The Ear, Nose and Throat in MPS

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Preciptorship program on MPS

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Alterations of the outer and middle ear in MPS I

- narrowing of the external auditory canals, chronic otitis externa
- middle ear effusion, chronic inflammation of the middle ear
- deformation of the ossicles, especially the stapes
- thickened mucosa and granulation in the middle ear
Therapeutic Options

- adenoidectomy
- ventilation tubes

Optimizing and stabilizing of the hearing ability
Optimizing the ventilation of the middle ear

• does not automatically normalize hearing ability
• conductive component may persist after myringotomy
• clinicians should not delay considering hearing aids for MPS children
Inner Ear in MPS

- hearing loss seems to increase with age
- high variability

- degeneration of the organ of Corti
- Reissner’s membrane adherent to tectorial membrane
- lack of neurons, Hurler cells in the spiral ganglion
- distended congested vessels in the stria vascularis
Audiogram of a 12 yrs old patient with MPSI
Mainz data hearing loss:

nach WHO: 0.5/1/2/4 kHz
Many patients need hearing aids:
Fitting of hearing aids in permanent hearing losses

- conductive losses caused by deformation of the ossicles, especially the stapes, thickened mucosa and granulation in the middle ear
- Sensorineural losses
- only possible if the hearing loss is not too much fluctuating
- impedes the ventilation of the outer ear channel and increases the probability of inflammations
Mouth

• Thickened lips
• Gingival hyperplasia, gapped teeth
• Macroglossia
• Tonsillar hypertrophy
• Sore throat
  – swelling of the mucosa,
  – mucus
Incisor gap (in mm); n = 87

<table>
<thead>
<tr>
<th>MPS subgroup</th>
<th>Mean (mm)</th>
<th>Range (mm)</th>
</tr>
</thead>
<tbody>
<tr>
<td>MPS I</td>
<td>33</td>
<td>25-47</td>
</tr>
<tr>
<td>MPS II</td>
<td>39</td>
<td>22-60</td>
</tr>
<tr>
<td>MPS IV</td>
<td>39</td>
<td>36-40</td>
</tr>
<tr>
<td>MPS VI</td>
<td>36</td>
<td>22-47</td>
</tr>
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</table>

MPS: mucopolysaccharidoses
Incisor gap

Subgroups of MPS

- MPS I: n = 18, MW = 3.3
- MPS II: n = 38, MW = 3.8
- MPS IV: n = 5, MW = 3.9
- MPS VI: n = 26, MW = 3.6

n = 87
Mallampati classification

<table>
<thead>
<tr>
<th>Class</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>I</td>
<td>Soft palate, uvula and palatal arch are completely visible</td>
</tr>
<tr>
<td>II</td>
<td>Palatoglossal arch is partially visible, tip of the uvula is not visible</td>
</tr>
<tr>
<td>III</td>
<td>Only soft palate visible</td>
</tr>
<tr>
<td>IV</td>
<td>Only hard palate visible</td>
</tr>
</tbody>
</table>

Mallampati classification

<table>
<thead>
<tr>
<th></th>
<th>Klasse 1</th>
<th>Klasse 2</th>
<th>Klasse 3</th>
<th>Klasse 4</th>
<th>ALL</th>
</tr>
</thead>
<tbody>
<tr>
<td>MPS I</td>
<td>3</td>
<td>2</td>
<td>6</td>
<td>6</td>
<td>17</td>
</tr>
<tr>
<td>MPS II</td>
<td>--</td>
<td>6</td>
<td>11</td>
<td>23</td>
<td>40</td>
</tr>
<tr>
<td>MPS IV</td>
<td>7</td>
<td>2</td>
<td>1</td>
<td>--</td>
<td>10</td>
</tr>
<tr>
<td>MPS VI</td>
<td>2</td>
<td>15</td>
<td>6</td>
<td>8</td>
<td>31</td>
</tr>
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</table>
The progressive accumulation of partially degraded glycosaminoglycans (GAGs) within the cells of various body tissues leads to cellular dysfunction:

- Facial dysmorphism
- Widened nasal bridge, large nares
Nose

- Restricted nasal airflow
- Tonsillar hypertrophy
- Recurrent nose infections
  ("running nose")
Sleep apnea

- Significant upper airway obstruction
- Hypertrophy of the adenoids and tonsils
- Thickening of the mucosa
- Alteration of the base of the tongue
  - Hypopharynx
  - Larynx
Therapeutic options

- Enzyme replacement therapy (ERT)
- Increasing the input of fluids
- Antibiotic treatment in case of bacterial infection
- Steroids in case of sudden worsening
- Adenoidectomy, inferior turbinate reduction
- Tonsillectomy
- Continuous positive airway pressure (CPAP)
# Surgery in patients with MPS II

<table>
<thead>
<tr>
<th>Type of surgery</th>
<th>Percentage (%)</th>
<th>Median age (years)</th>
<th>10(^{th})-90(^{th}) percentile (years)</th>
</tr>
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<tr>
<td>Adenoidectomy</td>
<td>47.4</td>
<td>3.5</td>
<td>1.6-7.8</td>
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<tr>
<td>Tonsillectomy</td>
<td>36.6</td>
<td>4.3</td>
<td>2.0-8.3</td>
</tr>
<tr>
<td>Tracheotomy</td>
<td>4.2</td>
<td>11.9</td>
<td>2.1-24.8</td>
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Adenoids of patients with mucopolysaccharidoses demonstrate typical alterations

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ENT surgery in patients with MPS

• is often very helpful in children with narrow upper airways
• should be done only in specialized centres
Alterations of pharynx and larynx

• classification protocol in which the alterations of
  – mucosa in the posterior region of the larynx
  – the false
  – the true vocal folds and the
  – the epiglottis,
  were documented

• flexible or rigid endoscopy

### A: Posterior region of the larynx

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<tr>
<td>1</td>
<td>Minimally extended volume of the mucosa</td>
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<td>2</td>
<td>Markedly extended volume of the mucosa without obstruction of the airways</td>
</tr>
<tr>
<td>3</td>
<td>Markedly extended volume of the mucosa with obstruction of the airways</td>
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<tr>
<td>4</td>
<td>Mucosa is moved into and out of the larynx during inspiration and expiration</td>
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<tr>
<td><strong>4</strong></td>
<td>Mucosa of the region of the false vocal folds is moved up and down during inspiration and expiration</td>
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</table>
T: Laryngeal vestibule – false vocal folds
Before ERT

6 months under ERT
Ask an ENT specialist or pediatric audiologist to follow your MPS team!

• treatment of the inflammations in the upper respiratory tract
  – symptomatic treatment
  – adenoidectomy, tonsillectomy
  – insertion of ventilation tubes
• fitting of hearing aids
• documentation of the symptoms
Thank you for your kind attention!