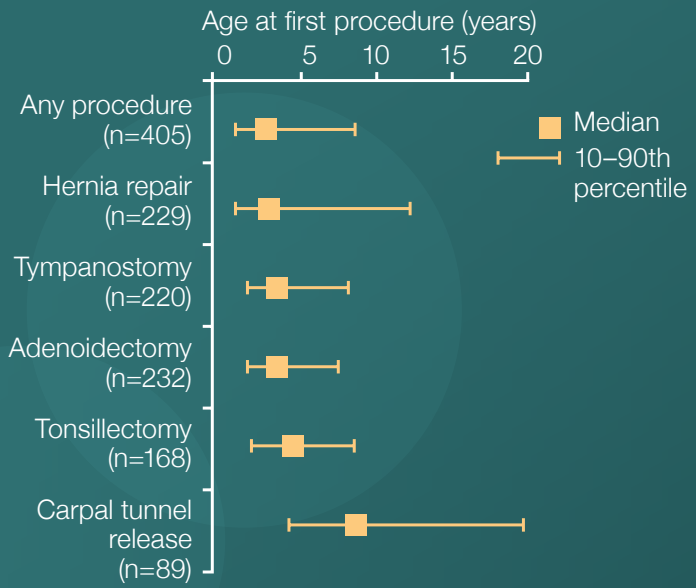
CONNECT THE DOTS: **THINK HUNTER SYNDROME**

As a pediatric surgeon, 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] A history of early and frequent surgeries is a defining feature of Hunter syndrome.[2] By recognizing key clinical features, you can help identify and refer patients who may be at risk from this disease.

Figure 1. Age at first surgical procedure[3]



Adapted from Mendelsohn NJ, *et al. Genet Med.* 2010;12(12):816-822

Silas, age 6

- Broad nose[4]
- Flared nostrils[4]
- Prominent supraorbital ridges[4]
- Thick lips[4]
- Large jowls[4]
- Large head circumference[4]

ACT EARLY

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

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

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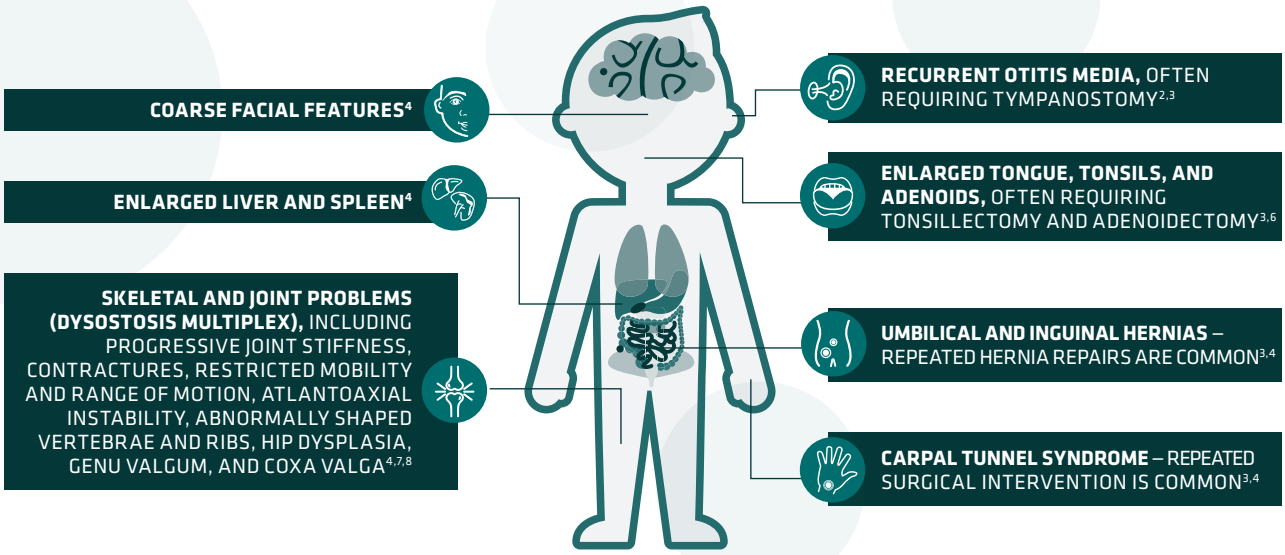
OVERVIEW OF **HUNTER SYNDROME**

Hunter syndrome is a progressive genetic disease

- X-linked recessive genetic disorder[1] affecting approximately 1 in 162,000 live births[5], almost exclusively males[1]
- Lysosomal storage disorder caused by the deficiency or absence of iduronate-2-sulfatase (I2S).[1,4] The widespread pathological lysosomal storage of glycosaminoglycans (GAGs) leads to progressive damage and dysfunction in cells, tissues, and organs throughout the body[2,4]
- 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[4]
- Difficult to diagnose before irreversible damage occurs due to insidious onset and symptom overlap with common childhood complaints[2]

NOTABLE FEATURES FOR PEDIATRICIANS



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. Mendelsohn NJ, *et al. Genet Med*. 2010;12(12):816-822. 4. Martin R, *et al. Pediatrics*. 2008;121(2):e377-e386. 5. Meikle PJ, *et al. JAMA*. 1999;281(3):249-254. 6. Muenzer JM, *et al. Pediatrics*. 2009;124(6):e1-e12. 7. Morishita K, *et al. Rheumatology* (Oxford). 2011;50(suppl 5):v19-v25. 8. White KK. *Rheumatology* (Oxford). 2011;50(suppl 5):v26-v33.

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