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ORIGINAL RESEARCH

Exploring the Impact of Epidermolysis Bullosa on Parents and Caregivers: A Cross-Cultural Validation of the Epidermolysis Bullosa Burden of Disease Questionnaire

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Purpose: Epidermolysis bullosa (EB) is a heterogeneous group of genetically inherited skin and mucosal fragility disorders. EB may have a profound impact on parental physical and psychosocial health. This study was designed to evaluate the disease burden in parents of patients with EB and identify out-of-pocket (OOP) expenditures for EB care in Saudi Arabia.

Patients and Methods: Thirty-eight caregivers of patients with EB were recruited from the Saudi EB registry to participate. All participants completed the EB Burden of Disease (EB-BoD) questionnaire. Data were collected between May 2020 and December 2020. The sample included 10 patients with EB simplex (EBS), 10 with junctional EB (JEB), 14 with dystrophic EB (DEB), and 4 with an unknown type.

Results: Mothers were the primary caregivers in 89.5% of cases. The mean EB-BoD score was 53 ± 21.5. The family-life and child's life dimensions had the higher burden. The mean EB-BoD score observed in patients with DEB was 62.4±16.8 versus 45.7 ± 19.42 for EBS. The EB-BoD score was correlated with the patient's family income. Most caregivers (97.4%) reported OOP expenditure, with a mean monthly OOP expenditure of $575.5± $701.1. OOP expenses increased with the severity of the condition.

Conclusion: This study highlights the need for support services for parents caring for patients with EB.

Keywords: burden, caregivers, epidermolysis bullosa, quality of life

Introduction

Epidermolysis bullosa (EB) is a heterogeneous group of genetically inherited skin and mucosal fragility disorders. It is classified into four major classical EB types based on the level of skin cleavage: EB simplex (EBS), Junctional EB (JEB), Dystrophic EB (DEB), and Kindler EB.1 Clinical severity and extracutaneous manifestations vary considerably between EB types and subtypes.[1,2] EB Management focuses on wound care, symptom control, and treatment of complications.[3]

Few studies have addressed the psychosocial impact of EB on affected families, including its impacts on family size and relationships.[4–6] Caregivers face many challenges, and previous studies have found that parents experience increased family expenses and significant stress due to the disease.[4,7,8] The health costs of EB management affect the caregiver's budget, particularly if there are substantial out-of-pocket (OOP) expenditures for care needs. Proper recognition and understanding of the impact of EB on affected patients and their families are crucial. This can help to implement family support programs to ease the burden on families and improve their quality of life.[6,9] This study aimed to assess the EB disease burden on the caregiving parents of patients with EB and measure the OOP expenditures of EB care in Saudi Arabia.

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Materials and Methods

Study Population Variables and Measurement

This was a cross-sectional study of EB families. The parents of patients from the Saudi EB registry were approached to participate in this survey. Patients were eligible for inclusion if they were diagnosed with EB at any age, if Arabic was their native language, and if their caregivers provided informed consent. All participants were informed about the study objective and data confidentiality and were asked to indicate their understanding of the study conditions and agreement to participate. The Institutional Review Board of the Imam Mohammad Ibn Saud University approved the study protocol.

Variables and Measurement

We measured the EB burden using the EB Burden of Disease (EB-BoD) questionnaire, as shown in Table SI.[10] The EB-BoD is a validated and reliable family burden-specific tool comprising 20 items.[10] It is a multidimensional instrument that evaluates family life, the child's life, disease and treatment, and economic and social impacts. For each item, a six-point Likert scale was used (never, rarely, sometimes, often, very often, and constantly) to limit missing data. "Not applicable" was also included. Dimension scores were calculated by totaling the individual item scores. A global score, the total of all individual item scores, was transformed into a scale of 0–100 points, with a higher score reflecting a higher EB burden. The English version of the EB-BoD questionnaire has been translated and has undergone linguistic and cultural adaptation to the Arabic language.

Procedure

We retrieved a list of patients with EB who met the inclusion criteria from the Saudi EB database. The parents of patients with EB were contacted and invited to participate in this study. A link to the questionnaire was sent to participants via phone or email. Data were collected from patients' primary caregivers. The participants received the informed consent form, EB-BoD, and clinical research form for demographic and clinical information. Data were collected between May 2020 and December 2020. The Ethics Committee of Imam Mohammad Ibn Saud University approved this study (02–2020). This study adhered to the Declaration of Helsinki.

Statistical Analysis

We conducted data analysis using SPSS Windows software version 26. Continuous variables were presented as mean ± standard deviation (SD) and range. Categorical variables are presented as frequencies and percentages. Continuous variables were compared using *t*-test or Mann–Whitney *U*-test according to the normality of the distribution, while categorical variables were compared using the Chi-squared or Fisher's exact tests, as appropriate. All reported P-values were two-sided, and P-values <0.05 were considered statistically significant.

Results

A total of 38 caregivers completed the questionnaires. The patients' mean age ± SD was 11.3 ± 8.9 years (range: 1–35 years). Of them, 24 were men (63.2%), and 14 were women (36.8%). Table 1 shows the characteristics of the study participants. The cohort comprised 10 patients (26.3%) with EBS, 10 (26.3%) with JEB, 14 (36.8%) with DEB (3 dominant DEB and 11 recessive DEB), and 4 patients (10.5%) with an unknown type. The instrument's reliability was determined by analyzing the internal consistency of each domain using Cronbach's alpha. The alpha coefficient was considered good at 0.8.

Most primary caregivers had a college education or higher (63.1%). Mothers were the primary caregivers in 89.5% (n = 34) cases. Seven (20.6%) out of 34 mothers declared that they had to stop working altogether to care for their children with EB. Most patients (89.5%) needed assistance with their wound care regimen. Wound care was provided by the mother and a live-in home caregiver, by the mother alone, by both parents, and by a registered nurse in 50%, 23.5%, 23.5%, and 5.9% of cases, respectively. Approximately 84% of parents of patients with EB reported that the burden of care reduces family time such as going on vacation.

Table 1 Demographic and Clinical Characteristics of the 38 Patients with Epidermolysis Bullosa

	DEB	JEB	EBS	Unknown	Total
Number of patients, n (%)	14 (36.8%)	10 (26.3%)	10 (26.3%)	4 (10.5%)	38 (100%)
Gender					
Female	6 (42.9%)	4 (40%)	2 (20%)	2 (50%)	14 (36.8%)
Male	8 (57.1%)	6 (60%)	8 (80%)	2 (50%)	24 (63.2%)
Age (years), mean	9±6.43	10±8.52	18.6±10.01	4.25±2.5	11.3±8.9
Range	(1–21)	(2–24)	(6–35)	(3–8)	(1–35)
Nationality					
Saudi	11 (78.6%)	8 (80%)	10 (100%)	4 (100%)	33 (86.8%)
Other	3 (21.4%)	2 (20%)	0 (0%)	0 (0%)	5 (13.2%)
Number of families who lost a child due to EB	3 (21.4%)	4 (40%)	0 (0%)	1 (25%)	8 (21.1%)
Family history of EB, n (%)					
Yes	4 (28.6%)	5 (50%)	6 (60%)	3 (75%)	18 (47.4%)
No	9 (64.3%)	4 (40%)	4 (40%)	1 (25%)	18 (47.4%)
Unknown	1 (7.1%)	1 (10%)	0 (0%)	0 (0%)	2 (5.3%)
History of consanguineous marriage	11 (78.6%)	9 (90%)	6 (60%)	4 (100%)	30 (78.9%)
Average financial costs/month in US dollars	1036.75±1188.09	397.86±246.22	386.56±116.71	333.24±282.76	575.52±701.08
Range	(133.30–3998.88)	(106.64–799.78)	(266.59–533.18)	(133.30–533.18)	(106.64–3998.88)
Average monthly salary in US dollars[a]	1707.19±819.05	2042.93±1075.00	4038.87±3148.93	2532.62±188.51	2694.82±2205.91
Range	(799.78–2932.51)	(399.89–3199.10)	(293.25–10,663.68)	(2399.33–2665.92)	(293.25–10,663.68)

Notes: [a]This variable contains two influential outliers, with participant salaries of $6933 and $10,666.
Abbreviations: DEB, dystrophic epidermolysis bullosa; JEB, junctional epidermolysis bullosa; EBS, epidermolysis bullosa simplex; US, United States.

Patients above 18 years were more likely to be dependent on their family if they had JEB or DEB (50%) compared with EBS (10%). The mean EB-BoD score was 53 ± 21.5 (Table 2). The EB-BoD scores differed significantly between EB subtypes. Comparison of DEB and EBS groups showed a significant difference: 62.4±16.8 and 45.7 ± 19.42, respectively, p = 0.035 (Tables 2 and 3). The family and child life dimensions had higher burden scores (p = 0.041, p = 0.009, respectively), whereas the disease and socioeconomic dimensions were not significantly different (p = 0.361, p = 0.254, respectively) (Table 3). The EB-BoD score did not correlate with the patient's age (p = 0.113) or gender (p = 0.208). Families with incomes > $1800 per month had a mean EB-BoD score of 45.3 ± 21, while families with smaller incomes had an EB-BoD score of 62 ± 15.7, p = 0.051. Among the four dimensions, family life was the most impacted, with an EB-BoD score of 15.6±8 in the group with higher income compared with 22±6.8 in the lower-income group, showing a difference of 31%, p = 0.146. Most caregivers (97.4%) reported OOP expenditures, with a mean monthly OOP expenditures of $575.5± $701.1. This amount differed according to EB type: $386.6± $116.7 for EBS, $397.9± $246.2 for JEB, and $1036.8± $1188.1 for DEB. Approximately 70.3% of OOP expenditures were related to wound dressing products. The purchase of medication and emollients was reported by 46% of participants. Approximately 16.2% reported OOP expenditures related to special clothing items. Other reported OOP expenses included nutritional supplements (13.5%), cleansing products (5.4%), alternative and complementary medicines (2.7%), and travel costs (2.7%).

Table 2 Different Dimensions of the Epidermolysis Bullosa Burden of Disease Questionnaire in the Different Epidermolysis Bullosa Subtypes of the Cohort

	DEB	JEB	EBS	Unknown	Total
Global EB-BoD score	62.36±16.8	40.5±21.38	45.7±19.42	69.25±22.74	52.95±21.5
Range	(42 to 87)	(17 to 75)	(23 to 76)	(42 to 88)	(17 to 88)
Family life	20.5±7.04	16.5±7.79	13.2±9.44	23.25±3.2	17.82±8.17
Range	(8 to 35)	(7 to 28)	(4 to 28)	(20 to 26)	(4 to 35)
Economic and social impact	12.64±2.27	10±2.79	11.6±1.96	12±2.31	11.61±2.49
Range	(10 to 15)	(6 to 14)	(10 to 15)	(10 to 14)	(6 to 15)
Disease and treatment	11.43±6.19	6.6±7.43	9.3±4.37	15.5±10.34	10.03±6.89
Range	(4 to 22)	(0 to 17)	(3 to 17)	(3 to 24)	(0 to 24)
Child's life	17.79±4.95	7.4±5.82	11.6±5.48	18.5±7.14	13.5±7
Range	(10 to 25)	(2 to 18)	(6 to 21)	(9 to 24)	(2 to 25)

Note: Values are reported as mean ± SD.
Abbreviations: DEB, dystrophic epidermolysis bullosa; JEB, junctional epidermolysis bullosa; EBS, epidermolysis bullosa simplex.

Table 3 Comparison of the EB-BoD Scores Among Epidermolysis Bullosa Subtypes: P-values Determined by Student's *t*-Test

	DEB	JEB	EBS
DEB		0.010	0.035
JEB			0.576
EBS			

Abbreviations: DEB, dystrophic epidermolysis bullosa; JEB, junctional epidermolysis bullosa; EBS, epidermolysis bullosa simplex.

Discussion

Measuring EB's psychosocial impact and economic burden on patients' families helps to set priorities for healthcare resource allocation.[11–13] Few studies have explored the effects of EB on family units.[6] We examined the impact of EB on caregivers' daily lives and its financial consequences. The mean EB-BoD score for caregivers of patients with EB was 53 ± 21.5. The EB-BoD score was also evaluated according to the EB subtype. A significant difference was found between parents of patients with lower disease severity (EBS) and those with higher disease severity (DEB). The small sample size of the adult group may explain the absence of statistical differences in the EB-BoD scores concerning patient age. Our results suggest that EB places a considerable burden on caregivers. These results are in concordance with those of previous reports. A study of 28 families found that a higher perceived caregiver burden was associated with disease severity.[13] A study of 21 caregivers of children with EB reported a lower quality of life, including difficulties in life attitudes and satisfaction, compared with parents of healthy children.[14]

Furthermore, most primary caregivers (approximately 90%) in the current study were mothers. Mothers sometimes sacrifice their careers to care for affected children. Only 5.9% of participants reported having wound care performed by a registered home care nurse. Daily dressing changes can be a time-consuming and stressful procedure.[15] This is consistent with previous studies showing that mothers were the main caregivers for patients with EB and usually gave

up work to have time to provide the care needed.[16,17] This highlights the role of mothers in the care of patients with EB in Saudi Arabia and the need to initiate support programs to reduce their stress, thereby improving their quality of life.

The financial impact of EB on families was evident in our study. Families with a higher income > $1800 per month reported lower mean EB-BoD scores of 45.2 ± 20.5, compared with families with lower incomes (average EB-BoD score of 61 ± 20.7).

The mean monthly OOP health expenditures were $575.5± $701.1. To put these numbers into perspective, the mean monthly OOP health expenses for the general population in Saudi Arabia are, on average, $70.9, as reported by the Saudi General Authority for Statistics.[18] Hence, the OOP health expenditures of EB families are eight times higher than those of the Saudi population. Moreover, previous studies assessing OOP expenditures required to manage EB in France found mean monthly OOP to be $363.5± $380.4.[19] The considerably high OOP expenses reflect the complex care and management required for EB. Our findings indicate considerable OOP expenditure on specialized dressings for EB wounds. This finding is consistent with a previous study of 249 patients with EB and their caregivers living in the United States, wherein 73% of participants reported a significant or moderate financial impact on wound care supplies.[20] EB dressings should be nonadherent and atraumatic to protect fragile skin and prevent blistering.[21] These dressings are covered for medical costs by the government and some health insurance; however, increased demand and differences in insurance coverage might affect the supply of wound dressings. Therefore, healthcare authorities should re-evaluate nationally funded EB dressing schemes to ensure sufficient bandage coverage.

Furthermore, the current and previous studies indicate that EB considerably burdens patients and their families. This study explored the burden on caregivers of patients with EB in Saudi Arabia using the EB-BoD tool, which includes social and economic dimensions.[10]

This study has some limitations and highlights the challenges associated with measuring disease burden in a population with a rare disease. These include missing data, recall bias, and a small sample size.

Conclusion

Understanding and measuring the disease burden on the entire family is essential for prioritizing community projects for children and families living with chronic illness. Future research is required to better understand the need for support systems to improve the quality of life of patients with EB and alleviate the burdens faced by their caregivers.[9]

Our study demonstrated that EB places a considerable burden on caregivers. Therefore, multidisciplinary care and specific family support programs are necessary.

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Disclosure

The authors report no conflicts of interest in this work.

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