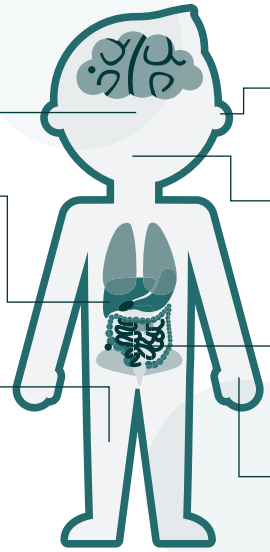
CONNECT THE DOTS: THINK HUNTER SYNDROME

As a pediatrician, 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] Many of the signs and symptoms of Hunter syndrome overlap with common childhood complaints, but clinical suspicion can be triggered by particular symptom clusters that are unlikely to appear in an unaffected child.[2] By recognizing key clinical features, you can help identify and refer patients who may be at risk from this disease.

NOTABLE FEATURES FOR PEDIATRICIANS



- **COARSE FACIAL FEATURES**[3]
- **ENLARGED LIVER AND SPLEEN**[3]
- **SKELETAL AND JOINT PROBLEMS (DYSOSTOSIS MULTIPLEX),** INCLUDING PROGRESSIVE JOINT STIFFNESS, CONTRACTURES, RESTRICTED MOBILITY AND RANGE OF MOTION, ATLANTOAXIAL INSTABILITY, ABNORMALLY SHAPED VERTEBRAE AND RIBS, HIP DYSPLASIA, GENU VALGUM, AND COXA VALGA[3-5]
- **RECURRENT OTITIS MEDIA,** OFTEN REQUIRING TYMPANOSTOMY[3,6]
- **ENLARGED TONGUE, TONSILS, AND ADENOIDS,** OFTEN REQUIRING TONSILLECTOMY AND ADENOIDECTOMY[6,7]
- **UMBILICAL AND INGUINAL HERNIAS** – REPEATED HERNIA REPAIRS ARE COMMON[3,6]
- **CARPAL TUNNEL SYNDROME** – REPEATED SURGICAL INTERVENTION IS COMMON[3,6]


Silas, age 6 months


Silas, age 2


Silas, age 4.5

Silas, age 6

- Broad nose[3]
- Flared nostrils[3]
- Prominent supraorbital ridges[3]
- Thick lips[3]
- Large jowls[3]
- Large head circumference[3]

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

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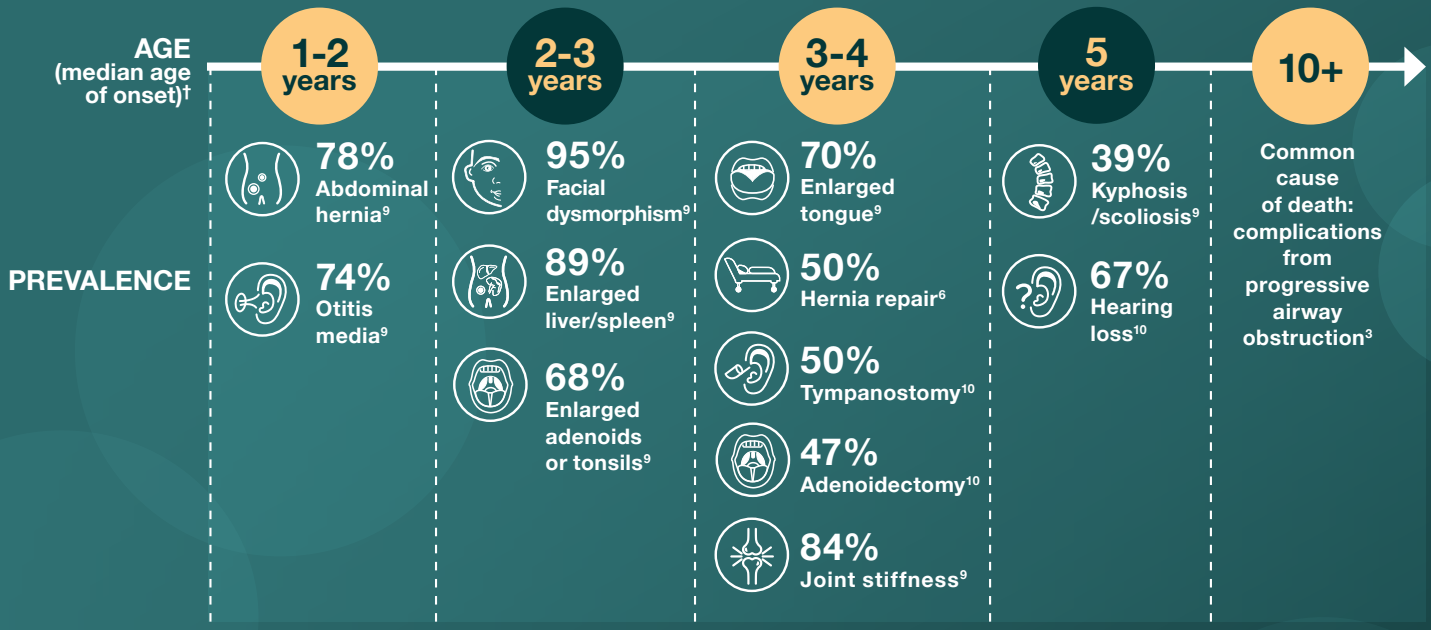
OVERVIEW OF **HUNTER SYNDROME**

Hunter syndrome is a progressive genetic disease

- X-linked recessive genetic disorder[1] affecting approximately 1 in 162,000 live births[8], almost exclusively males[1]
- Lysosomal storage disorder caused by the deficiency or absence of iduronate-2-sulfatase (I2S).[1,3] The widespread pathological lysosomal storage of glycosaminoglycans (GAGs) leads to progressive damage and dysfunction in cells, tissues, and organs throughout the body[2,3]
- 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[3]
- Difficult to diagnose before irreversible damage occurs due to insidious onset and symptom overlap with common childhood complaints[2]

PREVALENCE OF CLINICAL FEATURES IN HUNTER SYNDROME



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

- **Respiratory problems, including noisy breathing and snoring**
- **Chronic rhinorrhea**
- **Recurrent watery diarrhea**
- **Developmental delay and/or speech delay***

*Neuronopathic type only

Hunter syndrome is highly heterogenous. Symptoms vary in type, severity, age of onset, and progression.[3]

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. Burton B, *et al. Eur J Pediatr*. 2012;171(4):631-639. 3. Martin R, *et al. Pediatrics*. 2008;121(2):e377-e386. 4. Morishita K, *et al. Rheumatology* (Oxford). 2011;50(suppl 5):v19-v25. 5. White KK. *Rheumatology* (Oxford). 2011;50(suppl 5):v26-v33. 6. Mendelsohn NJ, *et al. Genet Med*. 2010;12(12):816-822. 7. Muenzer JM, *et al. Pediatrics*. 2009;124(6):e1-e12. 8. Meikle PJ, *et al. JAMA*. 1999;281(3):249-254. 9. Wraith JE, *et al. Genet Med*. 2008;10(7):508-516. 10. Keilmann A *et al. J Inherit Metab Dis 2012*; 35(2): 343 -353.

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