

Clinical manifestations of MPS VI

Organ system	Complications
Ear, nose, throat, and respiratory ^{5,6}	<p>GAG accumulation in the oropharynx and airway, combined with typical dysmorphic features and restrictive lung disease, can cause</p> <ul style="list-style-type: none"> • Thickening of the nose, lips, and tongue • Severe hearing impairment • Recurrent otitis media • Narrow trachea and excessive and thickened secretions • Obstructive sleep apnea <ul style="list-style-type: none"> • Recurrent pulmonary infections and pneumonia • Skeletal problems and reduced lung function and volume • The need for a respiratory device such as a CPAP machine or surgical insertion of an endotracheal tube to aid breathing
Cardiovascular ^{2,4,7}	<p>Cardiovascular abnormalities are a major cause of morbidity and mortality among patients with MPS VI</p> <ul style="list-style-type: none"> • Heart murmurs • Mitral and aortic valve degeneration • Electrocardiographic abnormalities <ul style="list-style-type: none"> • Coronary artery disease • Systemic vascular narrowing and hypertension • Cardiomyopathy
Skeletal ^{2,4,5}	<p>Skeletal deformities and other clinical manifestations are seen in patients with MPS VI</p> <ul style="list-style-type: none"> • Dysostosis multiplex • Spinal cord or nerve root injury • Coarse facial features • Short stature • Joint abnormalities <ul style="list-style-type: none"> • Chest rib cage restriction • Growth impairment • Profound dwarfism • Limited mobility • Claw hands
Ophthalmic ^{2,4,8,9}	<p>Visual impairment occurs in ~40% of patients with MPS VI</p> <ul style="list-style-type: none"> • Most patients are farsighted • Corneal clouding occurs in 95% of all patients • Retinopathy <ul style="list-style-type: none"> • Optic nerve abnormalities • Ocular hypertension and glaucoma
Dental ¹⁰	<p>Dental abnormalities are common in patients with MPS VI and include</p> <ul style="list-style-type: none"> • Mandibular condylar hypoplasia • Malposition of unerupted teeth • Large dental follicles <ul style="list-style-type: none"> • Anterior open bite • Maxillary constriction • Taurodontism
CNS/PNS ^{2,4}	<p>MPS VI involves no direct impairment of CNS activity, such that the patient's intelligence is typically normal despite the great physical disease burden</p> <ul style="list-style-type: none"> • GAG accumulation causes carpal tunnel syndrome, intracranial pressure, and progressive compressive myelopathy • Loss of dexterity and fixed flexion <ul style="list-style-type: none"> • CNS stenosis and spinal cord compression • Severe pain caused by compressed or traumatized nerves and nerve roots
Organ systems ²	<p>The abdomen in patients with MPS VI is large and protruding due to the enlarged liver and spleen, often with the presence of inguinal and/or umbilical hernia</p>