INFORMATION for ENT Specialists

This leaflet outlines:

- **the common symptoms and clinical consequences of MPS II**
- **current opinion from the literature on how ear, nose and throat complications are managed in MPS II patients**

This leaflet is a summary of the published information as of February 2018, is not exhaustive and does not constitute any recommendations. It is the responsibility of the ENT specialist to determine the optimal management plan for the individual patient.

Visit **Huntersyndrome.info** to learn more

The information in this leaflet is intended for healthcare professionals only.

This information is intended for an international audience outside of the USA and has been produced by Takeda. The leaflet has been developed in accordance with industry and legal standards to provide information for healthcare professionals about Hunter syndrome health topics. Takeda makes every reasonable effort to include accurate and current information. However, the information provided in this leaflet is not exhaustive.www

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INFORMATION for ENT Specialists

MPS II

A combination of common childhood complaints in your patient could indicate Hunter syndrome (MPS II), a rare genetic disorder that predominantly affects males. MPS II is caused by a mutation that stops the lysosomal enzyme iduronate-2-sulfatase (I2S) from being produced. I2S normally breaks down molecules called glycosaminoglycans (GAGs); without I2S, GAGs accumulate, causing progressive, multisystemic disease, leading to early mortality.[1]

Early symptoms that should raise suspicion of MPS II include otitis media, abdominal hernia, nasal obstructions, and enlarged tonsils/adenoids and tongue.[2] Although these symptoms are common among children, in MPS II patients these symptoms occur early and in combination, and may be refractory to treatment.[3] It is therefore favourable to be able to diagnose MPS II in patients early, so that appropriate management may be initiated as soon as possible.[4]

ACT EARLY

Check the **abdomen** for **hernia**.

+

Check the ear, nose and throat for **otitis media** and **enlarged tonsils** or **adenoids**.

+

Check the rest of the body for **joint stiffness** and **prominent facial features**.

If you suspect Hunter syndrome: REFER TO A SPECIALIST TODAY

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Presentation

The course and presentation of disease for each MPS II patient is unique, but there are common features that are prevalent, due to the accumulation of GAGs in soft tissues. These features are noticeable from an early age, and include a large head circumference, a broad nose and flared nostrils, prominent brow, large jowl, thick lips, and an enlarged, protruding tongue.[2]

Common clinical symptoms of MPS II include frequent hearing loss due to otitis media or other conductive impairment or sensorineural degeneration, and breathing and chewing or swallowing difficulties due to respiratory obstructions (enlarged tonsils and adenoids, nasal obstructions), skeletal deformities and abdominal distension. Therefore, ENT specialists are amongst the first physicians to encounter MPS II symptoms when seeing patients.[2,4]

Clinical consequences

Tympanostomy, adenoidectomy and tonsillectomy are among the most common surgical procedures performed to relieve symptoms in MPS II patients (these are often performed before the diagnosis of MPS II is even made). For example, fewer than 10% of children in the general population receive tympanostomies, whereas over 51% of MPS II patients have this procedure. Hearing loss is a concern in MPS II patients, as it can cause behavioural and learning difficulties (in addition to any underlying cognitive impairment).[3]

Upper airway obstructions can cause obstructive sleep apnoea in patients, causing fatigue and also contributing to behavioural problems.[4] These obstructions are also a major contributor to mortality in MPS II patients; 39% of deaths in MPS II patients are due to respiratory/airway involvement, so it is important to carefully monitor the development of airway obstructions.[5]

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The following summary describes the current opinion in the literature (as of February 2018), however, it is the responsibility of the ENT specialist and the multidisciplinary team to determine the optimal management plan for the individual patient.

Management

Otologic and audiologic evaluations performed every 6 to 12 months can help to assess the hearing loss that contributes to behavioural problems and learning difficulties in MPS II patients. Myringotomy with placement of ventilating tubes and hearing aids may improve hearing.[4]

A diagnosis of the airway obstructions in a patient requires a comprehensive evaluation of the medical history of the patient, a current physical examination, and imaging procedures. Pulmonary function testing using spirometry can be employed, although it is difficult to perform on young or cognitively impaired patients, as it requires patient cooperation. An overnight sleep study, conducted at a hospital or at home, can evaluate the severity of obstructive sleep apnoea.[2]

For a more detailed assessment, a bronchoscopy can be used to evaluate respiratory involvement. Assessment can be performed with a rigid bronchoscope, which provides high-quality images but distorts the anatomy, and a flexible bronchoscope, which fits the anatomy and therefore visualises the dynamics of the obstruction.[4]

The first step in managing airway involvement is the removal of the obstructions, including the tonsils and adenoids. Preparation before surgery is a key aspect of management in MPS II and patients should be assessed by a multidisciplinary team that includes a cardiologist, an ENT specialist, and an anaesthetist before any surgical procedure. However, due to the progressive nature of airway involvement, surgery may only provide a temporary solution; continuous positive airway pressure, ventilation, or tracheotomy may be required at a later stage.[2,4]

Summary

In MPS II patients, ENT specialists will observe hearing loss (due to otitis media or other conductive impairment or sensorineural degeneration), and breathing and chewing or swallowing difficulties due to respiratory obstructions (enlarged tonsils and adenoids, nasal obstructions), skeletal deformities and abdominal distension.[2,4]

Hearing loss can be monitored with otologic and audiologic evaluations performed every 6 to 12 months, while a comprehensive evaluation of the medical history of the patient, a current physical examination, and imaging procedures are needed to diagnose airway obstructions.[2,4] Managing airway problems in MPS II patients often involves surgery, but preparation is crucial and ENT specialists should work with cardiologists and anaesthetists in evaluating the patient.[2]

References:

1. Burton BK, Giugliani R. Diagnosing Hunter syndrome in pediatric practice: practical considerations and common pitfalls. Eur J Pediatr 2012; 171(4): 631–639.
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5. Burton BK *et al.* Survival in idursulfase-treated and untreated patients with mucopolysaccharidosis type II: data from the Hunter Outcome Survey (HOS). J Inherit Metab Dis 2017; 40(6): 867–874.

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