



How to piece together the diagnosis of MPS I in your pediatric patients

If you see seemingly unrelated signs and symptoms in your patients, including hernia, frequent ear infections, joint contractures, and corneal clouding, consider mucopolysaccharidosis type I (MPS I).

Early identification of symptoms is key in pediatric patients with MPS I.

SHANNAE

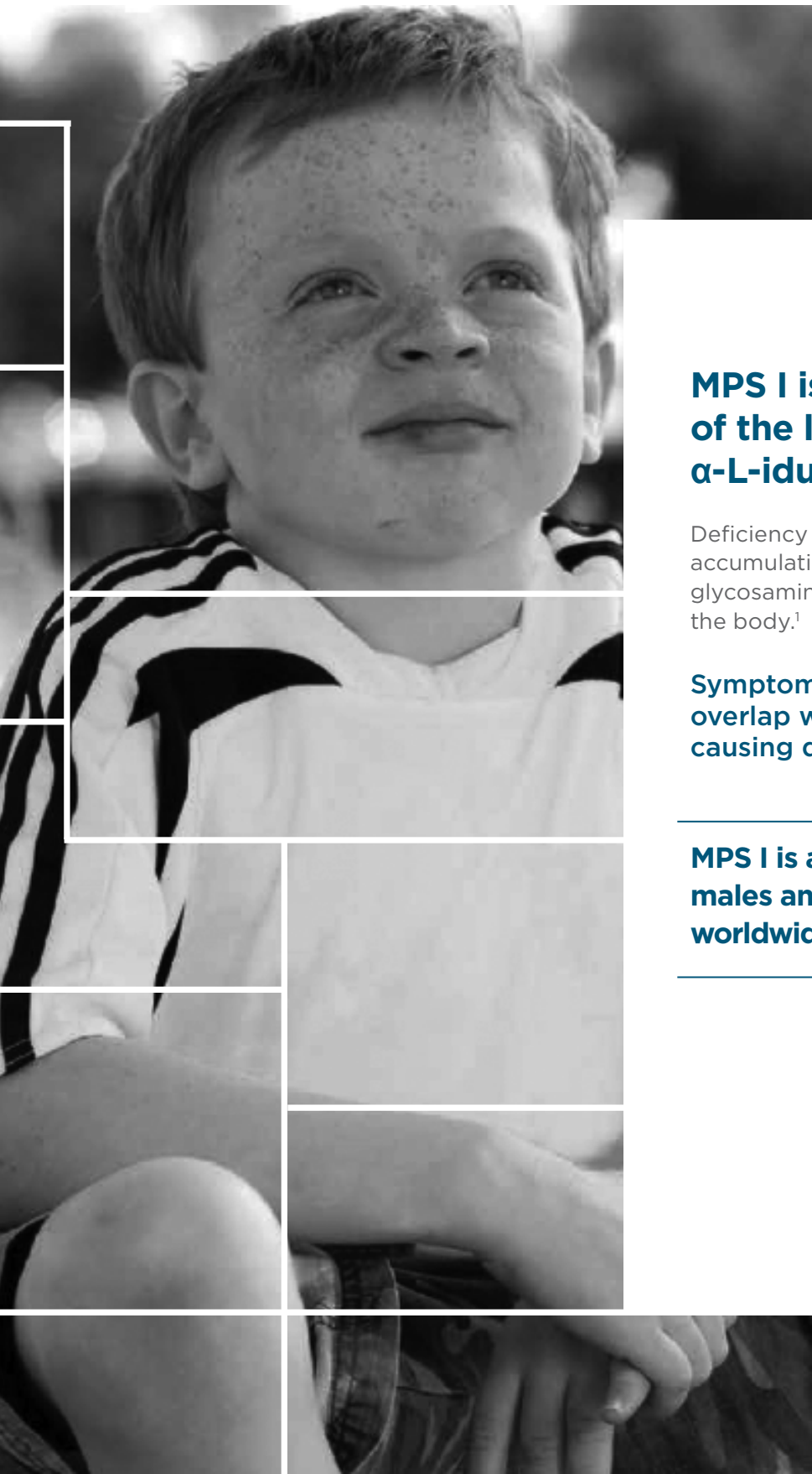
Country: Australia

Age at first symptom onset: 6 months old

Age at diagnosis: 4 years old



MPS I is a rare, progressive, multisystemic, and often under-recognized disease¹



MPS I is caused by the deficiency of the lysosomal enzyme α -L-iduronidase¹

Deficiency of this enzyme results in the progressive accumulation of non-degraded material (called glycosaminoglycans, or GAGs) in cells throughout the body.¹

Symptoms are highly variable and often overlap with other pediatric illnesses, causing delays in diagnosis.^{2,3}

MPS I is autosomal recessive and affects males and females equally. The estimated worldwide incidence of MPS I is 1:100,000.¹

Ben

Country: Australia

Age at first symptom onset: 6 months old

Age at diagnosis: 5 years old

MPS I has a wide spectrum of severity³⁻⁵

Hurler, Hurler-Scheie, and Scheie syndromes³⁻⁵

MPS I includes a highly heterogeneous spectrum of severity, signs, symptoms and affected organ systems, with symptom onset often in early childhood.⁶

MPS I Spectrum of Disease

Severe
Rapid Progression

Attenuated
Slower Progression



Age at Diagnosis



0.2-7
years

MPS I H
“Hurler”



0.2-36
years

MPS I HS
“Hurler-Scheie”



2-54
years

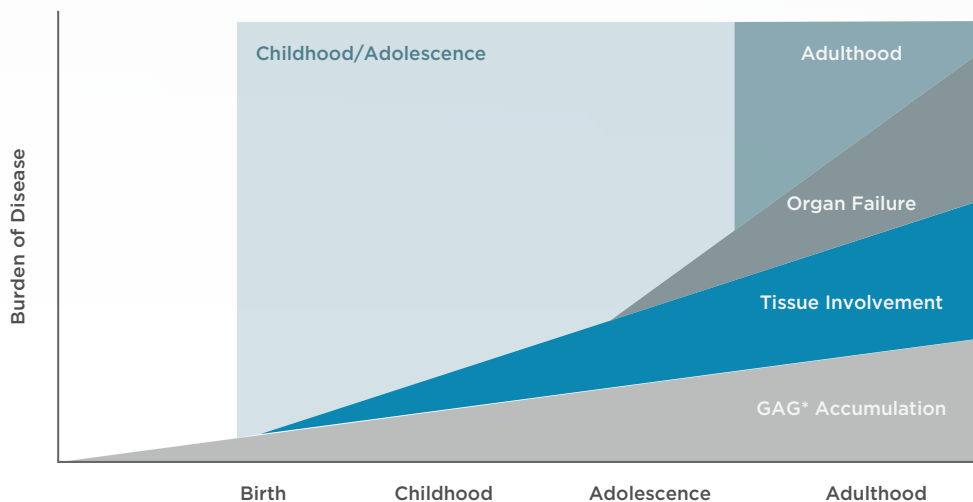
MPS I S
“Scheie”

Attenuated MPS I disease progression^{3,5-7,10}

Patients with the attenuated phenotype may have an average life span but may be burdened by considerable morbidity or, in some cases, may face premature death.^{3,4} Diagnostic delays among children with attenuated MPS I are unfortunately common. The progressive accumulation of GAGs can cause cardiovascular complications and respiratory disease, which are the leading morbidities associated with early death in patients with MPS I.³

Attenuated disease is often misdiagnosed

The varying disease presentation and seemingly unrelated signs and symptoms can lead to years of misdiagnoses and diagnostic delays in attenuated MPS I patients.^{6,7,10}



*GAG = glycosaminoglycan.

Could it be MPS I?

Consider the specific constellation of symptoms

Children presenting with **corneal clouding, short stature, restricted joint mobility, joint contractures, and/or recurrent hernias** should be tested for MPS I.

Possible differential diagnosis and syndromes^{2,3,8}

- Idiopathic short stature
 - Juvenile idiopathic arthritis
 - Autoimmune disease
 - Polymyositis
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Country: Australia

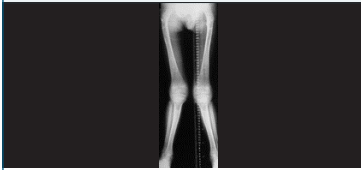



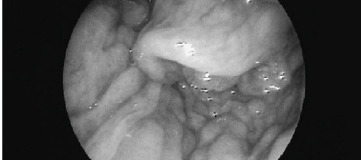
Age at first symptom onset: 6 months old

Age at diagnosis: 4 years old



What are the most common symptoms?

MPS I affects multiple body systems and is often characterized by clusters of symptoms¹

Clinical Depiction	Most common signs and symptoms ^{2,7}	Distinctive features ^{1,9}
	Joint stiffness/pain	<ul style="list-style-type: none">• Restricted shoulder flexion• Contractures without inflammation• Toe walking
	Poor hand function	<ul style="list-style-type: none">• Joint contractures• Trigger finger• Carpal tunnel syndrome
	Recurrent hernias	<ul style="list-style-type: none">• Multiple hernia repairs, especially after age 2.5 years⁹
	Poor vision	<ul style="list-style-type: none">• Corneal clouding (bilateral)• Photophobia
	Frequent ENT infections	<ul style="list-style-type: none">• Recurrent otitis media, sinusitis• Adenoidectomy/tonsillectomy procedures before age 5⁹

*Additional manifestations can include obstructive airway disease, sleep apnea, coarse facial features, growth deficiencies, skeletal abnormalities, spinal cord compression, hepatomegaly/splenomegaly, cardiac disease, and hearing loss.

You can piece together the diagnosis of MPS I



**If you suspect
MPS I disease,
refer your patient
to a Metabolic
Specialist or
Geneticist
immediately.**

To learn more about signs, symptoms, testing, and referrals, please visit www.hcp.mps1disease.com



NICK

Age at first symptom onset: 2 years old

Age at diagnosis: 4 years old

MPS I is rare, progressive, and often goes unrecognized

Early diagnosis can make a difference

Accurate early diagnosis is imperative to facilitate appropriate disease management.⁴

**2-4
years**

Average delay between symptom presentation and diagnosis in attenuated MPS I patients.¹⁰

**4-8
years**

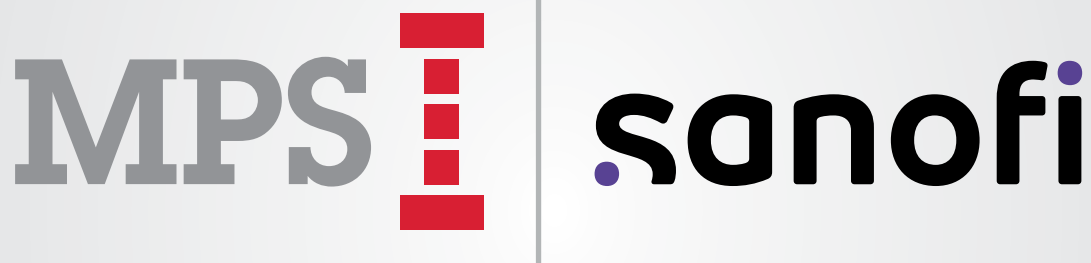
Average delay between diagnosis and initiation of disease-specific management in attenuated MPS I patients.¹⁰

TAMARA

Age at first symptom onset: 6 years old

Age at diagnosis: 16 years old





References

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