

Ongoing multisystemic assessments in patients with Morquio A¹

Morquio A manifestations	Assessments	Frequency
<p>Musculoskeletal Skeletal and joint abnormalities are the most apparent and prevalent disease manifestations of Morquio A syndrome</p>	<ul style="list-style-type: none"> Standardized upper extremity function test, radiographs Hips/pelvis, lower extremities 	<ul style="list-style-type: none"> At diagnosis/baseline, annually At diagnosis/baseline, as clinically indicated
<p>Respiratory 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"> Forced vital capacity Maximum voluntary ventilation Respiratory rate Oxygen saturation 	<ul style="list-style-type: none"> At diagnosis/baseline, annually
<p>Neurological Patients with Morquio A can develop neurological symptoms due to myelopathy secondary to spinal cord compression</p>	<ul style="list-style-type: none"> Neurological exam Plain radiograph MRI scan CT scan 	<ul style="list-style-type: none"> At diagnosis/baseline, every visit (minimally, every 6 months) At diagnosis, every 1 to 3 years At diagnosis, annually As clinically indicated
<p>Cardiovascular 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"> Electrocardiogram Echocardiogram Heart rate 	<ul style="list-style-type: none"> At diagnosis, every 1 to 3 years, as clinically indicated At diagnosis, every 2 to 3 years, as clinically indicated At diagnosis, annually
<p>Ophthalmological 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"> Refractive error and intraocular pressure 	<ul style="list-style-type: none"> At diagnosis, as clinically indicated
<p>Audiological 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"> Multimodal hearing assessments 	<ul style="list-style-type: none"> At diagnosis, annually
<p>Abdominal 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"> Assessments of gastrointestinal problems 	<ul style="list-style-type: none"> As clinically indicated
<p>Dental 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"> Evaluation of oral health 	<ul style="list-style-type: none"> At diagnosis, annually
<p>Endurance 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"> 6MWT 	<ul style="list-style-type: none"> At diagnosis, annually, before and regularly after initiation of ERT
<p>QoL 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"> Reproducible, age-appropriate QoL Questionnaires (eg, EQ-SD-SL) 	<ul style="list-style-type: none"> At diagnosis, annually

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

Abbreviations: 6MWT, 6-minute walk test; CT, computed tomography; MRI, magnetic resonance imaging; QoL, quality of life.