

Quality of Life and Independence in Activities of Daily Living in Epidermolysis Bullosa

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Abstract

Background: Epidermolysis bullosa is a rare genetic skin disorder with four main types. One of the most debilitating subtypes is recessive dystrophic epidermolysis bullosa (RDEB). Patients experience frequent blister formation that occurs spontaneously or as a result of minor pressure or friction. This often leads to scarring and fibrosis of the skin, contracture development, pseudosyndactyly of the hands, and chronic pain. Functionally patients experience decreased independence in their activities of daily living and quality of life.

The relationship of engagement in activities of daily living and perceived quality of life has not been studied for this population. The aims of this cross-sectional study were to find evidence to support this relationship through the correlation of a patient-reported independence in activities of daily living questionnaire and a patient-reported quality of life measure developed for persons with EB.

Results: 19 subjects with RDEB aged 9-25 were recruited from a multidisciplinary EB center from 2014-2017. The results demonstrate a slight positive trend in the correlation of independence in activities of daily living and quality of life, though the data was not sufficient to be statistically significant.

Conclusion: The results of this study have implications for further research with recommendations to recruit a greater number of subjects. Use of a more sensitive and robust assessment of activities of daily living that also includes leisure skills may yield more statistically significant data. In addition, the findings confirm the important role that occupational therapy has in addressing patient needs and facilitating greater functional independence.

Background

Epidermolysis bullosa (EB) is a group of inherited diseases characterized by mechanical fragility of the skin and mucous membranes¹. Blister formation typically presents at birth and can occur spontaneously or following friction or mechanical trauma². EB causes such fragility of the skin that those with the diagnosis are referred to as “butterfly children” by DEBRA International. According to the Orphan Drug Act of 1983, EB qualifies as a rare disease as defined as a condition that affects fewer than 200,000 people worldwide, affecting both sexes. In the United States, its prevalence is reported to be 11 per 1 million live births³.

EB can be divided into four major types: EB simplex (EBS), junctional (JEB), dystrophic (DEB), and Kindler EB (KEB). Types are determined by the level of skin cleavage, inheritance pattern, and clinical and molecular features⁴. Considerable variation may exist in disease severity and progression even within an EB subtype because of the influence of environmental and/or modifying genetic factors⁵. Generally speaking, for those with EBS, mechanical fragility and blistering is confined to the epidermis. With JEB blister formation is within the lamina lucida of the skin basement membrane. KEB patients have blister formation in multiple levels within or beneath the basement membrane. Patients with DEB have blister

formation in the uppermost dermis. DEB can be further classified by their mode of inheritance into either dominant, DDEB, or recessive, RDEB. In general, RDEB patients tend to have more severe disease progression. Persons with RDEB present at birth with widespread blisters and erosions of the skin and mucous membranes. Multisystem involvement may develop including fusion of fingers (mitten hand deformity) and toes, flexion contractures affecting the limbs and neck, osteoporosis and esophageal stricture⁶. Patients also present with chronic pain, decreased independence in activities of daily living (ADL) such as toileting, feeding dressing, and writing⁷, and decreased quality of life (QOL).

Patient care is optimally provided by a multidisciplinary team (MDT) that may include a dermatologist, nurse specialist, dietician, pain management expert, and physical and occupational therapist (OT)⁸. OTs are concerned with enabling patient engagement with their environments including independence in ADLs. OT interventions include improving and maintaining range of motion, strength, and fine motor function, and providing modifications and adaptations.

For this study, the definition of QOL is an abstract, multidimensional construct reflecting physical, psychological and social aspects of an individual's well-being⁹. The disabling condition of EB significantly affects a patient's perceived QOL.

ADLs are the essential tasks that each person needs to perform on a regular basis for basic survival and wellbeing. It includes various self-care skills such as bathing and showering, toileting, dressing, and eating. It also includes activities that support daily life and are oriented toward interacting with the environment such as working, going to school, and engaging in leisure activities.

The OT for EB clinical practice guideline recommends that patients with all subtypes of EB with functional or biomechanical impairments including contractures and decreased mobility receive an early OT referral for assessment of their functional independence in ADL with frequent re-evaluation. And that persons with EB are provided with modifications that are needed to limit cutaneous injury while enabling natural motor development, independence, and social integration that affects QOL. In addition, it is recommended that OTs work to provide adaptations to optimize participation and success in work and school, which can lead to confidence and even more opportunities for leisure and social participation in those settings¹⁰.

The purpose of this study was to find a correlation between patient reported performance of ADLs and patient perceived QOL in persons living with RDEB. This is based on our hypothesis that persons with RDEB, one of the most severe forms of EB, that have greater independence with ADLs, also have a higher perceived QOL.

Results

Analysis was completed by a research statistician who was provided with the subjects' ages and sex and was blinded to all other identifiers.

All subjects (n=20; 100%) with RDEB from the EB clinic who met our criteria consented to participate in the study. One subject was removed from the study due to not complying with instructions in answering the questionnaires .

Of the 19 RDEB patients, 10 (53%) were male and the average age was 15.1 ± 3.7 years old (Table 1a). ADL scores were not significantly associated with total QOLEB scores ($r_s = 0.31, p = 0.196$), functional QOLEB scores ($r_s = 0.30, p = 0.209$), or emotional QOLEB scores ($r_s = 0.23, p = 0.349$) (Table 1b).

QOL and ADL were not significantly correlated though there was a slight positive trend as evidenced by the data. (figure 1a, b, and c).

Table 1a. Descriptive Statistics

Variable	Count	%
<i>Gender</i>		
Male	10	53%
Female	9	47%

Table 1b

Variable	N	Mean	SD
Age	19	15.1	3.7
ADL Scores	19	7	(6-8)*
QOLEB Scores	19	34.6	9
QOLEB: Functional	19	22.3	5.8
QOLEB: Emotional	19	12	4.3

*Median and interquartile range reported

Table 1c. Statistical Analysis

Variable	Correlation	p-value
ADL vs. QOLEB (total)	0.31	0.196
ADL vs. QOLEB (funtional)	0.30	0.209
ADL vs. QOLEB (emotional)	0.23	0.349

Discussion

The present study reports on 95% (n = 19) patients with RDEB from a major metropolitan hospital over a 3-year period. Our data nearly represented 53 – 47% males to females. The number of subjects that was recruited is indicative of the rarity of this condition.

Another cross-sectional study of QOL and hand function recruited patients with various types of EB from across the United States. Of the 71 participants recruited from 20 states, 32 had RDEB. The results indicated that QOL was shown to be highly related to the degree of hand function with a significant correlation of the Quality of Life and EB (QOLEB) questionnaire with parent reported responses using the ABILHAND-kids questionnaire¹¹. Their findings also indicated that children with RDEB reported the worst hand function and QOL.

Data from the National Epidermolysis Bullosa Registry from 1986–2002, United States, showed that out of a total of 3280 patients enrolled nationwide, 386 (12%) had RDEB. They further showed that the frequency of totally independent patients with RDEB range from 42–73% (n = 162.12 to 281.78) for grooming, dressing, and bathing, and 30% (n = 115.8) needed assistance with feeding and toileting⁷.

Limitations to this study:

- The number of subjects was limited inherent in such a rare condition. However, 100% consented to participate.
- Non-validated survey of ADL was used. The tool was developed by the primary researcher due to lack of experience in the use and reporting of tools.
- Recruitment process (main researcher was a clinical expert in EB), could also influence a bias on the participant responses due to possible fear of effecting their care.

Future Research:

In the future it is recommended to use a patient's self-reported occupational performance of self-care using a validated tool such as the Canadian Occupational Performance Measure (COPM)¹². This questionnaire includes self-reported performance in the areas of self-care, productivity, and leisure and also provides another score for level of satisfaction. Other validated assessments that could potentially be used include the Child Occupational Self-Assessment (COSA) that is a self-report of occupational competence and values for everyday activities that uses visual cues for rating. This assessment is intended for ages 6–17 years¹³. It has been adapted for persons with EB¹⁴. In addition, the Pediatric Outcomes Measurement Information System (PROMIS) is a group of self-reported scales that include upper extremity skills and ADL for ages 8–17 and can be web based¹⁵.

Expanding this study to other multidisciplinary EB centers would improve statistical power through a greater number of subjects and generalizability through geographic diversity.

Implications for Occupational Therapy:

Persons living with RDEB present with challenging disabilities. Presently there is no cure and comprehensive care of EB patients requires the integration of MDT management including OT¹⁶. OT helps people across the lifespan to do the things they want and need to do through the therapeutic use of daily activities/occupations. One of the profession's core beliefs is in the positive relationship between occupation and health and its view of people as occupational beings¹⁷.

This study has potential implications for the role of OTs working with persons with EB and their caregivers including:

- ADLs that have value to patients should be identified
- Caregivers should be encouraged to allow their children with EB to engage in ADLs to the best of their abilities
- OTs are recommended to work with persons with EB to provide modifications to limit potential blistering and injury during the developmental stages
- Persons with EB should be encouraged to engage in ADL throughout their lifespan with OT consultation for adaptive equipment as needed

Conclusion

The results of this study highlight a positive trend of a correlation of independence in ADL and self-perceived QOL, but further research utilizing an ADL measure that is more robust and informative is needed. Ultimately this would provide information valuable to persons with EB, their caregivers, and treating OTs regarding the importance of allowing and encouraging individuals with one of the most debilitating forms of EB to perform ADL tasks to the best of their abilities.

Methods

Study design:

This is a qualitative, cross-sectional study, covering ages range from 9–25.

Participants: The inclusion criteria were as follows: all patients diagnosed with RDEB were recruited from the monthly epidermolysis bullosa clinic at a major metropolitan hospital from October 2014 through August 2017. Patients needed to be able to read English and answer the questions independently from their caregivers and the OT conducting the study.

Data collection:

The first questionnaire included a list of 4 ADL tasks and level of independence needed to complete them. Specific ADLs measured were the ability to perform dressing, toileting, and grooming self-care tasks and ability to perform the physical activities needed to complete schoolwork. The patients indicated their abilities as either dependent, independent, or able with some assistance. Items are scored 0–2 and summary scores range from 0–8 with higher scores indicating greater independence.

The second questionnaire, the QOLEB is a 17-item EB-specific measurement. The QOLEB tool has been reported to be validated and reliable for various subtypes of EB and has the potential to identify dimensions of QOL as targets for intervention and research¹⁸. The QOLEB assesses physical, psychological, and social aspects of QOL specific to patients with EB. Items are scored 0–3 and summary scores range from 0–51, with higher values indicating more disability. All questionnaires were completed on paper during one session with the OT.

Data Analysis: Parameters collected included descriptive statistics for continuous variables presented as means and standard deviations or medians and interquartile ranges, depending on normality. Categorical variables are presented with counts and percentages. Furthermore, the relationships between the ADL and QOLEB scores were analyzed using Spearman’s correlation test. The QOLEB scores were split up into two sub-scores based on identified categories: functional and emotional. The relationship between these two sub-scores and ADL scores were further analyzed using Spearman’s correlation tests. All analyses were completed in RStudio (Boston, MA) using a two-sided level of significance of 0.05.

Abbreviations

ADL

Activities of Daily Living; COPM:Canadian Occupational Performance Measure; COSA:Child Occupational Self-Assessment; CPG:Clinical Practice Guideline; DEB:Dystrophic EB; DDEB:Dominant DEB; EB:Epidermolysis Bullosa; EBS:EB simplex; JEB:Junctional EB; KEB:Kindler EB; MDT:Multidisciplinary Team; OT:Occupational Therapist; PROMIS:Pediatric Reported Outcomes Measurement Information; QOL:Quality of Life; RDEB:Recessive DEB

Declarations

Ethics and approval:

The Stanford University Institutional Review Board approved this cross-sectional study, #29122. Informed consent was obtained from patients 18 years and older or the parents of children under 18 years of age with the patient’s assent.

Consent for publication: Not applicable

Availability of data and materials: The datasets used and or analysed during the current study are available from the corresponding author on reasonable request.

Competing interests: The authors declare that they have no competing interests.

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Authors' contributions:

JC recruited the subjects for the study, acquired subjects' consent or assent an responses, and was the primary contributor in writing the manuscript.

NS analyzed and interpreted the patient data from patient questionnaires.

AL contributed to the conception and design of the study and acted as an advisor throughout the process.

All authors read and approved the final manuscript.

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Figures

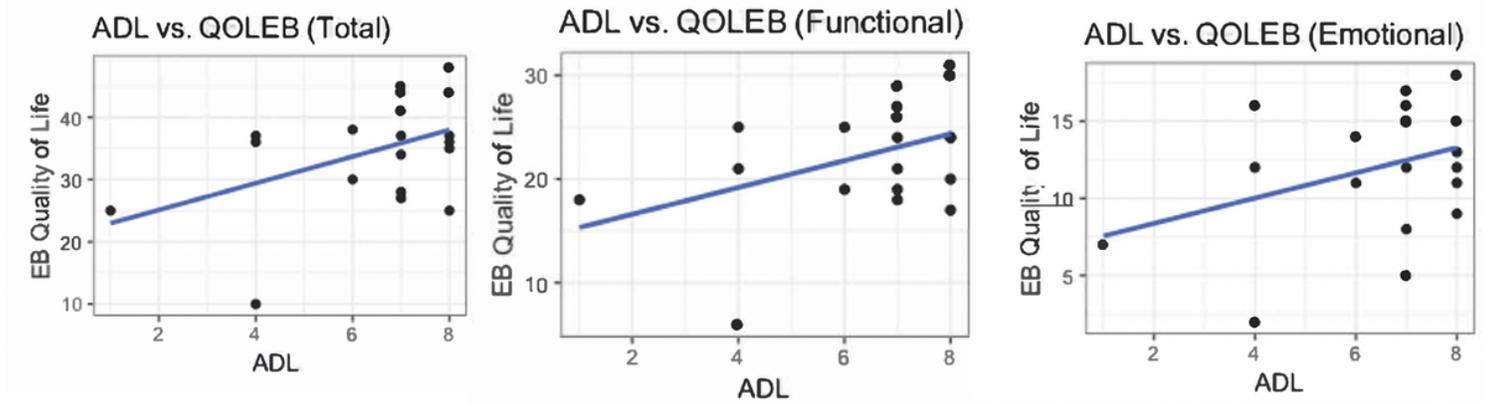


Figure 1

a: ADL vs QOLEB (Total) b: ADL vs QOLEB (Functional) c: ADL vs QOLEB (Emotional)