair may be indicated.
- Ataxia (difficulty coordinating smooth motor movement)
  - Unstable or non-walking children may benefit from physical supports in the classroom.
  - They may need extra supports/people to help them in their academic program and inclusion.
  - Individuals with motor issues may need extra space and/or minimal obstructions to be safe.
  - Physical therapy
    - Adaptive chairs or positioning support may be helpful
  - Occupational therapy
    - May help with fine motor and oral motor control

### 3. Behavioral and Sensory Support

#### What you need to know

Some children with MPS may present with a psychological complication, however this isn't the rule. It is important to discuss the child's individual issues.

- Psychological and psychosocial difficulties
  - Fatigue and pain may make psychological problems worse
  - Psychological distress may increase pain
  - Depression, anxiety, low self-confidence, negative thinking, hopelessness, and desperation may occur
  - Depression is often associated with chronic pain, disabilities



- Individuals may feel misunderstood, disbelieved, and/or alone
- Children may be self-conscious of physical differences.
- Pain
  - Fear of pain and joint instability may lead to avoidance behaviors, and make dysfunction and disability worse

## What you can do

- Counseling and support for pain may help
- Meditation and yoga could be helpful
- Antidepressants may be helpful
- May need help with adaptation and acceptance of issues and potential limitations.
- Some individuals need support for social skills development.
  - Misconceptions of abilities can cause insecurity and anxiety in social situations
- Make sure teaching strategies being used are appropriate for children who are already socially engaged.

Treat children who have MPS the same as you would with any other individual who might have a psychological condition. They may need:

- Behavioral supports
- Counseling
- Medication

## 4. Physical Activity, Trips, Events

### What you need to know

Physical Education:

- It is important for individuals with LSD to have an opportunity for physical activity to optimize physical and mental health.
- Certain LSD syndromes will have very specific recommendations regarding physical exercise and restrictions.
  - Exercise is important and should be encouraged, especially those exercises that build muscle mass and motor skills.
  - Individuals may be able to exercise but not participate in contact sports.
  - Adapted PE programs

- Downsize equipment (i.e. smaller bat)
- Have a designated runner
- If activity can't be modified to be safe, have child be a scorekeeper, umpire etc.
- Occupational therapy may help with accommodations and modifications
- Some individuals may need additional time to get to class
  - Limit extra movement between classes

#### Field Trips:

- If there is to be a lot of walking on the field trip it is important to be aware that it may take the child more time and they may be tired
  - Consider cutting down on walking when possible
  - Use alternative forms of transportation

### What you can do

#### Physical Education:

- Encourage academic and artistic activities
- Allow child to help with coaching or team management if there are restrictions.
- Consider 504 Plan for physical activity modifications if appropriate
- Instruct in self-monitoring technique.
  - Provide opportunities for practice using self-monitoring strategies
  - Instruct in relaxation techniques, safety, and breathing
- Consider any seizure plans, cardiac support (AED device or CPR) that might be needed.

#### Field Trips:

- Assistive devices are available to improve accessibility and independence in homes, school, and community.
- Take any seizure meds and seizure protocol on the trip
- Consider if an AED or CPR trained individuals will need to be on the trip

## 5. School Absences & Fatigue

## What you need to know

- Children with LSD may be absent due to surgery, illness and/or medical appointment.
- Fatigue may be a symptom of a condition (like Gaucher) or may be a side effect from disordered sleep.

## What you can do

- Help to make transitions in and out of school as seamless as possible.
- Allow extra time for assignments and/or provide make up work as needed.
- Make accommodations for resting or taking breaks.
- Have peers share class notes.
- Monitor work so that it is challenging, but there are attainable and realistic goals.
- Plan for absences and consider tutoring.
- Communication with parents is important to meet these challenges.
- Children with LSDs may be tired and require rest opportunities or breaks in their day.

## 6. Emergency Planning

### What you need to know

- There should be a plan for an emergency evacuation of children with physical limitations.
  - Need to avoid situations where individuals may be trampled
  - Child needs to be able to reach door handles.
- Someone assigned to help individuals with a back-up person if that person is out.
- Seizure plan should be in place if appropriate.
- AEDs (Automated External Defibrillator) or CPR (cardiopulmonary resuscitation) may be necessary if a child has heart involvement.

### What you can do

- Develop an emergency plan if necessary, depending on the needs of individual children.
  - Assign a back-up when needed.

## 7. Resources

## National MPS Society

<http://mpssociety.org/>

- Education Strategies and Resources – A Guide for Parents  
[http://mpssociety.org/wp-content/uploads/2011/07/Education\\_2008.pdf](http://mpssociety.org/wp-content/uploads/2011/07/Education_2008.pdf)
- MPS 1 (hurler, Hurler-Scheie, Scheie syndrome)  
<http://mpssociety.org/mps/mps-i/>

## Genetics Home Reference

Consumer-friendly information about human genetics from the U.S. National Library of Medicine

Hurler syndrome

- <https://ghr.nlm.nih.gov/condition/mucopolysaccharidosis-type-i>

Hunter syndrome

- <https://ghr.nlm.nih.gov/condition/mucopolysaccharidosis-type-ii>

Morquio syndrome

- <https://ghr.nlm.nih.gov/condition/mucopolysaccharidosis-type-iv>

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### *Resources for other more common LSDs:*

- International Advocate for Glycoprotein Storage Diseases  
[http://www.ismrd.org/glycoprotein\\_diseases](http://www.ismrd.org/glycoprotein_diseases)
- Lysosomal Disease Network <http://www.lysosomal diseases network.org/>
- National Gaucher Foundation <http://www.gaucherdisease.org/>
- National Tay-Sachs and Allied Diseases Association - Lysosomal Storage Disorders  
<https://www.ntsad.org/index.php/chart-of-allied-diseases>
- Niemann-Pick Disease Foundation, Inc. <http://nnpdf.org/overview/>
- United Pompe Association <https://www.unitedpompe.com/>

## 8. Meet a Child with MPS – “Tales of Travis”

“Travis is a kind-hearted and extremely thoughtful boy,” says his mother Steph. She shares that when he is playing soccer and someone is injured, “he is the first one to rush over and see if the other boy is okay.” She has so many positive things to say about her son! “He is also careful about other people’s feelings, he has a great sense of humor, and he is extremely brave through all of his medical procedures.” He is a busy little guy, involved in soccer, swimming, skiing, musical theatre and his favorite past time – Legos!



Travis was diagnosed with Attenuated MPS I when he was 2 ½ years old. They never suspected anything but he did have a lot of ear infections. While getting his pre-op physical for placement of ear tubes, the physician noticed some small differences and thought he might have MPS based on the ear infections, umbilical hernia, and a few other findings. She recommended they see a geneticist and the geneticist made the definitive diagnosis.

Travis started in early intervention right in his home, seeing speech, occupational, and physical therapists every week. He started in preschool 4 days per week and is now a robust second grader at age 8. “You would never know he has these medical issues,” says Steph. He has an IEP and sometimes it is hard for school staff to remember he has these medical and developmental challenges. But he is fully included in school and gets his services blended into typical parts of his day. “The only 1:1 time he has is for writing support.” He does have accommodations in the classroom to help him with focus and organization as he has an additional diagnosis of ADHD.

Travis goes to visit friends on play dates and birthday parties. Although a bit shy, he likes playing with his friends in the neighborhood and doesn’t have to take precautions. Steph said he is starting to notice his differences though, for instance he can’t throw the ball quite like his friends or do jumping jacks as well as he struggles with coordination. His condition affects his range of motion in his shoulder and hips, and he has fine motor challenges with his hands and wrists. She explained that he knows a bit about his condition, that he is missing an enzyme and he needs it to help his body stay strong and healthy.

He has a port placed so he can have his weekly enzyme infusions at home, working it into his family’s busy life activities. “We usually do the 4-hour infusion on Sunday nights and he is completely portable while the infusion is happening. We just have to make sure he is quiet so the needle doesn’t come out,” Steph explained. She puts the needle in his port and the medicine can go in a backpack so he can walk around during the infusion. “It was life-changing to be able to do this at home and not have to go to the hospital any longer for the infusions.”

Besides the infusions, Travis has had other medical procedures and operations. He has had 4 sets of ear tubes placed, hernia repair, port placement and two carpal tunnel surgeries. He sees every specialist once or twice a year.

Steph knows that the buildup of waste products in the cells can have a long term effect on his joints, especially his spine, but is cautiously optimistic that his early start on treatment will bode well for him in the future.

Steph got a quote from Travis: "My medical issues are no fun, but I am hopeful there will be a cure sometime soon. In the mean time I am happy to have fun playing with my toys and my brother and sister."

#### **Steph's Advice to Parents:**

- This condition impacts every child differently – no two children are alike and the presentation is different in each person.
- Since it is rare and complicated, it is hard to get answers but the MPS Society is the best resource. (Steph is a parent volunteer who speaks with parents of newly diagnosed children.)

#### **Steph's Advice to Teachers:**

- Children with MPS I can look like every other child. The issues aren't visually apparent in all presentations of the disease. Treating them like "typical" children is great....BUT, you really need to understand the medical conditions and read the IEP.
- Many needs are not obvious so make sure you read up on the diverse components of this condition.
- There is nothing to "worry about" in school. You just need to understand their needs well and make appropriate accommodations.