## Clinical manifestations of MPS VI

<table>
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<th>Organ system</th>
<th>Complications</th>
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| **Ear, nose, throat, and respiratory**<sup>1,2</sup> | GAG accumulation in the oropharynx and airway, combined with typical dysmorphic features and restrictive lung disease, can cause:  
• 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 |
| **Cardiovascular**<sup>3-5</sup> | Cardiovascular abnormalities are a major cause of morbidity and mortality among patients with MPS VI:  
• Heart murmurs  
• Mitral and aortic valve degeneration  
• Electrocardiographic abnormalities  
• Coronary artery disease  
• Systemic vascular narrowing and hypertension  
• Cardiomyopathy |
| **Skeletal**<sup>2-4</sup> | Skeletal deformities and other clinical manifestations are seen in patients with MPS VI:  
• 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 |
| **Ophthalmic**<sup>3,4,6,7</sup> | Visual impairment occurs in ~40% of patients with MPS VI:  
• Most patients are farsighted  
• Corneal clouding occurs in 95% of all patients  
• Retinopathy  
• Optic nerve abnormalities  
• Ocular hypertension and glaucoma |
| **Dental**<sup>4</sup> | Dental abnormalities are common in patients with MPS VI and include:  
• Mandibular condylar hypoplasia  
• Malposition of unerupted teeth  
• Large dental follicles  
• Anterior open bite  
• Maxillary constriction  
• Taurodontism |
| **CNS/PNS**<sup>3,4</sup> | MPS VI involves no direct impairment of CNS activity, such that the patient’s intelligence is typically normal despite the great physical disease burden:  
• 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 |
| **Organ systems**<sup>3</sup> | 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 |

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

**References:**