ARE YOU MISSING HUNTER SYNDROME?

ACT EARLY*

HUNTER SYNDROME IS A PROGRESSIVE GENETIC DISEASE

*Consider the importance of early assessment, diagnosis and follow-up by a specialist

A **rare combination** of common childhood complaints could indicate **Hunter syndrome (MPS II)**

Age 1-2 years

78% of HOS patients developed an **abdominal hernia**[1]

72% of HOS patients had **otitis media**[2]

Age 2-3 years

68% of HOS patients had enlarged **tonsils** or **adenoids**[1]

95% of HOS patients were described as having **facial dysmorphism**[1]

REFER TO A SPECIALIST TODAY

For more information visit:

Huntersyndrome.info

MPS II = mucopolysaccharidosis type II HOS = Hunter Outcome Survey

Median age of onset and prevalence data from HOS (Hunter Outcome Survey). 1. Wraith JE **et al**. Genet Med 2008; 10(7): 508–516. 2. Keilmann A **et al**. J Inherit Metab Dis 2012; 35(2): 343–353.

Hunter

Birth and first symptoms

“Hunter was my first child and he was just everything a mother could want from her first baby. But he had an umbilical hernia, ear infections and a runny nose from when he was just a few months old that never went away. And he had a big head. He didn’t start walking until he was 13 months old because his head was so big.

“I became pregnant with Kingston when Hunter was 6 months old. We were overjoyed with another little boy, but they were both chronically ill all the time, with ear infections or a runny nose.

“Kingston was more sick than Hunter. He was always on breathing treatments, which we attributed to asthma and breathing problems from his birth.”

“They were both chronically ill all the time, with ear infections or a runny nose.”

Hunter syndrome diagnosis

“We found a really good doctor who wanted to test for developmental delays, so we were going to start there. Then she said she had a presumption based on the way Kingston looked, and sent us to a geneticist.

“They really didn’t tell us anything other than that it was an enzyme disorder. Then we got the phone call, stating that they both had mucopolysaccharidosis II (MPS II). It was a hard time: lots of tears, lots of denial. Your children are perfect to you, and it’s like your dreams are crushed.

“Looking up MPS II, I found other children on the web that looked like my kids. Seeing these other kids, that aren’t diagnosed until 4 or 5, made me very thankful that the doctor caught it just by looking at the very strong, pronounced features in my 1-year-old at that time. I was very grateful that we caught it early, because it could have been way worse.”

Hunter

“I was very grateful that we caught it early, because it could have been way worse.”

The multisystemic nature of Hunter syndrome

Central nervous system

- Developmental delays [1]
- Mental impairment [1,2]
- Behaviour problems [1,2]
- Seizures [1,2]
- Hydrocephalus* [1,2]
- Spinal cord compression and cervical myelopathy resulting from atlantoaxial instability or stenosis* [1,3]

(*not primary CNS involvement) [4]

Respiratory

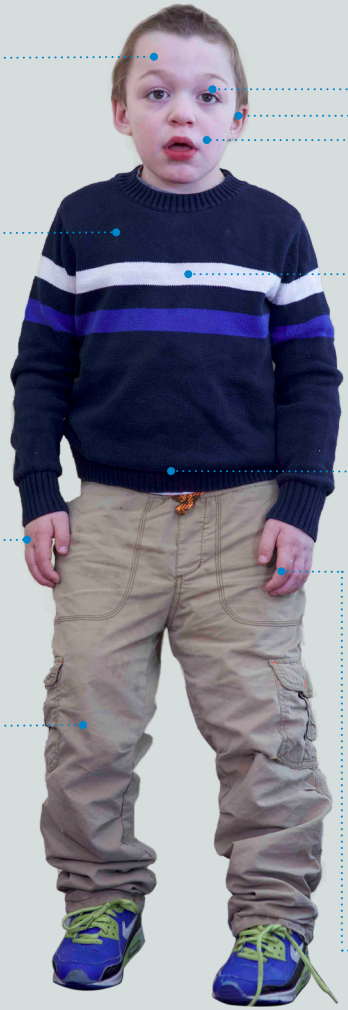
- Upper airway obstruction [1]
- Obstructive sleep apnoea [1]
- Frequent infections [1]
- Restrictive airway disease [1]

Peripheral nervous system

- Peripheral nerve entrapment (e.g. carpal tunnel syndrome) [1]

Skeletal

- Dysostosis multiplex [1]
- Joint stiffness and contractures [2]
- Claw-like hands [1]
- Short stature [3]
- Spine deformities (e.g. kyphosis, scoliosis) [3]



Eyes

- Vision loss [1,3]
- Optic disc swelling (e.g. papilloedema) [1,3]
- Retinal pigmentary degeneration [1]

Auditory

- Hearing loss [1,2,4]
  - Conductive
  - Sensorineural

Appearance

- Enlarged head [1,2]
- Distinct facial features (e.g. broad nose, flared nostrils, prominent forehead, thick lips) [1,2]
- Large tongue [1]

Cardiac

- Heart murmur [4]
- Cardiac valve disease [2]
- Cardiomyopathy [2]

Gastrointestinal

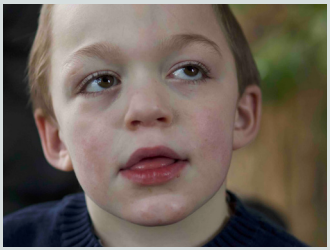
- Hepatosplenomegaly [2]
- Umbilical and inguinal hernia [2]
- Swallowing problems [4]
- Diarrhoea [1]

Skin

- Thickened, inelastic skin [1]
- Pebbled skin lesions [1]

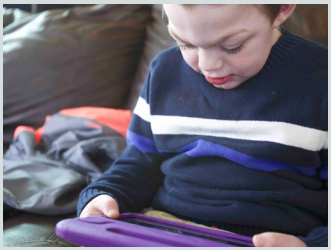
1. Martin R **et al.** Pediatrics 2008; 121(2): e377-e386.
2. Wraith JE **et al.** Eur J Pediatr 2008; 167(3): 267-277.
3. Scarpa M **et al.** Orphanet J Rare Dis 2011; 6(1): 72-90.
4. Holt J **et al.** J Pediatr 2011; 159(2): 320-326.

Visible physical signs



LARGE HEAD AND DISTINCT FACIAL FEATURES[1–4]

- Thick lips[1,3]
- Broad nose and flared nostrils[3]
- Excess facial hair[2]
- Prominent supraorbital ridges[1,3]
- Large jowls[1–3]



DEVELOPMENTAL DELAY[1–4]

Speech delay is seen in patients with CNS involvement[1–3]

SHORT STATURE*, SHORT TRUNK & NECK[3]

JOINT STIFFNESS AND CONTRACTURES[1–4]

Reduced range of movement[1]

CLAW-LIKE HANDS AND CARPAL TUNNEL SYNDROME[1–4]

Can result in loss of function[1]

1. Martin R et al. Pediatrics 2008; 121(2): e377-e386.
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Current disease severity and life impact

“My boys have severe MPS II, and they’re cognitively affected by the disease. Hunter has more storage in his face, and in his hands. He doesn’t understand things like a normal child would. He is more aggressive, a very big talker, and a huge social butterfly. Kingston doesn’t talk very often. He’s 6 years old now and only has about 60 words. He doesn’t understand things: he’s more of an eye contact boy. He has some of the MPS II facial features, and heart problems.

“They’re completely dependent on me. They’re my reason for living: it’s taking care of them, because they can’t do any of that themselves, playing with them, showing them attention until it’s bedtime; and we do it all over again the next day.”

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