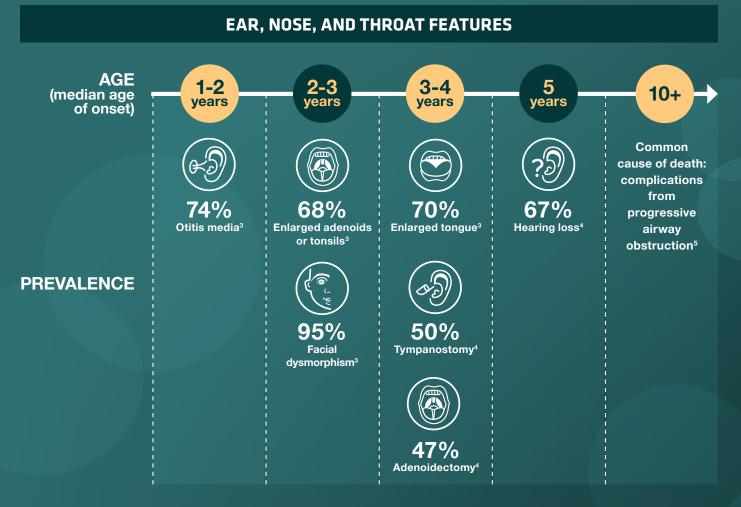
CONNECT THE DOTS: THINK HUNTER SYNDROME

As an ENT specialist, you may be one of the first to suspect Hunter syndrome (mucopolysaccharidosis II [MPS II]), a progressive genetic disease which affects almost exclusively males.¹ ENT manifestations are very common in MPS II, and are often the earliest clinical manifestations of the disease.² By recognizing key clinical features, you can help identify and refer patients who may be at risk from this disease.



Other symptoms that may be seen by age 2-4 include:⁶

Respiratory problems, including noisy breathing and snoring, and chronic rhinorrhea. Hunter syndrome is highly heterogenous. Symptoms vary in type, severity, age of onset, and progression.⁵

ACT EARLY If you suspect Hunter syndrome, refer your patient to a metabolic geneticist for an accurate diagnosis.

Visit **hunterpatients.com/healthcare-professionals** for more information about Hunter syndrome



Silas, age 6 months

Silas, age 4.

OVERVIEW OF HUNTER SYNDROME

Hunter syndrome is a progressive genetic disease

- X-linked recessive genetic disorder¹ affecting approximately 1 in 162,000 live births⁷, almost exclusively males¹
- Lysosomal storage disorder caused by the deficiency or absence of iduronate-2-sulfatase (I2S).^{1,5} The widespread pathological lysosomal storage of glycosaminoglycans (GAGs) leads to progressive damage and dysfunction in cells, tissues, and organs throughout the body⁵
- A newborn infant who has Hunter syndrome may appear unaffected in the first months and years of life, but between the ages of 2 and 4, physical abnormalities begin to emerge and, in some cases, cognitive impairment may develop⁵
- Difficult to diagnose before irreversible damage occurs due to insidious onset and symptom overlap with common childhood complaints⁶

Silas, age 6

- Broad nose⁵
- Flared nostrils⁵
- Prominent supraorbital ridges⁵
- Thick lips⁵
- Large jowls⁵
- Large head circumference⁵

1. Neufeld EF, et al. The mucopolysaccharidoses. In: Scriver CR, Beaudet AL, Sly WS, et al, eds. The Metabolic and Molecular Bases of Inherited Disease. 8th ed. New York, NY: McGraw-Hill; 2001:3421-3452. 2. Mesolella M, et al. Acta Otorhinolaryngol Ital. 2013;33(4):267-272. 3. Wraith JE et al. Genet Med 2008;10(7):508-516. 4. Keilmann A et al. J Inherit Metab Dis 2012;35(2):343-353. 5. Martin R, et al. Pediatrics. 2008;121(2):e377-e386. 6. Burton B, et al. Eur J Pediatr. 2012;171(4):631-639. 7. Meikle PJ, et al. JAMA. 1999;281(3):249-254.

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