

# EAR, NOSE, AND THROAT SPECIALIST GUIDE

Ear, nose, and throat disorders affect >90% of MPS patients and appear in the early stages of the disease.<sup>1</sup> Most MPS II patients have recurrent infections, and nearly all experience progressive hearing loss.<sup>2</sup>

## Pathophysiology & Clinical Consequences

Changes to the soft tissues cause most of the respiratory problems associated with MPS II.<sup>3</sup> Normal breathing is inhibited because of the skeletal manifestation of Hunter syndrome, such as stiffness of the chest wall, abnormally shaped ribs, and restricted movement of the temporomandibular joints, and also as a result of the abdominal distention caused by an enlarged liver and spleen.<sup>2,3</sup>

Additionally, the enlarged tongue, hypertrophic adenoids and tonsils, and skeletal changes in the jaw and neck limit the opening of the mouth and can affect patients' ability to chew and swallow.<sup>2,3</sup> Upper airway obstruction is a major contributor to the morbidity and mortality of MPS II patients.<sup>3</sup>

Hearing loss is caused by both conductive and sensorineural deficits and middle-ear effusion.<sup>2</sup> Chronic otitis media, otosclerosis, and tympanomastoid abnormalities contribute to conductive hearing loss.<sup>2,3</sup> Hearing loss is nearly universal in MPS II patients and can contribute to behavioral problems and learning difficulties.<sup>3</sup>

## Management

Diagnosis of airway obstruction may be aided by a comprehensive evaluation of medical history, physical examination, and imaging studies.<sup>3</sup> A first step in managing airway involvement often includes the removal of obstructions, including the tonsils and adenoids, however, due to the progressive nature of airway involvement at a later stage, it can be important to consider continuous positive airway pressure, ventilation, or tracheostomy.<sup>2,3</sup>

## Assessment Tools

Spirometry may be used to monitor progressive changes in respiratory function, although as this test requires patient cooperation it is limited in very young or severe patients.<sup>3</sup>

An overnight sleep study, conducted at a hospital or at home, can be utilized to evaluate the severity of, and effect of treatment on, obstructive sleep apnea.<sup>3</sup> Bronchoscopy can be used for a more thorough evaluation of respiratory involvement as it can provide an accurate observation of the dynamic obstruction that occurs during breathing.<sup>3</sup>

1. Bianchi PM et al. *Ital J Pediatr* 2018; 44(Suppl 2): 127. 2. Martin R et al. *Pediatrics* 2008; 121(2): e377-386. 3. Muenzer J et al. *Pediatrics* 2009; 124(6): e1228-1239.

**HUNTER  
SYNDROME IS A  
MULTISYSTEMIC  
AND PROGRESSIVE  
DISEASE**

**Nash, age 6**



**OTITIS  
MEDIA**



**CHRONIC  
RHINORRHEA**



**ENLARGED TONSILS  
AND ADENOIDS**

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