MUCOPOLYSACCHARDOSIS TYPE I RECOMMENDED MINIMUM EVALUATION SCHEDULE: Based upon Peer-Reviewed Publications

Mucopolysaccharidosis Type I (MPS I):

MPS I is a lysosomal storage disease that is caused by the deficiency of lysosomal enzyme alpha-L-iduronidase. Individuals with MPS I have a pathogenic variant in the *IDUA* gene that codes for the production of alpha-L-iduronidase. Alpha-L-iduronidase is needed to catabolize glycosaminoglycans (GAGs) in the lysosome. Reduced alpha-L-iduronidase activity results in the accumulation of GAGs, specifically heparan and dermatan sulfate.

Recommended Minimum Evaluation Schedule:

The listed recommendations were obtained from peer reviewed publications cited below. Physicians and health care providers will determine the assessments and their actual frequency according to the patient's individual needs.

Evaluations	Initial Evaluation	Every 6 Months	Every 12 Months	Every Other Year
General				
Demographic characteristic	X			
Patient diagnosis	X			
Medical history	X	X		
Physical examination	X	X		
General appearance	X	X		
Neurological/Central Nervous System				
CT or MRI scans of brain	X			X
MRI scans of spine	X			X
Median nerve conduction velocity	X			X
Cognitive testing (DQ/IQ)	X		X	
Auditory				
Audiometry	X		X	
Ophthalmologic				
Visual acuity	X		X	
Retinal examination	X		X	
Corneal examination	X		X	
Respiratory				
Spirometry including Forced Vital Capacity/Forced Expiratory Volume (FVC/FEV)	X	X		
Sleep study	X		X	
Cardiac				
Echocardiography	X			X
Electrocardiography	X			X
Musculoskeletal				
Skeletal survey with radiographs[1]	X			X
Gastrointestinal				
Spleen volume[2]	X			X[3]
Liver volume[2]	X			X[3]
Vital Signs and Laboratory Tests				
Height and weight	X	X		
Head circumference[1]	X	X		
Blood pressure	X	X		
Enzyme activity level	X			
Urinary glycosaminoglycan level	X	X[3]		
Urinalysis	X	X[3]		
Molecular Sequencing of the *IDUA* gene	X			
Functional Outcome Measurements				
Mucopolysaccharidosis Health Assessment Questionnaire or other tools exploring functional ability and quality of life[4]	X		X	

1) Studies are only for pediatric patients, unless determined otherwise by the treating physician.
2) The recommended method for determining organ volumes is MRI or computed tomography, to enable quantitative analysis. If it is unsafe to sedate the patient, in the opinion of the clinician, then ultrasonography may be substituted.
3) Studies are only for patients that are treated, unless determined otherwise by the treating physician.
4) Assessment may not be possible for uncooperative patients or patients younger than 5 to 6 years of age.

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Musculoskeletal-Specific Recommended Minimum Imaging Evaluation Schedule:

The listed recommendations were obtained from peer reviewed publications cited below. Physicians and health care providers will determine the assessments and their actual frequency according to the patient's individual needs.

Imaging Studies	Initial Evaluation	Every 12 Months	Comments
Cervical spine			
Radiographs - Flexion/extension lat	X		Follow neurological exam yearly, with repeat radiographs every 3-5 years
MRI	X	X	
Thoracolumbar spine			
Radiographs - AP/lat 36-inch Cassette	X	X	Less frequent as growth slows
MRI	X		Further exams warranted only with changes in neurological exam
CT Scan – primarily warranted for pre-operative evaluation			Primarily warranted for pre-operative evaluation
Hips/Pelvis			
Radiograph - AP pelvis	X	X	
Lower extremities			
Radiograph - Standing AP	X		Further exams warranted based on clinical exam
Skeletal Survey			
Radiograph - AP/lat skull	X		
Radiograph - AP/lat spine	X		
Radiograph - AP pelvis	X		
Radiograph - AP forearms	X		
Radiograph - AP hands	X		
Radiograph - AP feet	X		
Radiograph - Lat cervical spine	X		

Lat – Lateral; AP - Anterior-Posterior

Cardiac-Specific Recommended Minimum Imaging Evaluation Schedule:

The listed recommendations were obtained from peer reviewed publications cited below. Physicians and health care providers will determine the assessments and their actual frequency according to the patient's individual needs.

Evaluations	Initial Evaluation	Every 1 to 2 Years
ECG		
Surface 12-lead	X	X
Holter 24-hours	X	X
ECHO		
2D Transthoracic	X	X
M Mode	X	X
Doppler	X	X
Color Doppler	X	X
Tissue Doppler	X	X

References:

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