The impact of OI on jaw function and oral health-related quality of life

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Centralization of specialized oral health care service for people with rare inherited diseases
Dental symptoms in adults with OI

• Dentinogenesis imperfecta (DI)
  – Discoloration of teeth
  – Pulpal obliteration
  – Deviations in dental morphology

• DI in 25 % of patients with OI
  – Type 3-4: 94 % with DI
  – Type 1: 2 % with DI

• Enamel and crown fractures

• Thuesen et al 2018
Craniofacial morphology and occlusion in patients with OI

- Relative mandibular prognatism and mandibular overjet
  - *Stenvik et al 1985*
  - *Waltimo Siren et al 2005*
- Most severe craniofacial abnormalities in OI type 3 and 4
- Near normal jaw morphology in OI type 1
  - *Jensen & Lund 1997*
- Mandibular overjet & posterior crosbite prevalent in OI type 3 and 4
  - *Bendixen et al. 2018*
Necessities for the ability to chew, eat, speak, and smile: Structures of the orofacial complex

- Jaw & teeth
- TM joint & discus
- Muscles

The presence of dental abnormalities like DI and skeletal abnormalities like OI, as well as the presence of malocclusion, might have impact on jaw function and oral health-related quality of life.
Bad jaw function = Temporomandibular disorder  TMD

• TMD are a heterogeneous group of pathologies affecting the temporomandibular joint (TMJ), the jaw muscles, or both.

• They are characterized by a classically described triad of clinical signs:
  – muscle and/or TMJ pain;
  – TMJ sounds;
  – and restriction, deviation, or deflection of the mouth opening path
Aims of study on jaw function

• to assess the occurrence of temporomandibular disorders (TMD)
  – in adults with osteogenesis imperfecta and

• to compare the occurrence of TMD
  – according to severity of OI

• Research Diagnostic Criteria for assessment of temporomandibular disorders RDC/TMD
Study population: 75 adults with genetically verified OI

<table>
<thead>
<tr>
<th>OI mild (type 1)</th>
<th>Females</th>
<th>Males</th>
</tr>
</thead>
<tbody>
<tr>
<td>Numbers</td>
<td>31</td>
<td>25</td>
</tr>
<tr>
<td>Age (yr) mean</td>
<td>49 (14.5)</td>
<td>49 (17.4)</td>
</tr>
<tr>
<td>Age (yr) range</td>
<td>23-72</td>
<td>22-80</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>OI severe (type 3/4)</th>
<th>Females</th>
<th>Males</th>
</tr>
</thead>
<tbody>
<tr>
<td>Numbers</td>
<td>9</td>
<td>10</td>
</tr>
<tr>
<td>Age (yr) mean</td>
<td>49 (12.3)</td>
<td>44 (10.3)</td>
</tr>
<tr>
<td>Age (yr) range</td>
<td>37-72</td>
<td>28-63</td>
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</table>
Diagnoses on myofacial pain in
56 with OI I – 19 with OI III/IV – 100 general – 100 TMD

RDC/TMD I - myofacial pain

<table>
<thead>
<tr>
<th></th>
<th>OI I</th>
<th>OI III/IV</th>
<th>General</th>
<th>TMD</th>
<th>100%</th>
<th>80%</th>
<th>60%</th>
<th>40%</th>
<th>20%</th>
<th>0%</th>
</tr>
</thead>
<tbody>
<tr>
<td>Myofacial pain (1)</td>
<td>51</td>
<td>18</td>
<td>90</td>
<td>55</td>
<td>ns</td>
<td>55</td>
<td>45</td>
<td>10</td>
<td>1</td>
<td>5</td>
</tr>
<tr>
<td>No pain</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
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</tbody>
</table>

ns: not significant
Chronic Pain Grade (CPG 0-4) in 56 with OI I - 19 with OI III/IV

- **CPG 0**: no TMD pain in the prior six months
- **CPG 1**: low disability and low pain intensity
- **CPG 2**: low disability and high pain intensity
- **CPG 3**: high disability and moderately limiting
- **CPG 4**: high disability and severely limiting
Conclusions on OI and jaw function
Bendixen et al 2018

• Irrespective of OI severity, very few patients had TMD diagnoses on myofacial pain, disc displacement, or joint disorder

• The bodily pain complaints frequently occurring in OI was not reflected in the orofacial area
Necessities for the ability to chew, eat, speak, and smile:
Structures of the orofacial complex

- Jaw & teeth
- TM joint & disc
- Muscles

The presence of dental abnormalities like DI and skeletal abnormalities like OI, as well as the presence of malocclusion, might have impact on jaw function and oral health-related quality of life.
Aim of study on **oral health-related** quality of life in patients with rare skeletal disorders

- To assess the impact upon oral health-related quality of life in two rare diseases, characterized by the presence of both skeletal and dental symptoms (XLH and OI)
- To compare the impact on oral health related quality of life according to the disease, XLH or OI
Two cross-sectional studies on and OI respective XLH in Denmark

X-linked Hypophosphatemia
- *PHEX* mutation, X-linked
- Renal depletion of phosphate and hypophosphatemia
- Hypomineralization and softening of bones
- Short stature and bowed legs
- Enlargement of growth plates
- Costochondral thickening
  - “rachitic rosary”
- Pulpitis and pulp necrosis
  - in teeth without decay
- Dental abscesses
- Recruitment by MD PhD Signe S Beck-Nielsen

Osteogenesis Imperfecta
- Interdisciplinary assessment
  - Clinical, radiological, histological, genetical, QoL, dexas, otomicroscopical, dental etc

Recruitment by MD PhD Jannie D Hald
Study populations XLH and OI

<table>
<thead>
<tr>
<th></th>
<th>Females</th>
<th>Males</th>
</tr>
</thead>
<tbody>
<tr>
<td>XLH</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Numbers</td>
<td>24</td>
<td>11</td>
</tr>
<tr>
<td>Age (yr) mean (SD)</td>
<td>41 (14.6)</td>
<td>42.5 (19.6)</td>
</tr>
<tr>
<td>Age (yr) range</td>
<td>21-75</td>
<td>19-73</td>
</tr>
<tr>
<td>OI</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Numbers</td>
<td>40</td>
<td>35</td>
</tr>
<tr>
<td>Age (yr) mean (SD)</td>
<td>49 (13.9)</td>
<td>48 (15.7)</td>
</tr>
<tr>
<td>Age (yr) range</td>
<td>23-73</td>
<td>22-80</td>
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</table>
Method: quantifying the impact of oral health on quality of life

Tool: Oral Health Impact Profile OHIP-D


**Standardized questionnaire**
- 49 questions
- Frequency of symptoms or experiences
- Score 0 – 4
  - 0 = never
  - 4 = daily / always

**7 domaines**
- Functional limitations
- Physical pain
- Psychological discomfort
- Physical disability
- Psychological disability
- Social disability
- Handicap
OHIP profiles by means (t-test) in OI severe, OI mild, XLH and Reference

![OHIP profiles graph](image-url)
Comparison (Wilcoxon rank-sum test)
OI patients (N=75) and XLH patients (N=35)
Comparison (Wilcoxon rank-sum test)
Mild OI (N=56) and severe OI (N=19)
Conclusions on oral health related quality of life

• In both XLH and OI groups, the domaine scores in all domaines - except one (social disability) - deviate from reference group
• The impact of XLH exceeds the impact of OI in nearly all domaines
  – Functional limitations
  – Pain
  – Psychological discomfort
  – Psychological disability
  – Handicap
• According to OHIP, the impact of severe OI hardly exceeds the impact of mild OI
Thank you for your attention

Thank you for support from

- participating patients
- The Danish Society for Osteogenesis imperfecta
- Care for Brittle Bones (C4BB)

Thank you for collaboration with

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  - Aarhus University Hospital
    - Dept of medicin and endocrinology
    - Center for Rare Diseases
  - Department of Dentistry and Oral Health, Aarhus University
- the staff at
  - Dept. Maxillofacial Surgery, Aarhus University Hospital
  - Department of Dentistry and Oral Health, Aarhus University

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