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ORIGINAL RESEARCH

Assessing Quality of Life in Patients with Epidermolysis Bullosa in the Saudi Population: Validation of the QOLEB Questionnaire

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Background: Epidermolysis bullosa (EB) is a rare condition where skin easily blisters. Using an Arabic-translated Quality of Life in Epidermolysis bullosa (QOLEB) questionnaire, we evaluated quality of life across persons with EB in Kingdom of Saudi Arabia.

Methods: Respondents were selected through nonprobability convenience sampling. Data were collected using a pre-validated questionnaire. We statistically compared response data across the four EB phenotypes using the Kruskal–Wallis test. Validity was assessed through confirmatory factor analysis and Cronbach's alpha was used to evaluate reliability with results 0.88.

Results: The study included 91 participants with Epidermolysis Bullosa (EB), comprising 60.4% of them were males. Adults and adolescents constituted 61.5% of the study sample. Children with EB faced more bathing challenges, with 64.3% always needing assistance compared to 31.4% of adults/adolescents. Writing adaptations varied significantly (p=0.03), with children exploring alternatives like typing, while adults primarily struggled to hold a pen.

Conclusion: Our findings underscore the profound physical, psychological, and social burdens associated with this rare condition, emphasizing the critical need for multidisciplinary care approaches. Addressing gaps in public awareness, improving access to specialized care, and providing psychosocial support for patients and their families are essential steps toward enhancing quality of life.

Keywords: epidermolysis bullosa, quality of life, questionnaire, Saudi Arabia

Introduction

Inherited epidermolysis bullosa (EB) refers to rare mutations that cause pathological disruptions in the anatomy—and hence physiology—of connective tissues.[1] Epidermolysis bullosa is often misconceived as merely a skin disease, when in reality, it is a complex, multi-organ condition, given that EB-causing mutations bring about defects in mucosae, this disease is also manifested in the gastrointestinal tract, oral cavity and the largest organ of the body (ie, the skin).[2] In EB, mutations preclude proper cohesion of skin tissue which, in turn, cause the skin to blister very easily.

Epidermolysis bullosa occurs equally in women and men; the prevalence of this condition is—more or less—equal across all races and ethnicities.[3] As per the peer-reviewed literature, the prevalence of EB is 10 per 1 million population —while the incidence of it is 20 per 1 million live births.[4] There are 4 major types of EB,[2] where about 70% of EB cases are of the Epidermolysis Bullosa Simplex (EBS) type; Dystrophic Epidermolysis Bullosa (DEB) and Junctional Epidermolysis Bullosa (JEB) constitute 25% and 5% of cases, respectively; Kindler Epidermolysis Bullosa (KEB), the rarest type, has been reported in 400 individuals globally.[5] Conventionally, EB usually refers to "inherited epidermolysis bullosa" (or "hereditary epidermolysis bullosa"), and the condition is distinguished from the non-inherited form, namely,

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epidermolysis bullosa acquisita (EBA).[5] There are currently no curative therapies for EB and the available treatments address symptoms at best.[6]

Given that many tissues and organs constitute mucous membranes, this inherited disorder impacts many aspects of life which, in turn, hampers psychological and social well-being; thus, many questionnaires have been used to capture non-biological dimensions of the disorder—such as the "Quality of Life in Epidermolysis bullosa" (QOLEB),[7] Dermatology Life Quality Index (DLQI), the Family Dermatology Life Quality Index (FDLQI) and Epidermolysis bullosa Burden of Disease (EB-BoD).[7,8]

Treatment, which sometimes involves repeated bandaging, makes the management of the condition costly; having this disorder may also affect a person's interpersonal relationships. Downstream consequences of EB also include osteopenia, osteoporosis, delayed puberty, cardiomyopathy, renal failure, and susceptibility to skin squamous cell carcinomas.[6] Squamous cell carcinomas (SCCs) in patients with dystrophic epidermolysis bullosa (DEB) are typically more aggressive than those in the general population, with a higher propensity for early metastasis. Treatment options are limited, as wide surgical excisions often result in chronic, non-healing wounds. In such cases, immunotherapy with Cemiplimab has shown promise as a first-line treatment. Upon disease progression, conventional chemotherapy may be considered. Notably, SCCs in DEB patients tend to develop at a younger age—typically between 35 and 50 years—and predominantly affect the extremities.[9] The chronic skin blistering partly leads to patient keeping physical activity to a minimum, care takers of persons with EB often find the management of the condition burdensome.[10] Hence the impact of EB to "self" (ie, the psychological effects) and on others (ie, the social effects) can altogether influence quality of life. Anemia, reduced life-expectancy and delayed puberty are also consequences of this condition.[6] The non-biological challenges of EB include significant costs, both direct and indirect. Living with EB often—if not always—leads to missed days from school or work, affecting both the individual and their caretaker(s).[11]

This rare condition has been investigated in various countries[12,13] and endeavours have been made to study EB in the Saudi population.[2,14] For instance, a study conducted by Alharthi et al[2] genetically profiled and reported EB cases in Saudi Arabia they found a total of 24 homozygous genetic variations were identified, with 14 being novel mutations. The most frequently implicated gene was COL7A1, found in 12 cases (42.9%), followed by LAMB3 in 5 cases (17.9%) and TGM5 in 4 cases (14.3%).[2] The majority of mutations were autosomal recessive (89.3%), and 87.5% of cases had homozygous mutations.[2] To explore the impact, distribution of this disease and provide baseline information, this study, therefore, aimed investigated quality of life across persons with EB in Saudi Arabia also assessed the reliability and validity of the Arabic translated QOLEB tool.

Materials and Methods

An analytical cross-sectional study was conducted amongpatients with EB. Also it was done in accordance with the Strengthening the Reporting of Observational Studies in Epidemiology (STROBE) guidelines.[15]

We translated the QOLEB questionnaire into Arabic (from English) and back translated it for clarity, and this translation was approved by Professor Dedee Murrell (license number 41) from the Australasian Blistering Disease.[6] Additionally, we incorporated two questions addressing pain and itching scales from the Yazdanshenas et al[16] study to enhance the questionnaire's comprehensiveness. The questionnaire comprises 5 domains with likert scale from (strongly agree to strongly disagree). To ensure comprehension by the participants, a pilot study involving 10 participants was conducted. Although the pilot study answers were excluded from the final analysis, the input was carefully reviewed, and adjustments were made to rectify any issues with double-barreled, confusing, and misleading questions. This iterative process resulted in developing a finalized version of the questionnaire; a copy of the finalized Arabic-translated QOLEB questionnaire is provided in the supplementary materials (Table S1). We administrated the Arabic-translated tool to participants identified through an "EB Network Group" participants were selected through nonprobability convenience sampling and provided their answers through "Electronic Forms".

We also utilized the completed survey to evaluate the validity and reliability of the Arabic-translated QOLEB questionnaire. Validity was assessed using confirmatory factor analysis (CFA), while reliability was measured with a standardized Cronbach's alpha. The Cronbach's alpha for our survey data was 0.88 which, according to the literature, represents a good level of reliability as it exceeds the accepted threshold of 70%.[17] Similarly, the results of our CFA

supports an acceptable/good level[18] of validity (Comparative Fit Index (CFI) = 0.913; Tucker-Lewis Index (TLI) = 0.899; Root Mean Square Error of Approximation (RMSEA) = 0.076).

Survey responses were collated and managed in Microsoft Excel spreadsheets. All data analyses, including those for reliability and validity, were conducted with the *R* software.[19] Frequency distribution was performed for categorical variables expressed in numbers and percentages. Pearson's Chi-squared test was used to compare response variables and explanatory variables. To explore age-related differences, subgroup analyses were conducted comparing children with adolescents and adults. This stratification was designed to assess variations in responses across age groups.

This study was ethically reviewed and approved by the Local Bioethics Committee at ministry of health, Jeddah, Saudi Arabia on February 14, 2024 (approval number: A01852). This study was conducted according to the Declaration of Helsinki. Participants were also assured of anonymity and the confidentiality of their responses to the questionnaire. Also the assent was taken from the parents for patients less than 14 years old. As per current regulations in the Kingdom of Saudi Arabia, research involving minors (participants under the age of 14) generally requires parental or legal guardian consent, which was obtained in our study. This is in accordance with the guidelines set by the National Committee of Bioethics (NCBE) and the Saudi Health Council.

Results

The study included 91 participants with Epidermolysis Bullosa (EB), comprising 60.4% males and 39.6% females. Adults and adolescents constituted 61.5% of the sample, with children representing 38.5%. The majority of participants (85.7%) were single, and 47.3% reported no income. EB subtypes were distributed as EB simplex (33%), Junctional EB (24.2%), Dystrophic EB (34%), and unclassified (8.8%). A notable characteristic was the high consanguinity rate of 78% (Table 1).

Functional Limitations

Table 2 provides a detailed comparison of functional limitations between children and adults/adolescents with Epidermolysis Bullosa (EB). Statistically significant differences were observed in several domains. Bathing and Personal Care (p=0.02) showed the most dramatic variation. While only 1.8% of children reported no bathing difficulties,

Table 1 Summary of Demographic Characteristics of Survey Participants with Epidermolysis Bullosa

Variables		Count (N)	Frequency (%)
Sex	Female	36	39.6%
	Male	55	60.4%
Age	Child	35	38.5%
	Adult/Adolescent	56	61.5%
Marital status	Married	12	13.2%
	Separated	1	1.1%
	Single	78	85.7%
Income	There is no income	43	47.3%
	Less than 10 thousand riyals (ie, $2666)	19	20.9%
	From 10 thousand to 20 thousand riyals (ie, $2666 – $5333)	5	5.5%
	More than 20 thousand riyals (ie, $5333)	5	5.5%
	I do not want to disclose	19	20.9%

(*Continued*)

Table 1 (Continued).

Variables		Count (N)	Frequency (%)
Education	Uneducated	32	35.2%
	Primary	26	28.6%
	Secondary-High school	10	11%
	Undergraduate-Bachelor	12	13.2%
	Diploma	2	2.2%
	Intermediate	8	8.8%
	Postgraduate studies	1	1.1%
Subtypes of Epidermolysis Bullosa	Simplex EB	30	33%
	Junctional EB	22	24.2%
	Dystrophic EB	31	34%
	Unclassified	8	8.8%
Consanguinity	Yes	71	78%
	No	20	22%

Table 2 Comparison of Functional Limitations in EB Between Children and Adults/Adolescents

Question	Response Options	Children N = 56	Adult/ Adolescent N = 35	P-value
Q1: Does your EB affect your ability to move around at home?	• It does not affect me at all	3 (5.4%)	8 (22.9%)	0.09
	• A little	26 (46.4%)	12 (34.3%)	
	• Somewhat	20 (35.7%)	11 (31.4%)	
	• A lot	7 (12.5%)	4 (11.4%)	
Q2: Does your EB affect your ability to bath or shower?	• No, it does not affect me at all	1 (1.8%)	7 (20.0%)	**0.02**
	• Yes, I sometimes need assistance	10 (17.9%)	11 (31.4%)	
	• Yes, I often need assistance	9 (16.1%)	6 (17.1%)	
	• Yes, I always need assistance while bathing	36 (64.3%)	11 (31.4%)	
Q3: Does your EB cause you physical pain?	• It does not cause me any pain	1 (1.8%)	1 (2.9%)	0.98
	• It causes me pain occasionally	15 (26.8%)	9 (25.7%)	
	• It causes me pain frequently	22 (39.3%)	14 (40.0%)	
	• It causes me constant pain	18 (32.1%)	11 (31.4%)	
Q4: How does your EB affect your ability to write?	• It does not affect my ability to write at all	15 (26.8%)	13 (37.1%)	**0.03**
	• I have difficulty holding a pen	19 (33.9%)	18 (51.4%)	
	• I find typing easier than handwriting	12 (21.4%)	1 (2.9%)	
	• I cannot write due to my condition	10 (17.9%)	3 (8.6%)	

(*Continued*)

Table 2 (Continued).

Question	Response Options	Children N = 56	Adult/ Adolescent N = 35	P-value
Q5: Does your EB affect your ability to eat?	• No, I eat normally	10 (17.9%)	4 (11.4%)	0.79
	• A little	19 (33.9%)	14 (40.0%)	
	• A lot	25 (44.6%)	15 (42.9%)	
	• I rely on a feeding tube for nutrition	2 (3.6%)	2 (5.7%)	
Q6: Does your EB affect your ability to go shopping?	• It does not affect me at all	5 (8.9%)	5 (14.3%)	0.15
	• A little	18 (32.1%)	15 (42.9%)	
	• I always need assistance	18 (32.1%)	4 (11.4%)	
	• A lot	15 (26.8%)	11 (31.4%)	
Q7: How does EB affect your involvement in sports?	• It does not affect me at all	0 (0.0%)	1 (2.9%)	0.08
	• I need to be cautious when playing sports	12 (21.4%)	5 (14.3%)	
	• I avoid certain sports	15 (26.8%)	17 (48.6%)	
	• I avoid all sports	29 (51.8%)	12 (34.3%)	
Q9: Does your EB affect your ability to move around outside of your home?	• It does not affect me at all	3 (5.4%)	6 (17.1%)	**0.03**
	• A little	20 (35.7%)	17 (48.6%)	
	• Somewhat	25 (44.6%)	6 (17.1%)	
	• A lot	8 (14.3%)	6 (17.1%)	
Q10: How does your EB affect your relationships with family members?	• It does not affect me at all	27 (48.2%)	19 (54.3%)	0.35
	• It has a small impact	20 (35.7%)	7 (20.0%)	
	• It has a significant impact	7 (12.5%)	6 (17.1%)	
	• It has a very significant impact	2 (3.6%)	3 (8.6%)	
Q12: Have you needed to, or do you need to modify your home (installing ramps etc.) due to your EB?	• I do not need it at all	19 (33.9%)	18 (51.4%)	0.18
	• A little	19 (33.9%)	8 (22.9%)	
	• A lot	11 (19.6%)	8 (22.9%)	
	• Yes, I urgently need it	7 (12.5%)	1 (2.9%)	
Q13: Does your EB affect your relationships with friends?	• It does not affect me at all	16 (28.6%)	11 (31.4%)	0.67
	• A little	18 (32.1%)	13 (37.1%)	
	• A lot	14 (25.0%)	5 (14.3%)	
	• It severely restricts my social interaction	8 (14.3%)	6 (17.1%)	
Q15: How are you or your family affected financially by your EB?	• It does not affect us	6 (10.7%)	5 (14.3%)	0.33
	• It affects us a little	10 (17.9%)	11 (31.4%)	
	• It affects us a lot	27 (48.2%)	11 (31.4%)	
	• It affects us severely	13 (23.2%)	8 (22.9%)	

Note: Data in bold indicates statistical significance ($p < 0.05$).

20% of adults/adolescents experienced no issues. Conversely, 64.3% of children always needed assistance while bathing, compared to 31.4% of adults/adolescents. Writing abilities differed significantly (p=0.03). Children demonstrated more varied adaptations, with 21.4% finding typing easier and 17.9% unable to write, while adults/adolescents primarily reported difficulty holding a pen (51.4%). Movement and mobility both inside and outside the home showed moderate impacts, though not statistically significant.

Emotional Impact

Table 3 examined the emotional dimensions of EB using the Quality of Life in Epidermolysis Bullosa (QOLEB) scores. Despite no statistically significant differences, notable variations emerged across emotional experiences. Frustration levels showed nuanced patterns, with 48.2% of children feeling "a little" frustrated compared to 40% of adults/adolescents. Embarrassment experiences were similarly distributed, with 51.8% of children and 57.1% of adults/adolescents reporting "a little" embarrassment. Anxiety and depression revealed interesting trends. Adults/adolescents reported more moderate anxiety (60% "a little" anxious) compared to children (33.9%). Depression scores showed similar distributed responses, with 31.4% of adults/adolescents reporting no depression versus 21.4% of children.

Table 3 Comparison of Emotional Impact (QOLEB) of EB Between Children and Adults/Adolescent

Question	Response Options	Children N = 56	Adult/ Adolescent N = 35	P-value
Q8: How frustrated do you feel about your EB?	• I do not feel frustrated	6 (10.7%)	6 (17.1%)	0.65
	• I feel a little frustrated	27 (48.2%)	14 (40.0%)	
	• I feel very frustrated	14 (25.0%)	11 (31.4%)	
	• I feel frustrated and angry most of the time	9 (16.1%)	4 (11.4%)	
Q11: How embarrassed do people make you feel about your EB?	• They do not embarrass me at all	9 (16.1%)	3 (8.6%)	0.73
	• I feel a little embarrassed	29 (51.8%)	20 (57.1%)	
	• I feel very embarrassed	9 (16.1%)	5 (14.3%)	
	• I feel extremely embarrassed	9 (16.1%)	7 (20.0%)	
Q14: How worried or anxious do you feel because of your EB?	• I do not feel any anxiety at all	5 (8.9%)	1 (2.9%)	0.09
	• A little	19 (33.9%)	21 (60.0%)	
	• A lot	18 (32.1%)	7 (20.0%)	
	• I feel highly anxious	14 (25.0%)	6 (17.1%)	
Q16: How depressed do you feel because of your EB?	• I do not feel depressed at all	12 (21.4%)	11 (31.4%)	0.18
	• I feel a little depressed	31 (55.4%)	12 (34.3%)	
	• I feel very depressed	9 (16.1%)	6 (17.1%)	
	• I feel depressed all the time	4 (7.1%)	6 (17.1%)	
Q17: How uncomfortable are you made to feel by others (eg teasing or staring) because of your EB?	• I do not feel any discomfort at all	10 (17.9%)	5 (14.3%)	0.59
	• A little	22 (39.3%)	15 (42.9%)	
	• A lot	19 (33.9%)	9 (25.7%)	
	• So much that I avoid all social activities	5 (8.9%)	6 (17.1%)	

EB Subtypes and Quality of Life

Table 4 provided a comprehensive analysis of EB subtypes in relation to QOLEB scores and related conditions. While overall QOLEB total scores did not differ significantly (p=0.17), specific severity classifications showed notable variations.

Mild score classifications differed significantly (p=0.03), with EB simplex showing 80% of mild cases. Moderate score classifications also varied significantly (p=0.03), with more diverse distribution across subtypes.

Epidermolysis Bullosa: Correlation Analysis

Figure 1 revealed correlations between Epidermolysis Bullosa (EB) types and quality of life measures. The functional score showed a strong correlation with total score (r=0.948, p<0.001), while the emotional score demonstrated significant correlation (r=0.814, p<0.001) with total score. Sleep quality exhibited a moderate correlation (r=0.632, p<0.001) with total score, and itching displayed a weak but significant relationship with emotional scores (r=0.302, p=0.004).

Discussion

This study explored the demographic, functional, and emotional impact of Epidermolysis Bullosa (EB) on a cohort of patients in Saudi Arabia, providing valuable insights into the disease's burden. With 91 participants, this study contributes to a growing body of literature on EB in the region, highlighting unique socio-demographic and disease-related characteristics. The findings underline the necessity of tailoring interventions to improve the quality of life (QoL) for patients while addressing broader systemic and societal challenges.

The study revealed that EB disproportionately affects males (60.4%) and individuals from socioeconomically disadvantaged backgrounds, with nearly half (47.3%) reporting no income. Additionally, the high prevalence of consanguinity (78%) is consistent with regional genetic predispositions. These findings are in line with studies conducted in other Middle Eastern populations, where consanguinity amplifies the risk of autosomal recessive disorders, including EB subtypes such as Junctional and Dystrophic EB.[18] The significant proportion of uneducated participants (35.2%)

Table 4 Distribution of the Epidermolysis Bullosa Subtypes in Relation to the QOLEB Scores and Other Related Conditions

Study Sample N=*91*	**EBS n = *30***	**JEB n = *22***	**DEB n = *31***	**Unclassified n = *8***	**P-value**
QOLEB median scores: Median					
Total score (0–51)	17	19	20	14	0.17
Functioning (0–36)	6	6	7	7	0.84
Emotions (0–15)	25	25	27	21	0.27
Classification of the QOLEB total score: n (Row %)					
Very mild 0–4 points	0 (0)	0 (0)	1 (100)	0 (0)	0.5
Mild 5–9 points	4 (80)	0 (0)	1(20)	0 (0)	**0.03**
Moderate 10–19 points	5 (25)	7 (35)	4 (20)	4 (20)	**0.03**
Severe 20–35 points	18 (35)	11 (21.6)	18 (35)	4 (7.8)	0.9
Very severe 35–51 points	3 (21.4)	4 (28.6)	7 (50)	0 (0)	0.8
Other related conditions: Median					
Quality of Sleep (0–3)	1	1	2	1	0.12
Itching (0–3)	2	2	2	2	0.07

Note: Data in bold indicates statistical significance (p < 0.05).

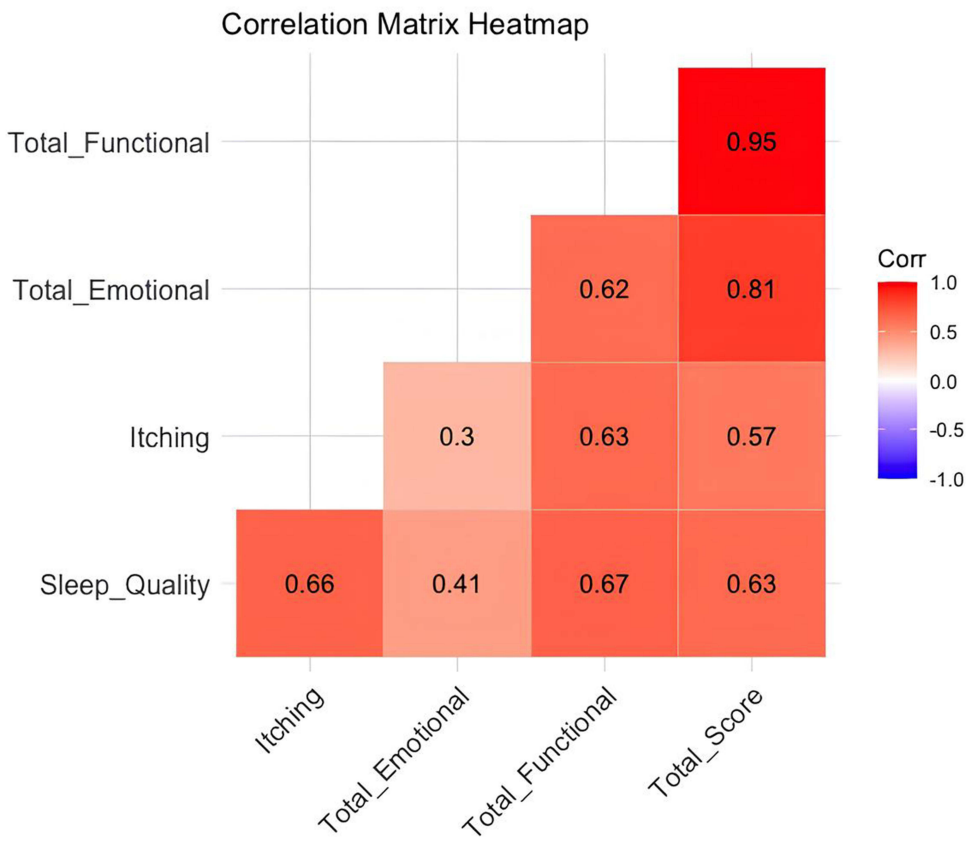

Figure 1 Correlation matrix heatmap of QOLEB sections and other related conditions.

further emphasizes the socioeconomic burden of EB, which may limit access to healthcare resources, awareness campaigns, and employment opportunities. Addressing these disparities through genetic counseling, public health initiatives, and educational outreach is essential.[20]

The comparison of functional limitations between children and adults/adolescents highlighted critical age-related differences in disease impact. Statistically significant variations were observed in bathing difficulties (p=0.02), writing abilities (p=0.03), and mobility outside the home (p=0.03). Adults and adolescents exhibited greater independence, likely due to adaptive strategies developed over time.[21,22] However, children were disproportionately affected in essential daily activities, with 64.3% always requiring bathing assistance compared to 31.4% of adults. These findings align with previous studies suggesting that the functional burden of EB is particularly severe in younger patients, potentially hindering their social development and independence.[22,23] Tailored occupational therapy and assistive technologies may help bridge these gaps, promoting greater autonomy across all age groups.[24]

Despite the absence of statistically significant differences in emotional parameters, the data revealed nuanced variations in frustration, embarrassment, anxiety, and depression across age groups.[25,26] Children reported higher levels of frustration and severe depression, while adults and adolescents experienced moderate anxiety more frequently. These patterns are consistent with the cumulative psychosocial burden of EB, which has been well-documented in the literature.[25,26] Chronic pain, visible skin lesions, and dependence on caregivers exacerbate feelings of isolation and stigmatization.[26] Psychological support and community-based interventions are critical to mitigating these impacts, particularly for children transitioning to adolescence.[26]

Interestingly, while both age groups reported embarrassment due to social interactions, adults appeared better equipped to navigate these challenges, possibly reflecting their accumulated coping mechanisms.[27] This underscores the importance of fostering resilience and self-efficacy in younger patients through targeted mental health interventions.[27] The findings also call for greater societal awareness to reduce stigma and improve the social integration of individuals with EB.[27,28]

Analysis of the EB subtypes revealed notable differences in QoL scores, although overall scores did not reach statistical significance. Mild and moderate severity classifications exhibited significant variation across subtypes (p=0.03). EB simplex patients, who typically experience milder symptoms, dominated the mild classification group, while Junctional and Dystrophic EB were associated with more severe disease manifestations.[29] These findings highlight the heterogeneity of EB and its varying impact on QoL, emphasizing the need for subtype-specific interventions.[27] The relationship between EB subtypes and related conditions such as sleep quality and itching was also explored.[30] While these parameters did not differ significantly across subtypes, the high median scores for itching (2 out of 3) across all groups highlight the pervasive and distressing nature of this symptom. Pruritus management should therefore be prioritized in clinical practice, as it directly impacts sleep and overall QoL.[24] The National EB Registry in the United States reported that by the age of 35 this risk is 67.8% and reaches 90.1% at age 55". This influences QOL in patients with EB dramatically.[9]

Implications for Clinical Practice and Policy

This study underscores the multifaceted impact of EB, which extends beyond physical symptoms to encompass profound emotional and socioeconomic challenges. Healthcare providers must adopt a multidisciplinary approach to manage the disease, incorporating dermatological, psychological, and social support services.[31] Routine assessments of functional and emotional well-being using validated tools such as the QOLEB should guide individualized care plans.[32] For instance, learning aids play a crucial role in helping children with EB overcome physical challenges in education. Tools such as tablets, voice-to-text software, and ergonomic devices minimize discomfort, while interactive technologies promote engagement and creativity. These resources support inclusive learning environments, empowering children with EB to succeed academically and build self-confidence. Similarly, specialized shower aids can significantly improve their independence and quality of life. Features like handheld showerheads with gentle settings, padded surfaces, and non-abrasive cleansing options help reduce pain and prevent injury during bathing. By enabling safe participation in hygiene routines, these aids foster autonomy, enhance self-esteem, and lessen reliance on caregivers. Additionally, adaptive technologies, including soft-touch materials and temperature controls, ensure a more comfortable and stress-free experience.

On a policy level, the findings highlight the urgent need for genetic screening programs to address the high consanguinity rate and prevent the intergenerational transmission of EB.[31] Furthermore, financial assistance programs for affected families could alleviate the economic burden, enabling better access to treatment and supportive care.

Limitations

This study has several limitations that should be acknowledged. The sample size, while comparable to similar studies, may limit the generalizability of the findings to the broader Saudi population. The reliance on self-reported data introduces the potential for recall and social desirability bias, particularly in sensitive areas such as emotional well-being. Additionally, the cross-sectional design precludes causal inferences about the relationships between disease characteristics and quality of life. The study did not account for potential confounders such as comorbid conditions or treatment adherence, which may have influenced the results.

Conclusion

This study sheds light on the multifaceted challenges faced by individuals living with Epidermolysis Bullosa (EB) in Saudi Arabia. Our findings underscore the profound physical, psychological, and social burdens associated with this rare condition, emphasizing the critical need for multidisciplinary care approaches. Addressing gaps in public awareness, improving access to specialized care, and providing psychosocial support for patients and their families are essential steps toward enhancing quality of life. Moreover, provide personalized care plans, assistive devices and training to the patients can help in improving their quality of life. Future research should focus on larger, diverse populations and longitudinal designs to further explore the long-term impacts of EB and to develop targeted interventions for comprehensive patient care. The burden associated with EB leads to the need of a multidisciplinary approach dermatologists,

genetics, pediatrics, internal diseases, oncologists in cases of squamous cell carcinoma. Establishing a comprehensive EB registry to monitor prevalence and outcomes.

Ethics Approval Statement

This study was ethically approved by Research Ethics Committee at King Abdullah Medical Complex (approval number: A01852).

Disclosure

The authors have no conflict of interest to declare.

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