

## **MPS (Hurler/Hunter/Morquio) For Healthcare Providers**

This is a customized health care provider version of our website. Please visit the main website to find more comprehensive information for families and schools ([www.gemssforschools.org](http://www.gemssforschools.org)).

Mucopolysaccharidoses are a group of conditions also known as lysosomal storage disorders. Enzyme deficiencies lead to the accumulation of glycosaminoglycans (long chains of sugar molecules used in the building of connective tissues). This accumulation leads to progressive multi-organ dysfunction. The types of MPS are based upon the substance that builds up and the clinical spectrum is very broad and varies from mild to severe. 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

### Physical characteristics and/or symptoms

*Note: With any of the types of Mucopolysaccharidoses (MPS) disorders, not all people will have all of the features listed below.*

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

- **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
  - 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
    - Possible learning disabilities
    - Sleep apnea
    - Hernias
    - Progressive pulmonary disease
    - Hearing loss (moderate to severe)
    - Cardiac valvular disease
    - Short stature
    - Hepatomegaly
    - Corneal clouding can lead to significant vision disability

### **Mucopolysaccharidosis Type II (MPS Type II): Hunter syndrome**

- 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

### **Mucopolysaccharidosis Type IV (MPS 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 intellectual disability

NOTE: Enzyme Replacement Therapy (ERT) is being or has been developed for all types of MPS. Types I, II and IV all have well established ERT protocols. The treatment requires frequent infusions, is expensive and may not be available at all medical centers. The infusion protocols should be initiated and administered by a physician and at a center familiar with the treatment. Hematopoietic Stem Cell Transplantation (HSCT) is a treatment primarily used in severe Type I and in Types VI and VII.

## Recommended Routine Surveillance

- Annual assessment by orthopedist to assess joint mobility
- Neurological evaluation
- Physical therapy to monitor carpal tunnel syndrome
- Cardiac surveillance
- ENT surveillance
- Ophthalmology with assessment of corneal clouding and retinal function
- Routine dental care

## Emergency Protocols

There are no specific emergency protocols for this particular condition as it is not typically associated with episodes of sudden and serious medical decompensation. However, the infusion therapies carry with them risks of reaction to the infusate and therefore need to be monitored carefully.

- Emergencies should be handled as with any child.

## Specialists Who May Be Involved

Follow up is need on a case-by-case basis. A multidisciplinary team approach to best meet the child's individual needs is recommended.

- Cardiologist
  - Cardiomyopathy
  - Mitral and aortic valve regurgitation/stenosis
  - Arrhythmia
- Developmental specialists
  - Speech therapy
  - Physical therapy, particularly to preserve range of motion in joints
  - Occupational therapy
- ENT
  - Hearing loss
  - Tonsil/adenoid enlargement
  - Sleep apnea
- Gastroenterologist
  - Inguinal hernias
  - Diarrhea/constipation
- Geneticist/Genetic Counselor:
  - Diagnosis
  - Coordination of care – often the geneticist is the specialist managing the ERT or HSCT
  - Genetic risk for family
  - Clinical trials
- Neurologist
  - Hydrocephalus
  - Arachnoid cysts
- Ophthalmology surveillance:
  - Corneal clouding
- Orthopedist
  - Progressive skeletal dysplasia
  - Spinal cord compression
  - Kyphosis
  - Scoliosis
  - Back pain
- Pulmonologist
  - Progressive pulmonary disease

## Sample Forms

- Sample paragraph to be used for Letters of Medical Necessity or Letters to the school:

*My patient \_\_\_\_\_ has been diagnosed with a form of Mucopolysaccharidosis (MPS). MPS vary in severity but are characterized by abnormalities of the brain, eyes, heart and developmental delays. Medical complications with MPS include management of progressive skeletal dysplasia, corneal clouding, hearing loss, GI issues, hydrocephalus, cardiac disease, and respiratory disease. In addition, \_\_\_\_\_ receives weekly infusions that require several hours each time. Because of these, \_\_\_\_\_ needs the following accommodations.*

## Seven Important Aspects of School Life

“[MPS \(Hurler/Hunter/Morquio\) at a Glance](#)” will help you talk with parents and schools about:

- Medical / Dietary Needs
- Education Supports
- Behavior & Sensory Supports
- Physical Activity, Trips, Events
- School Absences & Fatigue
- Emergency Planning
- Transitions



## Resources

### Gene Reviews:

#### **Mucopolysaccharidosis Type I: Hurler, Hurler-Scheie, Scheie**

<https://www.ncbi.nlm.nih.gov/books/NBK1162/>

#### **Mucopolysaccharidosis Type II: Hunter**

<https://www.ncbi.nlm.nih.gov/books/NBK1274/>

#### **Mucopolysaccharidosis Type IVA: Morquio syndrome**

<https://www.ncbi.nlm.nih.gov/books/NBK148668/>

#### **Lysosomal Storage Disorders – general information**

<https://www.ncbi.nlm.nih.gov/books/NBK6177/>

#### **National MPS Society**

<https://mpssociety.org/>