

Clinical manifestations of MPS VI

Organ system	Complications
<p>Ear, nose, throat, and respiratory^{1,2}</p>	<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 • 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
<p>Cardiovascular³⁻⁵</p>	<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 • Coronary artery disease • Systemic vascular narrowing and hypertension • Cardiomyopathy
<p>Skeletal²⁻⁴</p>	<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 • Chest rib cage restriction • Growth impairment • Profound dwarfism • Limited mobility • Claw hands
<p>Ophthalmic^{3, 4, 6, 7}</p>	<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 • Optic nerve abnormalities • Ocular hypertension and glaucoma
<p>Dental⁸</p>	<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 • Anterior open bite • Maxillary constriction • Taurodontism
<p>CNS/PNS^{3,4}</p>	<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 • CNS stenosis and spinal cord compression • Severe pain caused by compressed or traumatized nerves and nerve roots
<p>Organ systems³</p>	<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>

Abbreviations: CNS, central nervous system; CPAP, continuous positive airway pressure; GAG, glycosaminoglycan; MPS, mucopolysaccharidosis; PNS, peripheral nervous system.

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