

## Ongoing multisystemic assessments in patients with Morquio A (MPS IVA) <sup>1</sup>

<b>Morquio A manifestations</b>	<b>Assessments</b>	<b>Frequency</b>
<p><b>Musculoskeletal</b> Skeletal and joint abnormalities are the most apparent and prevalent disease manifestations of Morquio A syndrome</p>	<ul style="list-style-type: none"> <li>Standardized upper extremity function test, radiographs</li> <li>Hips/pelvis, lower extremities</li> </ul>	<ul style="list-style-type: none"> <li>At diagnosis/baseline, annually</li> <li>At diagnosis/baseline, as clinically indicated</li> </ul>
<p><b>Respiratory</b> Respiratory impairment is the leading cause of morbidity and mortality in patients with Morquio A and can be due to an obstructive or restrictive disease</p>	<ul style="list-style-type: none"> <li>Forced vital capacity</li> <li>Maximum voluntary ventilation</li> <li>Respiratory rate</li> <li>Oxygen saturation</li> </ul>	<ul style="list-style-type: none"> <li>At diagnosis/baseline, annually</li> </ul>
<p><b>Neurological</b> Patients with Morquio A can develop neurological symptoms due to myelopathy secondary to spinal cord compression</p>	<ul style="list-style-type: none"> <li>Neurological exam</li> <li>Plain radiograph</li> <li>MRI scan</li> <li>CT scan</li> </ul>	<ul style="list-style-type: none"> <li>At diagnosis/baseline, every visit (minimally, every 6 months)</li> <li>At diagnosis, every 1 to 3 years</li> <li>At diagnosis, annually</li> <li>As clinically indicated</li> </ul>
<p><b>Cardiovascular</b> It is important to be aware that the high heart rate in patients with Morquio A is needed to compensate for a small cardiac stroke volume</p>	<ul style="list-style-type: none"> <li>Electrocardiogram</li> <li>Echocardiogram</li> <li>Heart rate</li> </ul>	<ul style="list-style-type: none"> <li>At diagnosis, every 1 to 3 years, as clinically indicated</li> <li>At diagnosis, every 2 to 3 years, as clinically indicated</li> <li>At diagnosis, annually</li> </ul>
<p><b>Ophthalmological</b> Diffuse corneal clouding and refractive error problems (astigmatism, myopia, and hyperopia) are very common findings in patients with Morquio A and may lead to reduced visual acuity and photosensitivity</p>	<ul style="list-style-type: none"> <li>Refractive error and intraocular pressure</li> </ul>	<ul style="list-style-type: none"> <li>At diagnosis, as clinically indicated</li> </ul>
<p><b>Audiological</b> Neurosensory or mixed conductive and neurosensory hearing loss commonly develop in patients with Morquio A in the first decade of life</p>	<ul style="list-style-type: none"> <li>Multimodal hearing assessments</li> </ul>	<ul style="list-style-type: none"> <li>At diagnosis, annually</li> </ul>
<p><b>Abdominal</b> Abdominal manifestations of Morquio A include umbilical, inguinal, or bilateral diaphragmatic hernias, hepatomegaly, splenomegaly (less common), and other gastrointestinal disorders (eg, chronic constipation, diarrhea)</p>	<ul style="list-style-type: none"> <li>Assessments of gastrointestinal problems</li> </ul>	<ul style="list-style-type: none"> <li>As clinically indicated</li> </ul>
<p><b>Dental</b> Patients with Morquio A tend to have small, widely spaced teeth, often with thin, structurally weak enamel and small pointed cusps, spade-shaped incisors, pitted buccal surfaces, and other developmental abnormalities of primary and permanent dentition</p>	<ul style="list-style-type: none"> <li>Evaluation of oral health</li> </ul>	<ul style="list-style-type: none"> <li>At diagnosis, annually</li> </ul>
<p><b>Endurance</b> Patients with Morquio A may show reduced endurance due to impaired cardiac, respiratory, musculoskeletal, and/or neurological function, which may impact significantly on functional status/mobility and QoL</p>	<ul style="list-style-type: none"> <li>6MWT</li> </ul>	<ul style="list-style-type: none"> <li>At diagnosis, annually, before and regularly after initiation of ERT</li> </ul>
<p><b>QoL</b> Many factors may affect QoL in patients with Morquio A, including reduced endurance or mobility, difficulties in activities of daily living, dependence on caregivers, frequent surgical interventions, pain, and fatigue</p>	<ul style="list-style-type: none"> <li>Reproducible, age-appropriate QoL Questionnaires (eg, EQ-SD-SL)</li> </ul>	<ul style="list-style-type: none"> <li>At diagnosis, annually</li> </ul>

Adapted from Hendriksz, *Am J Med Genet Part A*, 2014.

Abbreviations: 6MWT, 6-minute walk test; CT, computed tomography; ERT, enzyme-replacement therapy; MPS, mucopolysaccharidosis; MRI, magnetic resonance imaging; QoL, quality of life.

**Reference:** 1. Hendriksz CJ, Berger KI, Giugliani R, et al. International guidelines for the management and treatment of Morquio A syndrome. *Am J Med Genet A*. 2014;9999A:1-15. doi:10.1002/ajmg.a.36833.