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.

EAR, NOSE, AND THROAT FEATURES

AGE (median age of onset)

1-2 years

74%
Otitis media[3]

PREVALENCE

2-3 years

68%
Enlarged adenoids or tonsils[3]

95%
Facial dysmorphism[3]

3-4 years

70%
Enlarged tongue[3]

50%
Tympanostomy[4]

47%
Adenoidectomy[4]

5 years

67%
Hearing loss[4]

10+

Common cause of death: complications from progressive airway obstruction[5]

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

Takeda

OVERVIEW OF **HUNTER SYNDROME**

Hunter syndrome is a progressive genetic disease

- X-linked recessive genetic disorder[1] affecting approximately 1 in 162,000 live births[7], almost exclusively males[1]
- 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 body[5]
- 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 develop[5]
- Difficult to diagnose before irreversible damage occurs due to insidious onset and symptom overlap with common childhood complaints[6]

Silas, age 6

- **Broad nose[5]**
- **Flared nostrils[5]**
- **Prominent supraorbital ridges[5]**
- **Thick lips[5]**
- **Large jowls[5]**
- **Large head circumference[5]**

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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