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Trabajo Original

Valoración nutricional

Health-related quality of life and clinical severity in people with epidermolysis bullosa — A proposal for assessing nutritional compromise by body mass index (Birmingham Epidermolysis Bullosa Severity Score)

Calidad de vida relacionada con la salud y gravedad clínica en personas con epidermólisis bullosa: una propuesta para evaluar el deterioro nutricional por el índice de masa corporal (Birmingham Epidermolysis Bullosa Severity Score)

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Abstract

Introduction and objectives: epidermolysis bullosa (EB) is a rare genetic disease characterised by skin fragility with blisters and erosions on the skin and/or mucous membranes. People with EB often experience several extracutaneous manifestations, including clinical and health-related quality of life (HRQoL) complications. Herein, we evaluate their HRQoL and clinical severity and propose an objective criterion for estimating nutritional compromise using the Birmingham Epidermolysis Bullosa Severity Score (BEBS) tool.

Methods: this series of cases included people with EB, monitored by a multi-professional team. Clinical severity was assessed with the BEBS, using body mass index ranges by age, as an objective proposal, to estimate the degree of nutritional compromise. To assess HRQoL, the Children's Dermatology Life Quality Index (individuals aged 4-16 years) and the Quality of Life Evaluation in Epidermolysis Bullosa – Brazilian Portuguese (individuals 17 years and over) were used.

Results: of the nine individuals with recessive dystrophic EB (88.9 % female and 12.91 (SD = 11.71) years), the mean total BEBS score was 24.47 (SD = 12.80) points on a scale of 0 to 100 points. Six participants had significant nutritional compromise according to the proposed criteria. Five of the six participants evaluated for HRQoL reported experiencing some impact, with individuals aged 17 and over being more affected and with greater clinical severity.

Conclusions: individuals with greater clinical severity of EB experience a more significant impact on their HRQoL. The proposed quantitative criteria for assessing nutritional compromise may help standardise assessments by professionals monitoring the nutritional status of individuals with EB.

Keywords:

Quality of life. Epidermolysis bullosa. Body mass index.

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Resumen

Introducción y objetivos: la epidermólisis bullosa (EB) es una rara enfermedad genética caracterizada por fragilidad de la piel con ampollas y erosiones. Las personas con EB experimentan manifestaciones extracutáneas y complicaciones clínicas y de calidad de vida relacionada con la salud (CVRS). Evaluamos la CVRS y la gravedad clínica y proponemos un criterio objetivo para estimar el deterioro nutricional con la herramienta *Birmingham Epidermolysis Bullosa Severity Score* (BEBS).

Métodos: esta serie de casos incluyó pacientes con EB monitoreadas por un equipo multiprofesional. Se evaluó la gravedad clínica con el BEBS utilizando rangos de índice de masa corporal por edad. Para evaluar la CVRS se utilizaron el *Children's Dermatology Life Quality Index* (individuos de 4 a 16 años) y el *Quality of Life Evaluation in Epidermolysis Bullosa – Brazilian Portuguese* (individuos de 17 años y más).

Resultados: de los nueve individuos con EB distrófica recesiva (88,9 % mujeres y 12,91 (DE = 11,71) años), la puntuación total media del BEBS fue de 24,47 (DE = 12,80) puntos en una escala de 0 a 100 puntos. Seis participantes tenían un deterioro nutricional significativo según los criterios propuestos. Cinco de los seis participantes evaluados en la CVRS informaron experimentar algún impacto, siendo los individuos de 17 años y más los más afectados y con mayor gravedad clínica.

Conclusiones: los pacientes con mayor gravedad clínica experimentan un impacto más significativo en su CVRS. Los criterios cuantitativos propuestos para evaluar el deterioro nutricional pueden ayudar a estandarizar las evaluaciones de los profesionales que monitorean el estado nutricional de las personas con EB.

Palabras clave:

Calidad de vida.
Epidermólisis bullosa.
Índice de masa corporal.

INTRODUCTION

Epidermolysis bullosa (EB) is a rare genetic disease characterised by the formation of blisters and erosions on the skin and/or mucous membranes, commonly caused by minor mechanical trauma due to mutations in the genes that are responsible for coding critical proteins for intraepidermal cell-cell adhesion and dermal-epidermal junction (1-3). The disease can present several extracutaneous manifestations and clinical complications, such as pain, cardiomyopathy, osteoporosis, infections, and anaemia, making regular follow-ups by a multi-professional team necessary (4-8). In this context, the diversity of clinical manifestations and complications of this disease leads to the compromise of nutritional status and health-related quality of life (HRQoL) (7,9,10).

The severity of EB may be related to individual, socioeconomic, and environmental factors (2) and, the more severe its manifestation, the greater the chance of serious secondary complications and nutritional compromise for the individual (11-13). The 'Birmingham Epidermolysis Bullosa Severity Score' (BEBS) is a tool that evaluates clinical manifestations and the impact of the disease on an individual's life. It identifies factors that contribute to the suffering of affected people. However, among the various items that make up the tool, 'nutritional compromise' is the only factor that does not present an objective criterion to be evaluated, scored as 0 (normal) or 5 (cachexia) points, based on the observer experience (14).

This study aimed to evaluate the HRQoL and clinical severity of a series of individuals with EB, followed by a multi-professional team. Considering that depending on the EB type, the individuals may present different levels of nutritional compromise (11-13), it is, therefore, difficult to determine, in a standardised way, nutritional status without numerical data. This study proposes a categorised criterion for the assessment of nutritional compromise for the BEBS, based on the body mass index (BMI) by age group.

MATERIALS AND METHODS

STUDY DESIGN

This series of cases is observational, cross-sectional, and descriptive.

PARTICIPANTS

The recruitment was regardless of sex, age, and physical condition, with all the individuals with recessive dystrophic EB (RDEB), monitored by a multi-professional team specializing in care for people with EB in Federal District - Brazil. Clinical severity and HRQoL were assessed and data on sociodemographic, nutritional, and clinical profiles were gathered. Individuals under 4 years were excluded from HRQoL assessment because of the lack of an adequate tool for this age group. However, they participated in the other analyses (Fig. 1).

ETHICAL ASPECTS

The study was approved by the Human Research Ethics Committee of the University of Brasilia (protocol no. 2.870.738). All individuals of legal age or legal guardians of minors signed the Free and Informed Consent Form, besides the authorisation term for image use.

DATA COLLECTION

Sociodemographic and clinical profiles

Date of birth, sex, EB type, diagnosis method, presence of daily care due to the disease, and nutritional supplements consumption were collected using a sociodemographic, nutritional, and clinical profiles questionnaire.

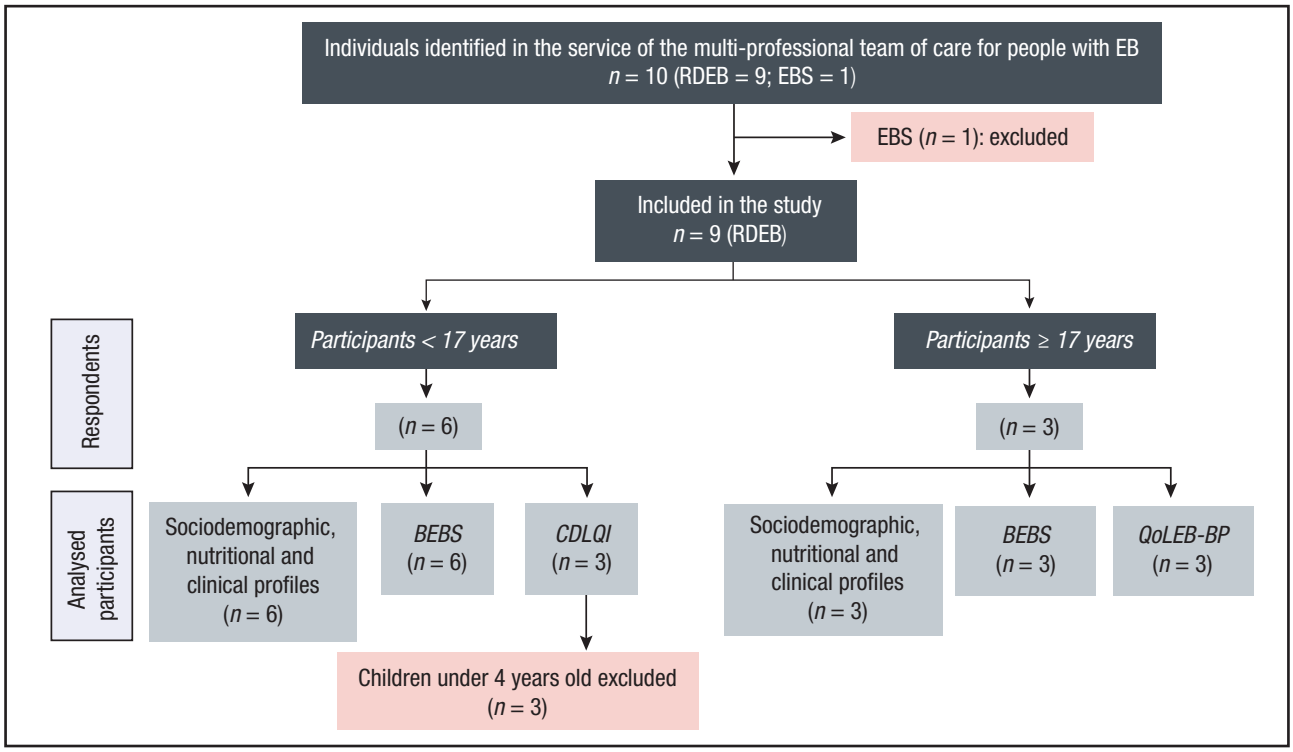

Figure 1.

Flow diagram of participants during the study (EB: epidermolysis bullosa; RDEB: recessive dystrophic epidermolysis bullosa; EBS: epidermolysis bullosa simplex; BEBS: Birmingham Epidermolysis Bullosa Severity Score; CDLQI: Children's Dermatology Life Quality Index; QoLEB-BP: Quality of Life Evaluation in Epidermolysis Bullosa – Brazilian Portuguese).

Anthropometric assessment

Anthropometry was performed by a trained team. In children under 2 years, weight was measured on a digital paediatric scale, without removing the bandages, in their guardian's presence. For height, an infantometer was used, with the child lying on a horizontal, firm, and flat surface measured from the soles of the feet to the top of the head (15). In volunteers aged ≥ 2 years, weight was measured on a digital platform scale, without removing the bandages. A vertical anthropometer was used to measure the height (15).

Clinical severity

The BEBS was used to assess this variable. Due to the lack of a validated version for Brazilian Portuguese, first, a free translation to Portuguese of the versions for both children and adults was carried out.

The tool had 11 items to assess clinical severity in EB, where, in each item, 0 point suggest a lower complication and the maximum of 5 points a higher complication (or 50 points for the area of lesions): 1. nails (maximum 5 points); 2. area of lesions (maximum 50 points); 3. mouth (maximum 5 points); 4. eyes (maximum 5 points); 5. larynx (maximum 5 points); 6. oesophagus (maximum 5 points); 7. scarring of hands (maximum 5 points); 8. skin cancer (maximum 5 points); 9. chronic wounds (maximum 5 points); 10. alopecia (maximum 5 points), and 11. nutritional compromise (maximum 5 points). The total BEBS score ranges from 0–100, according to each of the categories considered for each item. There are no cut-off points to classify the final score; however, the authors suggest that the higher the score, the greater the individual's level of clinical impairment, and consequently, a more affected HRQoL (14).

Regarding the nutritional compromise item, this presents a subjective evaluation as it establishes a score from 0 to 5, wherein a score of 0 should be attributed to the individual with normal nutritional status and 5 to the cachectic individual, regardless of whether they are a child or an adult (14). As a more quantitative criterion for scoring the item, BMI was used according to age group to objectively discriminate its score. For children and adolescents, the BMI-for-age curves from the World Health Organization (WHO) 2006 and 2007 were used (16,17), and for adults, the WHO BMI classification (1995) was used (15) (Table I). Thus, the BEBS score for the item was categorised into 0, 3, and 5 points for children and adolescents, while for adults, the scores of 0 and 5 points remained, but with a quantitative criterion, as adapted, aiming to use the measures recommended by the WHO. To better classify, according to WHO criteria, the term cachexia of the original tool was changed to severe thinness for children and underweight for adults.

Table I. The score of the nutritional compromise item of the Birmingham Epidermolysis Bullosa Severity Score (14), for children, adolescents, and adults, according to the body mass index (BMI) classification (15-17)

Classification for children and adolescents	BMI (Z-score)	Item score
Normal	≥ Z-score -2 and ≤ Z-score +1	Score 0
Thinness	≥ Z-score -3 and ≤ Z-score -2	Score 3
Severe thinness	< Z-score -3	Score 5
Classification for adults	**BMI (kg/m²)**	**Item score**
Normal	≥ 18.5 and < 25	Score 0
Underweight	< 18.5	Score 5

HRQoL

People aged 4-16 years

To assess HRQoL, the Children's Dermatology Life Quality Index (CDLQI) was used, which assesses individuals aged 4-16 years. This questionnaire was validated for Brazilian Portuguese (18,19), with ten questions about symptoms, leisure, school or holidays, personal relationships, sleep, and skin care in the previous week. Each question has four response options and is scored from 0 to 3 points, with a total of 30 points. The higher the score, the more affected the individual's HRQoL (20). The cut-off points for the interpretation of the final score were: 0-1 points, no effect; 2-6 points, small effect; 7-12 points, moderate effect; 13-18 points, very large effect and; 19-30 points, extremely large effect (21).

People aged ≥ 17 years

To assess HRQoL in this age group, the Quality of Life Evaluation in Epidermolysis Bullosa – Brazilian Portuguese (QoLEB – BP) questionnaire was used, which is translated, culturally adapted, and validated for Brazil (22), from the original 'The Quality of Life Evaluation in Epidermolysis Bullosa (QOLEB)' (23). This was designed for different clinical EB types, with the potential to gather information for therapy and research purposes, without proposing a specific version for children. It is composed of 17 questions that assess the functional and emotional aspects of people with EB. Each question has four response options, with scores ranging from 0 to 3 points and a maximum of 51 points (23). The cut-off points for interpretation of the total score were: 0-4 points, very mild impact; 5-9 points, mild impact; 10-19 points, moderate impact; 20-34 points, severe impact and; 35-51 points, very severe impact (24).

DATA ANALYSIS

The variables were age, sex, diagnosis method, presence of special daily care, weight, height, BMI, nutritional status, use of nutritional supplements, and scores for each BEBS form item. HRQoL data were according to the items of each questionnaire (CDLQI and QoLEB-BP), with the scores obtained and respective classifications.

The statistical software Stata® (version 16.1) (25) was used to perform the Shapiro-Wilk normality test on quantitative variables, presented as mean and standard deviation, while categorical variables were presented as percentages and 95 % confidence intervals.

RESULTS

SOCIODEMOGRAPHIC, CLINICAL AND NUTRITIONAL PROFILES

The sample consisted of nine participants, with a mean age of 12.91 years (SD = 11.71) and predominantly female (88.9 %). All had special daily care due to EB, such as changing dressings, managing hygiene and perforating blisters, and wearing comfortable clothes/shoes. Further, 66.7 % used some nutritional supplements (Table II).

CLINICAL SEVERITY

The mean total score of the participants on the BEBS was 24.47 points (SD = 12.80). The area of lesions contributed the most to the clinical severity score, indicating damaged skin with blisters, erosions, crusts, skin healing, erythema, and atrophic scarring. However, nails, mouth, scarring hands (Fig. 2), and nutritional compromise also made important contributions to the clinical severity score. The laryngeal and skin cancer items contributed the least (Table III).

Table IV presents the individual results of clinical severity according to the proposed BMI classification for assessing nutritional compromise and the BEBS scores. Of the nine individuals, three had normal BMI. The two individuals with the highest BEBS scores were adults and classified as underweight by BMI. In the age group of up to 18 years, individuals 1 and 5 had the greatest nutritional compromise.

Table II. Sociodemographic, clinical, and nutritional data of individuals with recessive dystrophic epidermolysis bullosa assisted by a multi-professional team specialising in care for people with epidermolysis bullosa in the Federal District — Brazil (*n* = 9)

Variables	Total sample (*n* = 9) (% and 95 % CI)
Sex (female)	88.9 % (40.94; 98.93)
Diagnostic method	
Exclusively clinical	22.2 % (4.30; 64.48)
Clinical + Biopsy†	44.4 % (14.55; 78.98)
Clinical + Molecular genetic examination	33.3 % (8.92; 71.86)
Use of nutritional supplement	66.7 % (28.14; 91.08)
Daily special care	100 %

CI: confidence interval. †Individuals who checked this item did not know which analysis method would be used later, but there is an extra method used after the biopsy.

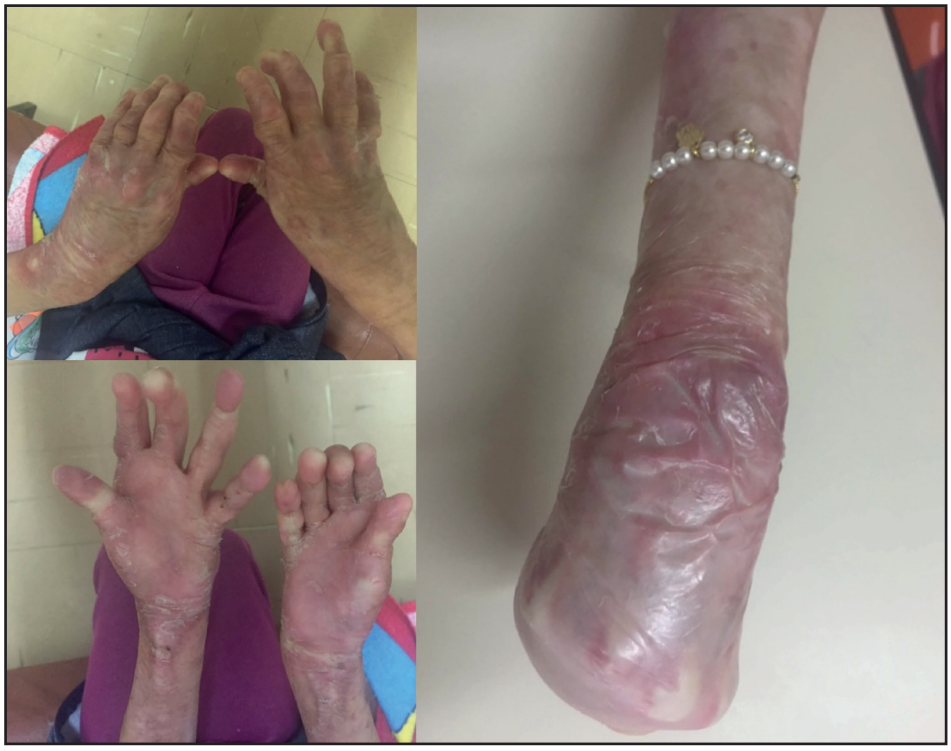

Figure 2.
Contractures and pseudosyndactyly in the hands of individuals monitored at the service of a multi-professional team for the care of people with epidermolysis bullosa at the Federal District – Brazil.

HRQoL

The results were described according to each used tool, as follows:

CDLQI

Participants 4, 8, and 9 did not have their HRQoL evaluated because they were younger than 4 years old. Participant 5 suffered no effect on HRQoL, while participant 6 presented a moderate effect, and participant 7 experienced a small effect. No participant reported embarrassment, annoyance, or sadness due to EB; no friendships, school activities, or vacations were affected; they were not intimidated by people staring or asking questions, and their sleep was not affected. As a result, their HRQoL was more affected in terms of physical aspects than emotional aspects.

The impact on HRQoL was compatible with the clinical severity score obtained in the BEBS as participant 5 suffered no effect on HRQoL, consistent with the lowest clinical severity score among the three participants; participant 6 experienced a moderate im-

Table III. Score on each of the items that make up the clinical severity assessed by the Birmingham Epidermolysis Bullosa Severity Score (14) of participants with recessive dystrophic epidermolysis bullosa assisted by a multi-professional team specialising in care for people with epidermolysis bullosa in Federal District — Brazil (*n* = 9)

Participant Items/score	1	2	3	4	5	6	7	8	9
Nails (0-5 points)	5	5	5	5	5	5	5	4.5	2.5
Area of lesions (0-50 points)	9.5	15.37	13.5	0.25	0.75	6	4.5	2.37	7
Mouth (0-5 points)	4	5	5	1	1	3	4	3	1
Eyes (0-5 points)	0	4	1	1	0	1	0	0	0
Larynx (0-5 points)	0	0	0	0	0	0	0	0	1
Oesophagus (0-5 points)	0	4	5	1	1	1	0	0	1
Scarring hands (0-5 points)	5	5	5	0	3	3	4	1	1
Skin cancer (0-5 points)	0	0	1	0	0	0	0	0	0
Chronic wounds (0-5 points)	1	2	1	0	1	3	2	3	2
Alopecia (0-5 points)	0	0	3	0	0	0	2	0	0
Nutritional compromise (0-5 points)	5	5	5	0	5	3	0	3	0
Total (0-100 points)	26.5	45.37	44.5	8.25	16.75	25	21.5	16.87	15.5

Table IV. Nutritional status according to body mass index (BMI) (15-17) and assessment of individual clinical severity by Birmingham Epidermolysis Bullosa Severity Score (BEBS) (14) of individuals with recessive dystrophic epidermolysis bullosa assisted by a multi-professional team specialising in care for people with epidermolysis bullosa in Federal District — Brazil (*n* = 9)

Participant	BMI (kg/m²)	Z-score	Nutritional status	BEBS score
1	12.75	< -3	Severe thinness	26.5
2	12.73	–	Underweight†	45.37
3	16.97	–	Underweight†	44.5
4	14.73	> -1 and < 0	Normal	8.25
5	10.94	< -3	Severe thinness	16.75
6	15.55	< -2 and > -3	Thinness	25
7	13.47	> -2 and < 0	Normal	21.5
8	13.69	> -3 and < -2	Thinness	16.87
9	15.12	> -1 and < 0	Normal	15.5

†*Classification of body mass index according to adult age group.*

Table V. Assessment of health-related quality of life (HRQoL) by Children's Dermatology Life Quality Index (CDLQI) (20) and Quality of Life Evaluation in Epidermolysis Bullosa – Brazilian Portuguese (QoLEB-BP) (22) and clinical severity score by Birmingham Epidermolysis Bullosa Severity Score (BEBS) (14) (n = 6)

Participant	CDLQI			QoLEB-BP		
	HRQoL score	Classification of impact on HRQoL	BEBS score	HRQoL score	Classification of impact on HRQoL	BEBS score
1	–	–	–	16	Moderate	26.5
2	–	–	–	29	Severe	45.37
3	–	–	–	20	Severe	44.5
5	1	No effect	16.75	–	–	–
6	8	Moderate effect	25	–	–	–
7	2	Small effect	21.5	–	–	–

pact, related to the higher score in clinical severity; and participant 7 underwent a small effect on HRQoL, compatible with the intermediate score between the three (Table V).

QoLEB – BP

The three participants aged ≥ 17 years had their HRQoL moderately or severely affected by EB, reporting a greater impact on their ability to shower and practice sports. Participants 2 and 3 experienced a severe impact on their HRQoL, which is consistent with what was found in their BEBS clinical severity assessments, considering that participant 2 had the highest clinical severity score and participant 3 presented the second-highest. In turn, participant 1 experienced a moderate impact on HRQoL, consistent with his lower clinical severity score compared to participants 2 and 3 (Table V).

DISCUSSION

In this series of cases, adults showed greater clinical severity than children and adolescents, and the area of lesions strongly contributed to this outcome, accompanied by manifestations in the nails, mouth, hands, and nutritional compromise. All participants reported daily special care for EB, which is challenging because it causes pain, is time-consuming, and wears out individuals with the disease and their caregivers, constituting one of the greatest concerns in EB (26,27). The area of lesions was also identified as having the highest weight by Moss et al. (2009) (14), since individuals with a larger area are more likely to experience severe pain, infections, malignancy in the lesions, nutritional compromise, and even early death. Therefore, effective treatment for EB should aim to reduce damaged skin (14).

Regarding nutritional status, more severely affected people with EB have nutritional compromise due to limiting factors in food consumption and hypermetabolism, mainly associated with open skin lesions, leading to greater protein turnover and heat loss (10,28), which is consistent with the nutritional compromise seen in most of the sample. However, according to the BEBS score, the most severely affected individuals were not the only ones to have nutritional compromise, confirming the difficulty in maintaining a good nutritional status in people with EB (29). Therefore, the Health Department of the Federal District - Brazil dispenses nutritional supplements for individuals with EB through the Home Enteral Nutritional Program, which aims to contribute to de-hospitalizations, subsidising Home Care Programs regarding food, reducing the number of readmissions for reasons related to nutritional status, improving HRQoL, and promoting the maintenance or recovery of nutritional status (30).

Considering that malnutrition leads to failure to thrive, delayed puberty, anaemia, and a cascade of clinical and biological events (31-33), it is important that the staging of nutritional compromise be carried out in an objective and standardised way to assess clinical severity. It is noteworthy that BEBS was prepared by professionals with experience, who were trained to monitor people with EB at a reference centre in England. These teams are used to seeing a large number of patients with the disease. Even so, Moss et al. (2009) reported difficulties in determining the nutritional compromise score precisely because of differences by age group (14). Filling out this topic may be impaired when applying this to services that serve a smaller number of individuals and are not characterised as reference centres. Depending on the evaluator's degree of training and the individual's EB type, the identification of nutritional compromise is difficult and not standardised (11-13).

Considering the clinical condition of this population, skinfold thicknesses and perimeters are not recommended, since the instruments used for these measurements would injure the skin

of these individuals (34). However, BMI use would be indicated, once it does not cause injuries to the individual skin and its purpose is to facilitate and standardise the nutritional status evaluation for the clinical severity. For this reason, the proposed criteria for assessing nutritional compromise, as recommended by the WHO for children, adolescents, and adults, prove to be very useful as a facilitator of this assessment (15-17).

Regarding the tools used to assess HRQoL in people with EB, Frew et al. (2009) (23), responsible for preparing the QoLEB, stated the need for a tool to assess the HRQoL of children (23). However, there was still no tool for this specific public at the time of research collection. For this reason, the CDLQI was adopted to assess the HRQoL of children and adolescents, which, despite not being specific to the population with EB, is used for dermatological diseases and considers the important aspects of HRQoL in this age group. Although it is important to emphasise that, currently, the 'EB-specific module of the Infants and Toddlers Dermatology Quality of Life (InToDermQoL-EB)' questionnaire was prepared, which underwent initial validation and will be specific for the evaluation of children with EB under 4 years old (35).

Concerning the assessment of HRQoL, it was noticed that individuals younger than 17 years were more affected in physical than emotional aspects, possibly because children who manage to accept the disease or distance themselves from it seem to feel better compared to those who tend to engage in cognitive palliative strategies or express emotional reactions, developing high levels of resilience (36). Individuals aged ≥ 17 years were more affected in their ability to shower and practice sports, as observed in a systematic review on the quality of life of people with EB, in which the difficulties faced by people with EB in these two activities were highlighted (9). In a study carried out in Brazil, it was observed that, concerning the QoLEB-BP assessment scales, individuals were more affected in the functional than in the emotional domain (22), again, a finding possibly explained by the high levels of resilience developed by this severely affected population (22,37,38).

Regarding the relationship between HRQoL and clinical severity, the clinical manifestations of EB vary from mild to fatal, and the more severe the manifestation, the greater the chance of secondary complications (11-13). According to a study by Yazdanshenas et al. (2020) (38), a high positive correlation was found between the assessments of the QoLEB and BEBS questionnaires, indicating that individuals with EB who have greater severity of the disease have a lower HRQoL (38). This is consistent with our findings, in which individuals with greater clinical severity experience a greater impact on their HRQoL, similar to the finding of Danescu et al. (2019) (39), who compared the HRQoL and clinical severity of people with EB in Romania using the QoLEB and the EB Disease Activity and Scarring Index (EBDASI), concluding that people with EB who have a greater perception of clinical severity have a lower HRQoL (39).

The strength of this study is its pioneering approach to assessing the clinical severity of a group with EB in Brazil using the BEBS and assessing HRQoL to verify the relationship between these two aspects. Additionally, objective criteria were proposed for the assessment of nutritional compromise in BEBS, according to the BMI by age group, as recommended by the WHO. However, there are limitations: the design of the case series study, which makes it impossible to extrapolate its results, and the limited number of participants for analysing the HRQoL, due to the lack of an evaluation questionnaire for children under 4 years of age at the time of the research.

Individuals with greater clinical severity presented a greater impact on their HRQoL. The proposed quantitative criterion for assessing nutritional compromise can facilitate and standardise assessments by professionals who monitor the nutritional status of individuals with EB, as the nutritional compromise, together with the area of lesions, nails, mouth and scarring of hands was one of the items that contributed the most to clinical severity scores. The tools used in this study should be included in the continuous monitoring of patients with EB to establish the best care strategies.

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