

# MUCOPOLYSACCHARIDOSIS I

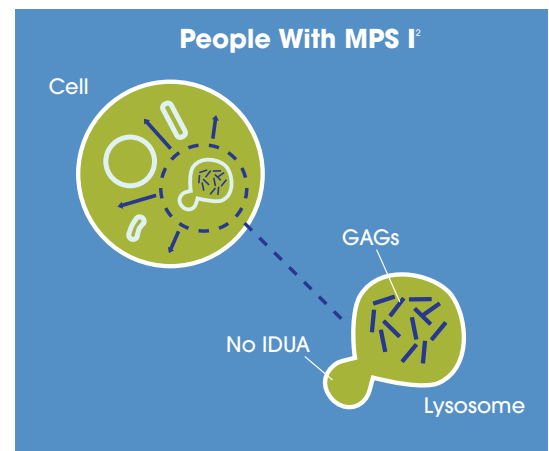
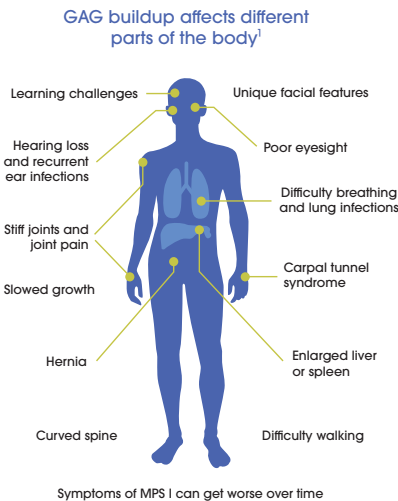
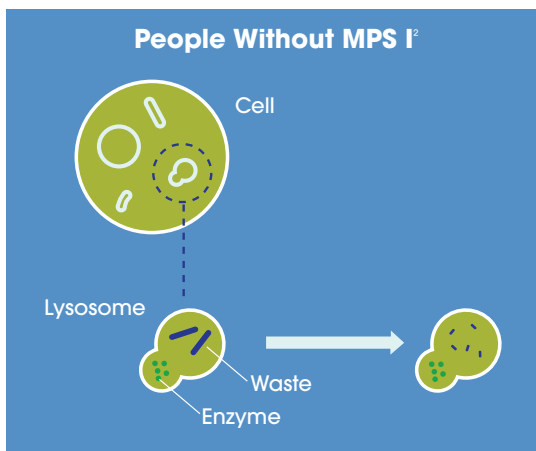
## DIAGNOSTIC TESTING FOR MPS I DISEASE

Mucopolysaccharidosis I (MPS I) is an inherited, multi-system progressive disorder caused by a deficiency of the lysosomal enzyme  $\alpha$ -L-iduronidase, leading to a buildup of its major substrates, the glycosaminoglycans (GAGs) dermatan and heparan sulfate, and resulting in developmental delay and regression, plus respiratory, cardiac, and musculoskeletal dysfunction.<sup>1</sup>



MPS I affects **1 in 100,000 people**<sup>2</sup>

**TEST NOW** and impact diagnostic delays, which may last up to 7 years.<sup>2</sup>



There are **3 types of MPS I** and they affect each person differently<sup>1</sup>

### Severe

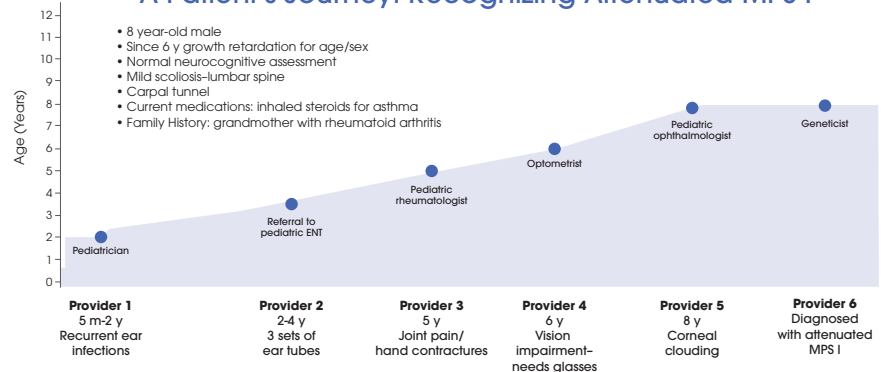
**Hurler**  
(60.9%)<sup>2</sup>

### Attenuated

**Hurler-Scheie**  
(23.0%)<sup>2</sup>

**Scheie**  
(12.9%)<sup>2</sup>

### A Patient's Journey: Recognizing Attenuated MPS I\*



### Symptom Presentation

\*Representative case, not a real patient.

A definitive diagnosis of MPS I is confirmed by an enzyme assay indicating low or absent  $\alpha$ -L-iduronidase enzyme, supported by DNA sequencing of the *IDUA* gene (indicating 2 mutations) and elevated urine GAGs.<sup>1</sup>

## A delay in diagnosis may result in delayed disease management<sup>2</sup>

Physicians are directed to contact the individual testing programs and/or laboratories for information on obtaining test requisitions, sample requirements, turnaround time, and the specific details of the services provided.

For commercial lab testing, coding accurately and obtaining reimbursement are the responsibility of the provider submitting a claim for the item or service.

SEE LISTING OF LABS ON REVERSE SIDE

## Testing Options for MPS I

Sanofi Genzyme does not review or control the content of non-Sanofi Genzyme websites. These listings do not constitute an endorsement by Sanofi Genzyme of information provided by any other organizations. The following is a selection of laboratories offering both IDUA enzyme assay ( $\alpha$ -L-Iduronidase) and IDUA sequencing and those offering an MPS enzyme panel. This is not an exhaustive list of labs that offer one or the other or an endorsement of any one lab. Other testing options can be found at [www.concertgenetics.com](http://www.concertgenetics.com) or [www.ncbi.nlm.nih.gov/gtr](http://www.ncbi.nlm.nih.gov/gtr). Content is current at time of printing; please check with individual laboratory before ordering. Tests may not be available in all states, so please call laboratory to confirm test availability.

Lab	Available Testing	Sample Requirements	Panel Type	Kits	Avg TAT	Mobile Blood Draw	Billing	Contact
EGL Genetics	Alpha-L-Iduronidase Enzyme	WB: 3-5 ml sodium heparin (green) tube	N/A	Blood, DBS, Saliva	7-10 d	No	Inst, Ins, Self-pay	P: 855-831-7447 W: <a href="http://www.egl-eurofins.com">www.egl-eurofins.com</a>
	IDUA Sequencing	For both del/dup and sequencing: WB: EDTA (lavender) tube (volume varies with age); Saliva (per Oragene kit)			2 wks			
	IDUA Del/Dup				4 wks			
	Qual/Quant GAGs				7-10 d			
Greenwood Genetic Center	Alpha-L-Iduronidase Enzyme	WB: 5-10 ml sodium heparin (green) tube; Serum; DBS card: 3 circles	MPS I, II, IIIA-D, IVA, IVB, VI, VII, MSD, mucopolipidosis II, III plus total GAGs and quantitative individual GAG components	Blood, DBS, Saliva	2 wks	No	Inst, Self-pay, Ins (SC residents only)	P: 800-473-9411 E: <a href="mailto:labgc@ggc.org">labgc@ggc.org</a> W: <a href="http://www.ggc.org">www.ggc.org</a>
	IDUA Sequencing	WB: 5-7 ml EDTA (lavender) tube; DBS card: 3 circles; Saliva			3 wks			
	IDUA Del/Dup	WB: 5-7 ml EDTA (lavender) tube			21 d			
	MPS 10 Enzyme Panel + GAGs	WB: 2 x 5-7 ml heparin (green) tube	3 wks					
	MPS 7 Enzyme Panel + GAGs	DBS: 5 circles	3 wks					
	Qual/Quant GAGs	3 ml random urine, frozen	MPS I, II, IIIB, IVA, IVB, VI, VII plus total GAG and quantitative individual GAG components	14 d				
LabCorp/Integrated Genetics	Alpha-L-Iduronidase Enzyme (code 800102)	WB: 2ml EDTA (lavender) or ACD (yellow) tube (note, performed by Mayo)	N/A	Blood, Buccal	7 d	Yes	Inst, Ins, Self-Pay	LabCorp Customers: P: 888-522-2677 W: <a href="http://www.labcorp.com">www.labcorp.com</a>  Integrated Customers: P: 800-848-4436 E: <a href="mailto:askIcclientervices@integratedgenetics.com">askIcclientervices@integratedgenetics.com</a> W: <a href="http://www.integratedgenetics.com">www.integratedgenetics.com</a>
	IDUA Sequencing (via Inheritest Gene-Specific NGS 451910)	WB: 3-7 ml EDTA (lavender) or ACD (yellow) tube, Buccal swab			14-21 d			
The Lantern Project (performed at PerkinElmer genomics)	Alpha-L-Iduronidase Enzyme	WB: 3-5 ml heparin (green) tube; DBS card: 5 circles	MPS I, II, IIIB, IVA, IVB, VI, VII	Blood, DBS, Saliva	3 d	Yes	No charge*	P: 866-354-2910 E: <a href="mailto:genomics@perkinelmer.com">genomics@perkinelmer.com</a> W: <a href="http://www.LanternProjectDx.com">www.LanternProjectDx.com</a>
	IDUA Sequencing (Incl Del/Dup)	WB: 3-5 ml EDTA (lavender) tube; DBS card: 5 circles; Saliva (per Oragene kit)			3 wks			
	MPS Enzyme Panel	WB: 3-5 ml EDTA (lavender) tube; DBS card: 5 circles			3 wks			
Mayo Medical Laboratories	Alpha-L-Iduronidase Enzyme	WB: 2 ml EDTA (lavender) or ACD (yellow) tube; DBS card: 2 circles	N/A	DBS (in some cases), Saliva	8-15 d	Yes	Inst (ins can be billed in some cases, Inst acct required)	P: 800-533-1710 E: <a href="mailto:mml@mayo.edu">mml@mayo.edu</a> W: <a href="http://www.mayomedicallaboratories.com">www.mayomedicallaboratories.com</a>
	IDUA Sequencing	WB: 3 ml (lavender) or ACD (yellow) tube; DBS card: 2-5 circles			14-20 d			
	Qual/Quant GAGs	3 ml first morning urine, frozen			10-18 d			
Sema4	Alpha-L-Iduronidase Enzyme	WB: 5-10 ml EDTA (lavender), ACD (yellow), or heparin (green) tube; min 3 ml	N/A	Blood, Saliva	7 d	Yes	Inst, Ins, Self-pay	P: 800-298-6470 E: <a href="mailto:clientservices@sema4.com">clientservices@sema4.com</a> W: <a href="http://www.sema4.com">www.sema4.com</a>
	IDUA Sequencing (incl Del/Dup)	WB: 2 x 5-10 ml EDTA (lavender) tubes; Saliva			10-14 d			

\*Testing is performed at no charge; local charges may apply for sample collection, processing, or shipping.

acct = account; avg TAT = average turnaround time; d = days; DBS = dried blood spot; del = deletion; dup = duplication analysis; GAGs = glycosaminoglycans; IDUA = iduronidase; incl = including; Ins = insurance; Inst = institution; min = minimum; N/A = not applicable; Qual = qualitative; Quant = quantitative; TAT = turnaround time; WB = whole blood; wks = weeks.