**Pediatric Patient-Reported Outcomes Assessment: A Case Study in Epidermolysis Bullosa**

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**BACKGROUND**

Pediatric Patient-Reported Outcomes Assessment

- Identifying the age or developmental stage at which children can reliably and reproducibly report that self-care is challenging.
- Children as young as 3 years of age have reported reliable assessment of outcome concepts such as pain. However, research supports a more conservative estimate of 8 to 10 years of age for children reporting an adequate level of self-care.

**Horn and Tidman,** 2002

- No established guidelines exist for age cutoffs for self-reporting. The Critical Task Forum (C-Task) recommends that specific age boundaries should be determined, in part by how abstract or concrete the reported concept is and further suggests the following cutoffs:
  - Age 3 years: often cited as the lower bound of the age range for self-report.
  - In patients aged 7 to 11 years, mixed validity and reliability results have been observed, such that a combination of self- and parent-reporting may be best.
  - In patients aged 15 years and older, psychometric testing has demonstrated that self-reporting is generally acceptable.

- Consideration of age may be increasingly important; intemperance of variability is comprehensivness and willingness/interest to respond should also be assessed.

**Epidermolysis Bullosa—A Rare Disease**

- Epidermolysis bullosa (EB) is a family of genetic skin fragility disorders, clinically characterized by blistering of the skin in response to friction or trauma.

- EB is extremely rare, with an estimated prevalence (all subtypes) of 0.10 per 100,000 population in the United States. 1 out of every 500,000 births is affected by EB.

- There is no cure for EB. The current standard of care for EB is support; palliative wound care consists of changing dressings and monitoring the wound site for excessive exudate and/or infection.

- No Related Quality of Life (EQ) measures in EB.

- Published literature highlights the following important health-related quality of life (HRQOL) aspects that are specific to patients with EB:
  - Limitations in physical functioning, including pain or discomfort on ambulation, bathing, grooming, dressing, and getting in and out of the house, sleeping, playing sports.
  - Emotional, social, and psychological effects, including anxiety, depression, thinking, relationships with friends and family.

- Aspects specific to wound treatment (e.g., pain during wound dressing change)

**OBJECTIVES**

- To identify and evaluate HRQOL measures used with a pediatric population (aged 3 to 18 years) with EB.

**METHODS**

- A structured PubMed search was conducted using Medical Subject Headings (MeSH) terms.

- Of the 136 abstracts identified, 40 were appropriate for further evaluation; 33 articles underwent full-text review.

- The following measurement properties were evaluated for each HRQOL measure based on standard criteria:
  - Purpose
  - Availability of age-appropriate versions
  - Number of respondents (i.e., respondent-based)
  - Readability
  - Content validity
  - Age-appropriate versions for patients with EB
  - Age-relevance of concepts addressed
  - Functional properties
  - Validity (known-groups, construct)
  - Reliability (test-retest, internal consistency)

**RESULTS**

- This review identified 8 HRQOL measures implemented in studies with patients with EB.

**Table 1. HRQOL Measures Implemented in EB Studies**

<table>
<thead>
<tr>
<th>Measure</th>
<th>Domain</th>
<th>Psychometric Properties</th>
<th>Purpose</th>
<th>Availability of age-appropriate versions</th>
<th>Number of respondents (i.e., respondent-based)</th>
<th>Validity (known-groups, construct)</th>
<th>Reliability (test-retest, internal consistency)</th>
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</thead>
<tbody>
<tr>
<td>SF-36</td>
<td>Physical, Mental</td>
<td>Excellent (all age groups)</td>
<td>Overall</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
</tr>
<tr>
<td>Dermatology Life Quality Index (DLQI)</td>
<td>Physical, Emotional</td>
<td>Excellent (all age groups)</td>
<td>Overall</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
</tr>
<tr>
<td>Skindex-29</td>
<td>Physical</td>
<td>Excellent (all age groups)</td>
<td>Overall</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
</tr>
<tr>
<td>CDLQI</td>
<td>Physical, Emotional</td>
<td>Excellent (all age groups)</td>
<td>Overall</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
</tr>
<tr>
<td>DQOLS</td>
<td>Physical, Emotional</td>
<td>Excellent (all age groups)</td>
<td>Overall</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
</tr>
<tr>
<td>SF-12</td>
<td>Physical, Emotional</td>
<td>Excellent (all age groups)</td>
<td>Overall</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
</tr>
<tr>
<td>HRQOL</td>
<td>Physical, Emotional</td>
<td>Excellent (all age groups)</td>
<td>Overall</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
</tr>
</tbody>
</table>

**DISCUSSION**

- EB HRQOL measures were identified and reviewed. An expected, no single measureOverview covers the full experience of EB across the age span of pediatric patients.

- The DLQI, a generic measure of HRQOL, may potentially be used to compare the burden of EB in pediatric patients aged 10 years with the burden of other pediatric conditions.

- The CDLQI covers the greatest portion of the age range of interest (4-16 years); this measure also is available in two different child-friendly versions (for children and adults) to aid in patient recruitment and understanding.

- The CDLQI item-sets are adequate for children and emotional and functional HRQOL domains. Deeply affected children (adolescents and young adults)

- Not all GLOD is content is relevant to children and adolescents:
  - Some items may be too abstract for children to understand.
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- Potential floor effects for patients with more severe EB on items that they may not experience.

**LIMITATIONS**

- The impact of EB will vary by age, consistent with normal childhood development.

- Future research is needed to document and assess HRQOL concepts in pediatric patients with EB.

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**REFERENCES**


