

Review

The impact of epidermolysis bullosa on the family and healthcare practitioners: a scoping review

Antoinette V Chateau^{1,2}, BSc, MBCHB, DCH, FC Derm (SA), MMedSci, 
David Blackbeard^{3,4}, MA Clin Psych, PhD and Colleen Aldous⁵, PhD

¹Department of Dermatology, Grey's Hospital, Pietermaritzburg, South Africa,
²University of KwaZulu-Natal, School of Clinical Medicine, Durban, South Africa,
³Department of Clinical Psychology, Grey's Hospital, Pietermaritzburg, South Africa,
⁴Department of Psychiatry, School of Clinical Medicine, University of KwaZulu-Natal, Durban, South Africa, ⁵University of KwaZulu-Natal, School of Clinical Medicine, Durban, South Africa

Correspondence

Antoinette V Chateau, BSc, MBCHB, DCH,
FC DERM SA, MMEDSCI,
Department of Dermatology
Greys Hospital
Pietermaritzburg
South Africa
E-mail: chateauav@gmail.com

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Introduction

Epidermolysis bullosa (EB) is a genodermatosis resulting in mucocutaneous fragility classified as a rare disease, affecting less than 1:2000 with an incidence and prevalence of 19.57 and 11.07 per 1 million live births, respectively.^{1,2} It is a heterogeneous group of disorders that are classified into four major groups with over 30 subtypes: simplex, junctional, dystrophic, and Kindler syndrome based on the defective protein at the dermal-epidermal junction. To date, 21 genes have been identified, with over 1000 mutations noted.³

The clinical presentation and complications vary from very limited vesicles or nail involvement to extensive disease resulting in skeletal deformity, systemic manifestations, and malignancy. Life expectancy varies depending on the subtype.

The focus of research has been around diagnostics and therapeutics with very little regarding the psychosocial well-being of the patient, their caregivers, and family as a component of holistic care. Both qualitative and quantitative studies have

Abstract

Background Epidermolysis bullosa (EB) is an inherited genodermatosis that results in mucocutaneous fragility. There is a lack of data on the impact of this disease on parents. There are no studies on the impact on siblings and few on healthcare professionals in dealing with this devastating disease.

Methods A scoping review was performed using the Arksey and O'Malley and PRISMA-ScR framework. Twenty-seven articles were reviewed, and a data-charting sheet was formulated.

Results Parents make great sacrifices and are resilient in caring for their sick children but are at risk of depression. Siblings play a vital role in caring for their siblings, but their needs may be overlooked because the main focus is on the sibling with EB. Healthcare professionals may suffer burnout and compassion fatigue in caring for patients and their families with EB.

Conclusion Comprehensive care of the family and the awareness of the challenges experienced by healthcare professionals is essential to the holistic care of a patient with EB.

investigated the quality of life of patients with EB and the burden of care of parents/caregivers and families caring for a patient with EB. Very few studies concentrate on the impact of EB on healthcare professionals caring for these families, and none evaluate the siblings of a child with EB.

In summary, EB is a rare genodermatosis, with a shortage of studies focusing on the family and healthcare professionals caring for these patients and their families. A better understanding of the impact on and needs of those close to the patient is essential for comprehensive care of the patient. Therefore, this scoping review aims to highlight the impact of EB on parents, siblings, and healthcare professionals caring for affected families and to determine the needs of the family in the context of EB.

Review question/objective

The objective of this scoping review is to review the global literature with regard to:

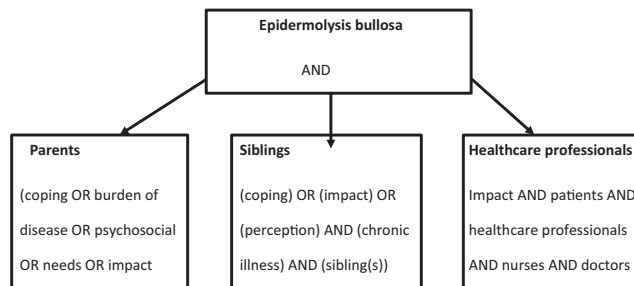


Figure 1 PubMed search

- 1 The psychological impact on parents or caregivers caring for children with EB
- 2 The impact of EB on the siblings of patients with EB
- 3 The impact on healthcare professionals (HCP) caring for patients with EB

Methodology

The scoping review was performed using Arksey and O'Malley⁴ and PRISMA-ScR framework.⁵ The studies included in this study were peer-reviewed qualitative, quantitative, mixed methods studies and a case report published between 1986 and 2020. Databases searched included PubMed; EBSCO host including Academic Search Complete, APA PsycArticles, APA PsycInfo, CINAHL, MEDLINE; Clinical Key; Science Direct; and Web of Science. The inclusion criteria were English full-text, peer-reviewed articles in humans, focused globally, articles in a foreign language, and animal studies were excluded.

Two reviewers, AC and CA, screened the articles found independently between January 4, 2021, and January 27, 2021, and the screening was verified by a third reviewer, DB. The last search was exported into the EndNote reference manager.

A complete search strategy for the PUBMED database is noted in Figure 1.

Boolean phrases with AND, OR, NOT, and parentheses were used with the following terms: Epidermolysis bullosa, coping, impact, the burden of disease, psychological, caregivers, parents, family, siblings, healthcare professionals, nurses, and doctors. A data-charting sheet was developed and included the author, year, country of origin, study design, the aim of the study, and key findings (Table 1).

Extracting and charting results

A total of 171 articles were identified, with 110 excluded as they were not relevant to the topic, and 30 were duplicated, resulting in 31 full-text articles being assessed for eligibility. Four further articles were excluded as they were not in English, and two could not be retrieved. Twenty-seven peer-reviewed articles were included in the study, which included qualitative, quantitative, mixed-methods studies, and a case report (Fig. 2).

Measuring tools for quality of LIFE and burden of disease studies

Over the years, significant progress has been made in developing assessment tools to determine the quality of life (QOL) of patients with EB; before this, generic QOL questionnaires were adapted for these patients.^{6,7} Frew et al. formulated and validated the first tool to assess the QOL of patients with EB through the development of the QOLEB questionnaire.⁸ Participants included are patients with EB, parents, siblings, and healthcare workers. Interviews were conducted, and the development of a questionnaire was subsequently refined and then validated for reliability. This QOL measurement tool took into account the various subtypes of EB and the different age groups of patients. As a consequence, the application of the QOLEB demonstrated that anxiety and depression responses were similar in patients with mild and severe diseases. This might have been because the presence of the disease was more related to depression and anxiety than its severity.

Developed and validated by Dufresne et al., the EB family/parental burden score determines disability by taking into account the psychological, social, physical, and economic aspects of the family. This burden of disease (BOD) score showed a significant difference between parents of children with a mild form, EB simplex, and those with the extensive form, recessive dystrophic EB. Complications were noted more in children older than 7 years of age.⁹ This observation is opposed to the view by Tabolli et al., who noted family burden due to epidermolysis bullosa is very high independent of disease type/subtype.¹⁰ Although both QOLEB and BOD are very important tools in EB studies, Frew and colleagues purported that the QOLEB questionnaire needs to be adapted and validated within different countries concerning the cultural setting, language, and other psychosocial aspects;⁸ and Dufresne et al. also highlighted that cross-cultural validation is important for international use.⁹

Impact of EB on the parents

There is a paucity of studies that focus on the impact of EB on the parents. Having a child with a potentially life-limiting disease, depending on the subtype, can be devastating for the

Table 1 Summary of identified studies in the literature

Author	Title	Year	Country	Study design	Aim	Findings
Norup M	Treatment of severely diseased newborns; attitudes among Danish physicians	1999	Denmark	Questionnaires O + G and Pediatrics	To determine the attitudes among healthcare providers toward the treatment of severely diseased neonates with EB, myelomeningocele, trisomy 13, and Down syndrome	<ul style="list-style-type: none"> a. Majority of physicians stated that they would refrain from life-prolonging treatment b. 52–60% of physicians would provide morphine in life-shortening doses c. Religious convictions played a role concerning infanticide in some physicians
Dures E	"You're whatever the patient needs at the time": The impact on the health and social care professionals of supporting people with epidermolysis bullosa	2010	United Kingdom	Two-part qualitative mixed methods: Interviews nurses and social care professionals Survey to EB specialists	Professionals' perception: <ul style="list-style-type: none"> - Ways in which their work impacts their well-being - Resources that may enhance their well-being 	Intensity and depth of involvement <ul style="list-style-type: none"> - Emotionally draining - Feelings of powerlessness - Guilt, demoralization, sorrow Limited resources <ul style="list-style-type: none"> - Reluctance of healthcare services and social services to become involved - nonspecialists not appreciating the complexity of the disease - Frustration at not having adequate resources - Need to look after yourself - Self-care - Support groups - Different patient-physician interactions - Mutual collaboration between patient and physician improves medical outcomes, improves compliance, and decreases medical expenses - Patients want to be empowered about their disease - Need to upscale doctors not familiar with EB - Parents' perception of how siblings coped. - Siblings were well adjusted - Supportive - Protective and helpful caregiver role toward their sibling - Emotionally supportive and provided physical comfort - Kept their sibling in touch with the outside world with the aid of technology- Facebook - Fundraising for medical bills Coping with the sibling's disease: <ul style="list-style-type: none"> - Sadness, helplessness, fear that they may get the disease Family restructuring: <ul style="list-style-type: none"> - Change in daily routine—caregivers are absent while caring for the sick sibling Experiences resulting from the sibling's hospitalization:
Budycha K	How do patients with rare diseases experience the medical encounter? Exploring role behavior and its impact on patient-physician interaction	2012	Germany	Interpretive, qualitative study, Semi-standardized interviews with patients with rare diseases	To explore experiences of patients with rare diseases in the interaction with their physician	<ul style="list-style-type: none"> - Support groups - Different patient-physician interactions - Mutual collaboration between patient and physician improves medical outcomes, improves compliance, and decreases medical expenses - Patients want to be empowered about their disease - Need to upscale doctors not familiar with EB - Parents' perception of how siblings coped. - Siblings were well adjusted - Supportive - Protective and helpful caregiver role toward their sibling - Emotionally supportive and provided physical comfort - Kept their sibling in touch with the outside world with the aid of technology- Facebook - Fundraising for medical bills
Nabors L	Family coping during hospitalization of children with chronic illnesses	2017	USA	Mixed methods Part 1: F-COPES Family Crisis-Oriented Personal Evaluation Scales Part 2: Interviews	To assess family resilience and coping during hospitalization of a child with a chronic illness	Coping with the sibling's disease: <ul style="list-style-type: none"> - Sadness, helplessness, fear that they may get the disease Family restructuring: <ul style="list-style-type: none"> - Change in daily routine—caregivers are absent while caring for the sick sibling Experiences resulting from the sibling's hospitalization:
Hilkner S	Perceptions of siblings of children hospitalized due to chronic illness	2019	São Paulo, Brazil	Qualitative narrative design	Perceptions of siblings of children hospitalized due to a chronic illness	Coping with the sibling's disease: <ul style="list-style-type: none"> - Sadness, helplessness, fear that they may get the disease Family restructuring: <ul style="list-style-type: none"> - Change in daily routine—caregivers are absent while caring for the sick sibling Experiences resulting from the sibling's hospitalization:

Table 1 Continued

Author	Title	Year	Country	Study design	Aim	Findings
Plumbridge G	Parents' communication with siblings of children affected by an inherited genetic condition	2011	United Kingdom	Semi-structured interviews	Parent communication about the risks of an inherited congenital condition on the siblings and to determine the level of support needed from healthcare professionals	<ul style="list-style-type: none"> - Fear of the equipment that their siblings were connected to - Church and prayer are a source of comfort - Siblings involvement in care is beneficial for the whole family and equips the family in the case of loss - Joy of being included in the care of their sibling <p>Exclusion experiences</p> <ul style="list-style-type: none"> - Siblings are invisible among family and those around the sick sibling - Condition is not explained to the sibling - Miss their sibling—no relationship - Communicate with parents on the phone - Cared for by grandparents - Social events excluded from certain events because their mother was at the hospital (Mother's Day) - Surprised at being asked to be interviewed <p><i>Parents talking to siblings about the genetic condition</i></p> <ul style="list-style-type: none"> - Children wanted to find out - Parents assumed that the children knew the condition - Some parents were reluctant to discuss the genetic - Parents did not reassure unaffected siblings who were not at risk - Few siblings did not want to discuss the risks - Some parents discussed the condition more with the siblings than the affected child. Siblings were thus entrusted to keep secrets from their siblings <p><i>Understanding of genetic risk and information given by the parent:</i></p> <ul style="list-style-type: none"> - Siblings found out about the heredity when they were tested or involved in sexual relationships - Parents felt that they did not need to know until they were likely to have sexual relationships - Siblings confronted parents when they gathered information from other sources outside the family such as the internet, school, or informational leaflets - Siblings did not discuss the genetic condition or the potential risks with each other <p><i>Emotional and behavioral outcomes for siblings:</i></p> <ul style="list-style-type: none"> - Parents did not expect siblings to help care for sibling - Sibling was willing to help and proud to do so - Some found their sibling's condition embarrassing - Cognoscente of the financial burden and time spent on their sibling, time away from family activities

Table 1 Continued

Author	Title	Year	Country	Study design	Aim	Findings
De Stefano F	Family burden of children suffering from epidermolysis bullosa	2015	Italy	Cohort study: Questionnaires	To determine the family burden of children affected with epidermolysis bullosa	<ul style="list-style-type: none"> - Where genetic conditions were not discussed, siblings showed resentment or withdrew from the affected child - When discussed, displayed understanding for their siblings and better understood their role in the family - Unaffected children worried about their siblings, feeling guilt that they were free of the disease - Some were reluctant to discuss the possibility of them or their future children having the disease with their parents as not to upset them - Some would have liked if their parents discussed the condition and reassured them <p><i>Importance of family communication for siblings:</i></p> <ul style="list-style-type: none"> - Support within the family was essential as many families felt that there was limited support for siblings from healthcare and social care services - Parents aware of the lack of time spent with the unaffected child <p>The entire family is at risk psychologically, socially, and financially independent of the type of EB</p>
Fine J	Impact of inherited EB on parental interpersonal relationship, marital status and family size	2005	USA	Standardized questionnaires	To determine the impact of the presence of EB in one or more children on the personal relationships between parents	<p>Relationship with their spouse was negatively impacted in severe forms, RDEB and junctional, not for EBS</p> <ul style="list-style-type: none"> - Lack of energy, interest, and time in anything else apart from caring for their child - Impact on the couple's sexual relationship - Impact on their choice to have more children - Risk of divorce, 22% of parents in this study were divorced (EBS did not attribute EB as a potential cause) - Financial burden did not play a major factor - Feelings of shock, anxiety, fear, and love. One mother described a feeling of rejection of her baby at birth - Variability in the information and emotional support they received at the birth of their children - Challenges faced included the appropriate clothing/shoes, dressings - Teachers in the junior school requested more information about the child's condition, some senior schoolteachers were less empathetic and understanding - Children's social life—some had problems making friends
Landsdown R	Practical and psychological problems for parents of children with epidermolysis bullosa	1986	London	Questionnaires and vignettes	The psychological and practical problems encountered by parents of children with EB	<ul style="list-style-type: none"> - Mothers usually gave up work to care for their children thus a loss of earnings - Increase in financial expenses
Duffresne H	Impact of a rare chronic genodermatosis on family daily life: The	2018	France	Semi-structured questionnaire	Impact of EB on the social dimension of the family	

Table 1 Continued

Author	Title	Year	Country	Study design	Aim	Findings
	example of epidermolysis bullosa					<p>Hospitalization resulted in the working parent taking time off of work to care for the home</p> <p>Siblings missed out on school life and extracurricular activities</p> <p>Social isolation</p> <p>Social support grants were insufficient</p> <p>Parents who had given up employment found it very difficult to enter the workforce</p> <p>EB patients and families are psychologically, socially, and financially at risk</p> <p>Multidisciplinary management and patient education programs are essential</p> <p>The study explores the mediation effect of the three coping strategies and self-esteem on the parenting stress and depressive symptom relationship in the family caring for a child with a genetic illness like EB</p> <p>Females had higher parenting stress using three coping strategies being problem focused, emotional focused, and dysfunctional coping, and more depressive symptoms. Stress had a negative correlation to self-esteem. Self-esteem had a negative correlation with dysfunctional coping and depressive symptoms</p> <p>Impact the family financially</p> <p>Time away from other children in the family</p> <p>Taking the child away from their known environment to adapt to a foreign environment and separation from the support structure could be detrimental to the development and quality of life of a child. Sometimes looking beyond the medical benefits and focusing on the quality of life even if it may result in a shorter life are difficult ethical decisions that need to be considered</p> <p>- This QOL measurement tool took into account the various subtypes of EB and the different age groups of patients</p> <p>- First questionnaire was developed that measured the QOL of patients with EB. It was designed to measure the impact EB has on a patient and has overcome the disadvantages of generic questionnaires</p> <p>- It is statistically reliable and valid.</p> <p>-The psychosocial aspects of QOLEB may require further investigation. This tool can be used to improve patient management or for monitoring the progress of a patient with EB</p>
Wen CC	Parenting stress and depressive symptoms in the family caregivers of children with genetic or rare diseases: The mediation effects of coping strategies and self-esteem	2019	Taiwan	Questionnaire: depression scale, self-esteem scale, brief COPE, pediatric inventory for parents	To determine how parent stress influences depressive symptoms in parents caring for a child with a rare disease by examining the mediators of the three coping strategies and self-esteem	
deSante-Bertkau J	When speciality care is unavailable to rural families	2019	USA	Case report	Ethical dilemma in treating a child with severe EB requiring a tracheostomy and weighing it up against the quality of life of the child and the family	
Frew J	Quality of life evaluation in epidermolysis bullosa through the development of QOLEB questionnaire: an EB-specific quality of life instrument	2009	Australia	Participants included patients with EB, parents, siblings, and healthcare workers. Interviews, development of a questionnaire that was subsequently refined and then validated for reliability	To develop a valid and reliable EB-specific QOL tool for measuring the effects of disease impact and interventions	

Table 1 Continued

Author	Title	Year	Country	Study design	Aim	Findings
Dufresne H	Development and validation of EB family/parental burden score	2015	France	Verbatim reports from parents followed by psychometric analysis. An EB-BOD questionnaire was then formulated and this was validated against the Short-Form 12-item questionnaire.	To develop and validate a questionnaire assessing the burden on families of children with EB: EB burden of disease	<p>The burden of disease in children is shared with the family</p> <p>The burden includes the financial cost of therapy, time away from work and lack of social support from family and friends, hours taken to do dressing changes</p> <ul style="list-style-type: none"> - Significant difference between parents of children with EBS compared to those with RDEB - Complications are noted more in children >7 years - EB-BOD takes into account the social and economic aspects of the patient and their family. It was suggested that cross-cultural validation is important for international use
Sampogna F	Measuring the impact of dermatological conditions on family and caregivers: a review of dermatology-specific instruments	2017	Italy	Systematic review	To identify the specific instruments that measure the impact of a dermatological disease on the quality of life of family members	<p>The Greater Patient, described by Basra and Findley, is a group of people who are close to the patient and whose lives are affected by the patient's disease. Patients are at the center of a family dynamic, and this needs to be taken into account when managing a patient</p> <p>Instruments that measure the impact of the disease on the family and the patient—take into account the physical, psychosocial, and economic impact of the disease</p> <p>Family Dermatology Life Quality Index (FDLQI) general, adverse effects of a disease on the family EB-BOD, the burden of EB on the family. Able to distinguish between the subtypes of EB</p> <p>Cultural aspects are usually not taken into account in these instruments and need to be considered when used in various countries</p>
Sampogna F	The evaluation of family impact of recessive dystrophic epidermolysis bullosa using the Italian version of the Family Dermatology Life Quality Index	2012	Italy	Family Strain Questionnaire and the FDLQI was completed	To determine the burden of recessive dystrophic EB on the family caregivers using the Italian version of the FDLQI and to validate the questionnaire	<ul style="list-style-type: none"> - Time spent - Increased household expenditure - Affected physical well-being - Emotional distress <p>Higher impact on mothers</p> <ul style="list-style-type: none"> - impact proportional to the extent of the disease - Parents felt that the information given by the medical staff was not sufficient and practical - Parents with a child with EB had a high burden of care, this was lower in parents who had assistance
Kahraman S	Determination of caregiving burden of parents providing care to their children with epidermolysis bullosa	2017	Egypt	Descriptive study. Questionnaires and Zarit caregiving burden scale	To determine the caring burden of parents with children with epidermolysis bullosa and its affecting factors	

Table 1 Continued

Author	Title	Year	Country	Study design	Aim	Findings
Van Scheppingen C	The main problems of parents of a child with epidermolysis bullosa	2008	Netherlands	Semi-structured interviews	To determine problems experienced by the parent concerning the child, family, and the healthcare providers	<ul style="list-style-type: none"> - Fathers were encouraged to be more involved and offer assistance in caring for their children - Healthcare professionals should evaluate the patient and the caregivers holistically and provide knowledge and personalized care plans <p><i>Child:</i></p> <ul style="list-style-type: none"> - Child stared at - Lack of empathy - Tormented seeing their child in pain <p><i>Parent and family:</i></p> <ul style="list-style-type: none"> - Feelings of uncertainty - Restrictions in employment and leisure time, never being off duty - Difficulty to organize care - Guilty about the unequal division of time between the sick child and sibling <p><i>Healthcare professionals</i></p> <ul style="list-style-type: none"> - Difficult to diagnose the condition - Lack of information provided - Lack of knowledge
Tabolli S	Family burden in epidermolysis bullosa is high independent of disease type/subtype	2010	Italy	Observational, cross-sectional study: Questionnaire	To assess the family burden of epidermolysis bullosa in children aged 1–7 years of age	<ul style="list-style-type: none"> - Had to navigate the disease on their own - Family burden increased with: <ul style="list-style-type: none"> - The severity of the disease - Parents perceived disease severity - Parents with depression/anxiety <p>The family burden due to EB is high and independent of disease type/severity</p>
Wu YH	Family caregivers' lived experiences of caring for EB patients: A phenomenological study	2020	Taiwan	Qualitative phenomenological approach	To explore the lived experiences of family caregivers of epidermolysis bullosa patients	<p>Helplessness is associated with a rare disease</p> <ul style="list-style-type: none"> - Parents becoming experts - Couple dynamics ad financial strain - Traditional attitudes can cause emotional stress - Emotional and physical stress, unbalanced life - Religion is a support structure - Support from family, rare disease foundation - Need for support from social services, psychology, and schools.
Yuen W	The needs of parents with children suffering from lethal EB	2012	Netherlands	Qualitative study, semi-structured in-depth interviews	The needs of parents who have lost children to lethal EB	<ul style="list-style-type: none"> - Need for rapid referral to a specialist clinic - Honest communication - Palliative care - To be involved in the medical care of their child

Table 1 Continued

Author	Title	Year	Country	Study design	Aim	Findings
Kearney S	Living with EB: daily challenges and healthcare needs	2020	Ireland)	Qualitative study, semi-structured in-depth interviews	To explore the needs of children and families affected by EB	<ul style="list-style-type: none"> - To be informed of end-of-life decisions - Need for genetic counseling - Parents need physical support from the healthcare team - Access to home-based care - Psychosocial support - Parents working with multidisciplinary care teams - Assistance with administrative tasks <p>Mothers are impacted more than fathers</p> <ul style="list-style-type: none"> - Financial impact of leaving work, impact on the social and leisure time <p>Physical exhaustion following dressing changes</p> <p>Distressed that they inflict pain during the dressing changes. Feelings of sadness and powerlessness</p> <p>Some parents felt guilty for others to assist with care, other parents allowed assistance to rest</p> <p>Parents hid emotions from their children</p> <p>Child and parent collaborated around dressing changes; child felt more in control of the pain. Distraction techniques used. Important for doctors and psychologists to discuss coping strategies with the family</p>
Mauritz P	Impact of painful wound care in EB during childhood: an interview study with adult patients and parents	2019	Netherlands	Qualitative study, semi-structured in-depth interviews	How parents and patients experience the impact of wound care and which coping strategies they consider helpful	<p>The burden of disease and the length of dressing changes were proportionate to the severity of the disease</p> <p>Financial implications due to decreasing hours or stopping work, expensive dressings, physicians' visits</p> <p>Impact on family planning decisions and divorce</p> <p>Diminished quality of life, feelings of depression, and anxiety</p> <p>Learnt about dressing changes by trial and error, other families, not healthcare practitioners. Attention should be placed on this aspect of management</p> <p>Type of EB impacted on parents coping</p> <p>Increased need for family support</p> <p>Support from the medical team was adequate</p>
Bruckner A	The challenges of living with and managing EB: insights from patients and caregivers	2020	USA	Survey	To determine challenges and burden of disease of caregivers and patients with EB	
Macik D	The need of social support, life attitudes and life satisfaction among parents of	2015	Poland	Interviews	The effect of having a child with EB on the parent's life and psychological well-being	

Table 1 Continued

Author	Title	Year	Country	Study design	Aim	Findings
Martin K	children suffering from EB					Life attitudes varied according to the severity of the child's illness Higher level of existential vacuum and a lower level of life control, purpose, self-realization, and life satisfaction <i>Psychosocial care of the individual:</i> - access to knowledge - collaborative with - easy access to psychological support to improve body image - support during transition periods - pain support - social integration - access to professional healthcare <i>Psychosocial care of the family:</i> - make parents aware that their reaction impacts their child - support for parents - home nursing, information, family support.
	Psychosocial recommendations for the care of children and adults with EB and their family: evidence-based guidelines	2019	United Kingdom	The expert panel performed systematic review: •Mixed methodsdeveloped guidelines	Guidelines for patients and their families with EB regarding psychosocial recommendations	- access to online EB communities for support Important for families to make time for themselves - dressings are stressful- form routine, support, and assistance from nurses and family <i>Self-care for professionals looking after patients with EB:</i> - professionals should be adequately trained to care for the physical and psychosocial needs of the family - online platforms to disseminate knowledge - promote a collaborative relationship with the patient/family - healthcare professionals should have a support structure - work in a multidisciplinary team <i>Social needs:</i> - Support group - Family and friend's support - Communicating with other families - Need to balance work and family <i>Informational needs:</i> - Need for general medical information, patients' condition, medication, the impact of the disease on the child's development and long-term health - Services available - Support structures - Concern over the knowledge of healthcare practitioners - Valued receiving an early diagnosis
Pelentsov L	The supportive care needs of parents caring for a child with a rare disease: A scoping review.	2015	Austria	Scoping review •Qualitative	To outline the supportive care needs of parents caring for a child with a rare disease	

Table 1 Continued

Author	Title	Year	Country	Study design	Aim	Findings
						<p><i>Emotional needs:</i></p> <ul style="list-style-type: none"> - Emotional needs of parent associated with child's health needs, the severity of child's illness, and parents caring responsibilities - Felt overwhelmed about their child's illness, feeling of uncertainty - Coping ability was influenced by their diagnostic experience. Some felt that the health professionals did not address their emotional needs and would have appreciated a higher level of knowledge and support from HCP <p><i>Practical needs:</i></p> <ul style="list-style-type: none"> - Financial concerns - Reduce work hours or stop working - Stress that others looking after their child in their absence would need a level of knowledge about the condition - Need for respite <p><i>Physical needs:</i></p> <ul style="list-style-type: none"> - physical and emotional exhaustion - sleep deprivation, loss of appetite and weight, headaches - some had the rare disease as well <p><i>Spiritual and psychological needs:</i></p> <ul style="list-style-type: none"> - religion assisted some to find comfort some parents had a crisis in their faith - felt powerless and helpless. Offered very little psychological support

FDLQI, Family Dermatology Life Quality Index.

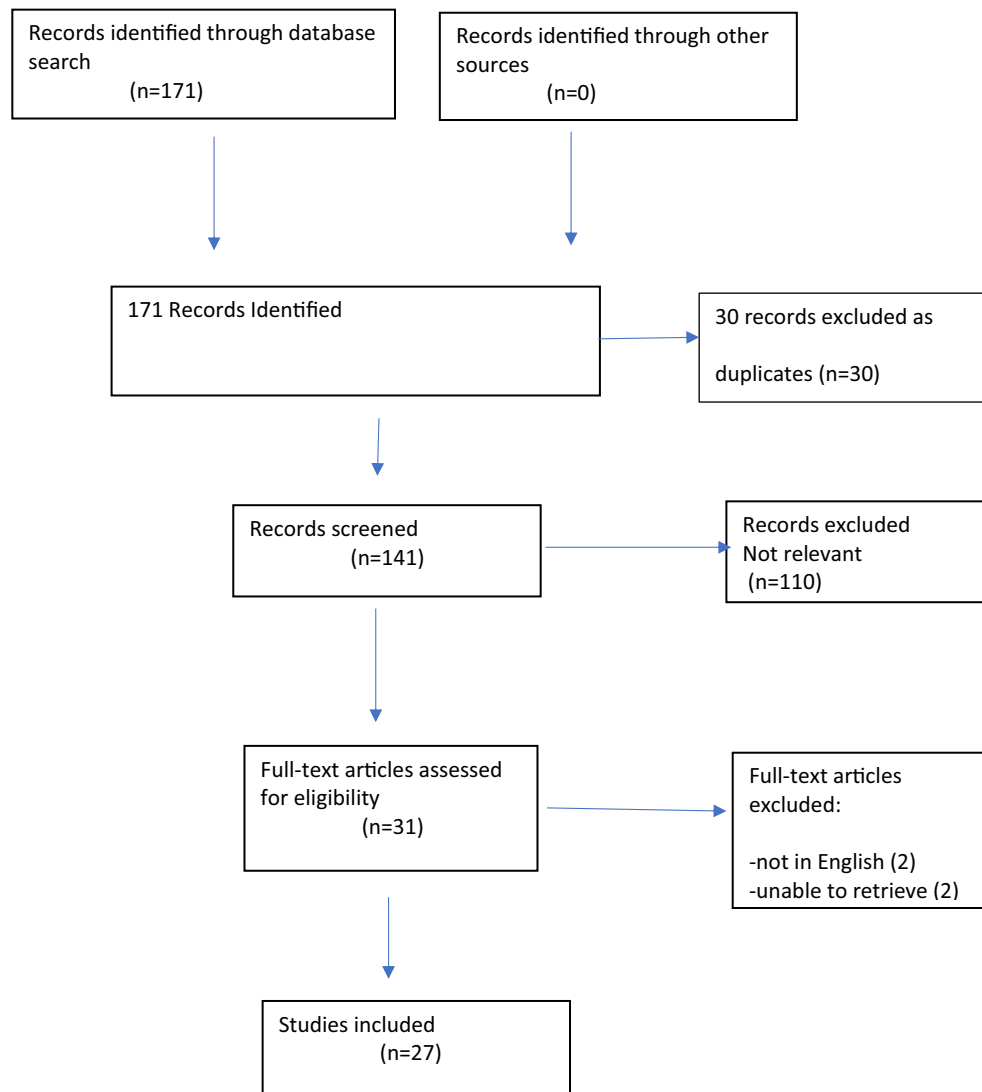


Figure 2 PRISMA flowchart of scoping literature

parents and the family as a whole. Strong family relations and a sturdy support structure are essential for the well-being of the patient and the family.

Parental interpersonal relationship

Constantly caring for a child with EB can place considerable strain on a couple's relationship.^{11,12} Parents can be so consumed with caring for their child that they spend little time together.^{11,13,14} EB has been shown to have a negative impact on the sexual relationships of couples, and this negative impact is correlated with the severity of their children's illness.^{11,15} A couple's decision to have more children was inversely proportional to their children's illness severity.^{11,16} Raising a child with EB is both physically and mentally challenging. The burden of care takes time away from doing things as a couple to focus on

their child's needs, which can strain the marital relationship.^{13,17} A 22% divorce rate among parents of children with EB is noted in a study by Fine et al.¹¹ The other reasons for marital strain and divorce included lack of support from a spouse in understanding the disease process or caring for their affected child.^{11,14,16}

Mothers were primarily responsible for caring for the child, and fathers were more removed and thus less affected. It is therefore encouraged that fathers of children with EB be more involved in providing care.¹⁸ It was suggested that it was important for the family to factor in leisure, recreation, vacations, and social life to prevent the breakdown of the family unit.¹⁹

Emotional impact

Parents experience a wide array of emotions at the birth of their baby with EB and throughout their child's life. There is initial

shock, anxiety, denial, helplessness, feeling overwhelmed, and fear at the birth of the baby, which may have been much anticipated.^{13,20} Some parents share feelings of loneliness, isolation, disconnection, and avoidance of social situations due to public embarrassment and humiliation.¹⁷ Parents have expressed feeling physically and emotionally exhausted, sleep-deprived, and frustrated that they are never off duty in caring for their child, with symptoms of loss of appetite and weight and headaches in need of respite.^{13,14,19,21,17} Physical and emotional exhaustion exacerbated by underlying depression and anxiety were associated with physical mistreatment of the child and parental suicidality.^{13,18} Lansdown et al. had noted that a parent had thoughts of initially rejecting their baby, but these thoughts gave way to affection and deep love.²⁰

Parents of ill children have a higher level of existential vacuum, a lower level of control, and life purpose and satisfaction. The greatest difficulties identified were the unpredictable nature of the illness, its impact on daily living, and the inability to make long-term plans.¹² Females were found to experience more depressive symptoms and used three coping strategies: problem-focused, emotional-focused, and dysfunctional coping, as well as having higher parenting stress levels that had a negative correlation with self-esteem.²² Physical dependency and the physical scars of EB cause an increased burden of disease on caregivers, while parents receiving assistance with caring had a lower burden score.

Dressing changes are long and painful procedures that can provoke negative emotions and lead to anxiety. Parents feel distressed and guilty that they are responsible for inflicting pain during dressing changes.^{14,19,21} Wu et al. mention that a mother had self-mutilated to identify with her child's pain.¹³ Parents face the difficulty of watching their child's condition deteriorate and feel guilty for wishing that their child passed away and also for passing on the disease to their children.^{14,15} Dyadic coping processes affect both the child and the parent. Parents hide their emotions from their children to protect them and focus on distraction techniques at difficult dressing changes. Nevertheless, children who assisted with the dressing changes felt more in control of their care.²¹

Regarding the receiving of the diagnosis, studies revealed that a parent's experience influenced their coping ability; that is, some parents felt that the HCPs did not address their emotional needs and would appreciate a higher level of knowledge and support from HCPs.¹⁷

Financial impact and sacrifices

Studies have highlighted that parents make great sacrifices for their families and give up careers and financial security to care for their children.^{13,14,16,21,23} Financial burdens are further compounded by medical practitioner costs, expensive medication, and dressings not covered by medical insurance.¹⁶ Hospitalization of the child may result in the working parent needing to take time off work to care for the home

and other children. However, it was observed that parents who had given up employment found it very difficult to reenter the work market.²³ Another question relevant to financial impacts, and one which raises ethical dilemmas too, is when the HCP suggests that families relocate to perform procedures such as a tracheostomy. Removing the family from their support structure and imposing a financial burden on the family may be detrimental to the patient's quality of life and the family.²⁴

Parental concerns regarding their children's health and needs and views of healthcare professionals

Parents felt uncertain about their children's future and the unpredictable nature of the disease.¹⁴ They expressed concerns about the lack of appropriate clothing and shoes for their children.^{15,20} Moreover, they were also apprehensive and stressed that others looking after their child in their absence would require a level of knowledge about the condition.¹⁷

In this line of thought, when the correct diagnosis and a prompt referral to a specialist center were given, there were challenges for many families. For instance, parents felt that healthcare practitioners lacked knowledge about the condition.^{13-15,25} Additionally, parents were not given appropriate information about their child's condition, and thus incorrect dressings were applied to their child's painful wounds.^{14,18,25} Parents felt a high turnover of home-based nursing care and therefore opted to treat their children themselves with a consequence of feeling that they had to navigate their child's care on their own and became the expert healthcare providers.^{13,14,17,21} They learned about wound dressing by trial and error, from patient communities and families rather than healthcare providers, except for specialist units.¹⁶

Nevertheless, parents expressed a necessity for rapid referral to an expert EB center for rapid diagnosis and prompt management.²⁵ There was a need for information about the disease, complications, impact of the disease on the child's development and long-term health, dressings, and appropriate clothing.^{15,17,25} There was a requirement for support groups accessible from rare disease foundations and online EB support groups.^{13,17,19} Also, parents felt that religion and traditional medication were a source of support and assistance.^{13,17} Maçik et al. noted that parents required family support but felt adequately supported by healthcare providers.¹² This was in contrast to what was noted in other studies.^{13-15,25}

On the positive side, specialist centers offer guidance, education, and relief for families, including a sense of empowerment and control.²⁵ Nurses trained in EB care allow parents the opportunity to escape the problematic dressing changes and time for downtime and respite.^{15,21} Patients require numerous doctors' appointments; parents felt that they should be appointed a care manager and an administrator to facilitate appointments and organize medical supplies, thereby eliminating fragmentation of care.^{14,15}

However, they preferred home-based care for their children over facility-based care.^{15,25} On the other hand, some parents felt that palliative home-based care was burdensome on the family and opted for a facility during end-of-life care.²⁵ In this sense, parents appreciated when they were informed honestly about their child's prognosis and could plan their time appropriately with the child. They did not want to prolong their child's pain and suffering and opted for pain and wound management only with no invasive procedures to prolong life.²⁵ Some parents in favor of euthanasia planned the location of death either at home or in a hospital.²⁵

In summary, there is a need for multidisciplinary care, including physicians, nurses, physiotherapists, psychologists, and pharmacists, in order to provide comprehensive and coordinated care.^{15,19} Although physicians might focus on the biomedical needs of the patient, there should be a holistic consideration of care, taking into account the physical, emotional, financial, or other strains on the patient and the family.^{11,13} There is a need for genetic analysis and counseling to assist parents to plan their families.²⁵

Family impact and support

Traditional attitudes and beliefs can cause emotional distress. Wu et al. noted that Chinese gender bias assigns blame to the daughter-in-law for genetic conditions, and coupled with the superstitious belief about sin being a form of punishment, this has led to emotional distress and strain within families.¹³ Family and friends offered support by cleaning, babysitting, and offering assistance with dressing changes; however, parents were reluctant to leave their children with family as they felt guilty if the child's fragile skin was injured.¹⁵

Societal reactions

Patients with EB have numerous vesicles that are covered in bandages and may have alopecia, onychia, and complications such as contractures and mitten deformities of their extremities, necessitating the use of wheelchairs. Parents have reported that people stare at their child and ask questions if the disease is contagious and if the child has been abused.^{14,20} Landsdown et al. noted that teachers in senior school lacked empathy and understanding, whereas junior schoolteachers asked for more information about the affected child's condition.²⁰

Impact of EB on the siblings

There have been very few studies that focus on the impact of EB on siblings. Reports have been within the context of the family and not solely focusing on their needs. Thus, studies that focus on the effects of a rare, chronic, and genetic disease on siblings have been extrapolated and included in this study.

Emotional experience/encounter

Siblings may experience a vast array of emotions, including sadness, helplessness, and fear, that they may get the

disease, guilt that they do not have the disease, embarrassment, resentment, and anger.^{26,27} Due to lengthy hospital stays, siblings are unable to develop relationships with their siblings.²⁶ Therefore, the impact of a debilitating, noncurable disease on siblings of children with EB should not be underestimated. The death of a sibling may result in separation anxiety; it is, therefore, essential that healthcare providers be aware of this and offer support.²⁵

Personal impact

Having a faith-based system can also function as a means of support for siblings of children with EB,²⁶ helping their sick siblings and being included in the treatment and care was important to them.²⁶ Studies showed that siblings were supportive, protective, and helpful, provided physical comfort, and took on a caregiver role in caring for their brother or sister.²⁸ Also, they play a social role as they kept their sick sibling in touch with the outside world through technology and social media; some were more active and lobbied and raised funds for their sibling's care.²⁸

However, the constant focus on the sick sibling makes the sibling "invisible" to the family,²⁶ with probably an unequal division of time and care of nonaffected siblings which, on the other hand, makes parents feel guilty and experience an increased burden of care.^{14,17,27} Due to the burden and focus of care, siblings may miss out on events associated with a parent, such as Mother's Day. This feeling of exclusion may contribute to further adjustment difficulties.²⁶ There may be changes in their daily routine, and they may miss out on school life and extracurricular activities.^{23,26} Another constraint is that parents may not explain the condition to the sibling or prepare them adequately for what they may encounter when they visit their sibling in the hospital; this may have a further negative impact.²⁶ Studies also demonstrate that parents did not explain the genetic implications of the disease, with some thinking it was only appropriate to discuss this when the sibling was age-appropriate and needed to make reproductive choices.²⁷ Parents who do explain the condition to the sibling might ask that they keep this a secret from their sick sibling, which may be additionally burdensome.²⁷

Impact of EB on healthcare providers

There is a paucity of studies that focus on the impact of EB on healthcare practitioners and providers (HCPs).

Emotional impact

Healthcare professionals felt the emotional toll and difficulty switching off after walking the long road with patients and families. They expressed feelings of sorrow during the palliative care process. There was frustration, self-criticism, guilt, powerlessness, and inadequacy that there was no cure and that the easing of pain was transitory.²⁹

Challenges

Epidermolysis is a rare disease with few trained specialists. Consultations are time-consuming, resources are limited, and working with service providers who do not understand the complexity of the disease and the needs of the patient/family can be taxing.²⁹ There are no boundaries to care; moreover, HCPs operate outside their area of expertise, such as being a counselor to the family.²⁹ There is a need for HCPs to be trained to care for the physical and psychosocial needs of the patient and their family.¹⁹

Personal needs

To cope, HCP requires self-care strategies, debriefing, and being part of support groups.²⁹ They should not work in isolation but rather in a multidisciplinary team, having access to psychological support, and reducing compassion fatigue and burnout.¹⁹

Attitude toward treatment

Healthcare professionals stated that they would refrain from life-prolonging treatment, they would provide life-shortening pain relief, and that a few would perform infanticide. Faith-based convictions influenced these choices.³⁰

Doctor–patient relationship

A good physician–patient relationship is essential to effective communication and a good therapeutic relationship. The physician may not be au fait with rare diseases, and thus patients may take on the role as the “expert patient.” Many physicians encourage a patient–physician partnership, while a minority oppose the assertive proactive patient behavior.³¹ Patients want to play an active role in their management, and the physician should acknowledge this and promote a collaborative relationship.¹⁹

Physicians’ responsibilities

Healthcare professionals should identify the psychosocial needs of the patients and their families. They should adopt a multidisciplinary approach to ease referrals and comprehensive, coordinated care of patients and their families.²⁹ Physicians should empower their patients to understand their disease, encourage self-reliance, and promote patient participation.³¹

Discussion

EB is a rare heterogeneous disease that varies in severity based on the subtype. Basra and Findley proposed the term the “Greater Patient” to describe the primary support system affected by a patient having a skin disease.³² Caring for a child with EB requires great care, love, and sacrifice from parents and their siblings and dedication, expertise, and empathy from the healthcare team.

Research thus far has largely been focused on diagnostics, classification of the disease, and therapeutics.³ There is a

paucity of research that focuses on the family and the healthcare team caring for patients with EB. This paucity ultimately impacts patients with EB. There is a lack of specialist centers that cater to the holistic management of these patients and their families. Many HCPs have little knowledge of the condition, resulting in a delay in diagnosis, appropriate referral, and essential management of these patients. Diagnostic delay is of great distress to the parents of these children.¹⁴ Healthcare professionals in the primary setting should be empowered with continued medical information in the form of courses and seminars with access to treatment guidelines to timeously diagnose, counsel, manage, and refer these patients and their families to specialist centers. COVID-19 has impacted meetings for continued medical information; in low-income countries, this information could be disseminated via online platforms such as Zoom® or Microsoft Teams®, for example. This could be offered by specialists within those countries or invited specialists from other countries. Treatment guidelines should be formulated taking into account the resource limitations within their setting.

The cultural setting of the family and the community and myths are often overlooked. Families need to be educated to dispel these myths, which may result in undue stress.¹³ Parents need to be educated about the disease process, potential complications, pathogenesis, and genetic counseling. They may require emotional and psychological support in the form of support groups, psychotherapy, and assistance of a palliative care team. Social support may refer to social work to assist with grants and funding for families at risk.

Siblings take on a caregiver role and show bravery to their families while navigating a wide array of emotions. Siblings make great sacrifices in missing out on social activities, with time away from school and friends. They may spend little quality time with their parents who care for their siblings at home or the hospital. They have to deal with a wide variety of emotions compounded by the societal reaction when their siblings are stared at and teased by peers. Poor school performance may be a result of coping difficulties. Awareness of a sibling's challenges and effective holistic intervention is essential, and support groups using technology may be helpful.

Patients and families of children with EB are psychologically, socially, and financially at risk. Multidisciplinary management and patient education programs are essential.²³ Care should be holistic by assessing disease burden, identifying families at risk of dysfunctional coping strategies and depression, and providing psychosocial interventions. Healthcare professionals should provide information, encourage supportive networks, coping strategies, and provide psychosocial interventions and problem-solving techniques.^{18,22}

Healthcare practitioners providing palliative end-of-life care are vulnerable to compassion fatigue and burnout, affecting the well-being of the clinician and the care that they provide to their patients.^{33,34} Healthcare professionals should be aware of their needs and access professional support.¹⁹ This assistance could

be sought from the employee assistance programs (EAP), psychologists, and psychiatrists at the healthcare professionals' place of work or in the private sector. The cost is usually free of charge at the healthcare professionals place of employment or covered by their health insurance.

Conclusions and implications for research practice

The patient suffering from EB impacts the parents, siblings, extended family, close communities, and healthcare providers. It is vital to be cognizant of these dynamics and be holistic and comprehensive in addressing the family's challenges, difficulties, and needs. There is a need for more specialist centers that holistically manage patients with EB and their families. Healthcare providers need to be aware of the impact of caring for these families and their need for self-care. Further research is required to address the gaps in knowledge regarding the impact of EB on siblings and healthcare practitioners. Further research should take into account the cultural context of the patients and their families.

Recommendations

There is a need for support groups for parents, siblings, and the family within the community. The use of technology is a powerful tool to aid communication, especially during the current COVID-19 pandemic. However, this may not be possible in a resource-limited environment in South Africa, where data and technology may not be accessible to our patients.

Healthcare professionals may benefit from debriefing sessions and the acquisition of coping skills. Sharing knowledge in continuing medical education seminars and outreach to general practitioners or primary care physicians will help the doctors who will initially diagnose and manage these patients before referral to a specialist center to improve care and prevent complications.

South Africa is faced with a significant burden of both communicable and noncommunicable diseases; healthcare policies do not recognize rare diseases as a public concern, and resources are directed to more common conditions. A utopian ideal would be for health policymakers, physicians, families, and patient associations to work together to address and manage patients with rare diseases. It would be advantageous for healthcare practitioners to partner with organizations such as Rare Diseases South Africa and DEBRA South Africa (Dystrophic Epidermolysis Bullosa Research Association) by partaking in research and forming patient and family support groups within various hospitals with annual meetings and sharing of ideas. This collaboration between these organizations/patient advocacy groups, healthcare practitioners, patients, and families could encourage engagement with policymakers to improve the lives of patients with rare diseases such as EB.

Funding should be made available for both qualitative and quantitative research in rare diseases such as EB. The management of EB requires a comprehensive approach that includes a rapid, accurate diagnosis. Following diagnosis, a multidisciplinary biopsychosocial treatment plan for the patient and the family needs to be implemented within the cultural context and with cognizance of resource limitations in our setting.

References

- Guttmann-Gruber C, Bauer JW, Piñón HJ. Hereditary bullous diseases: current and innovative models to study the skin blistering disease epidermolysis bullosa. *Drug Discov Today Dis Model* 2020; **32**: 17–25.
- Fine J-D. Epidemiology of inherited Epidermolysis Bullosa based on incidence and prevalence estimates from the National Epidermolysis Bullosa Registry. *JAMA Dermatol* 2016; **152**(11): 1231–1238.
- Has C, Liu L, Bolling MC, *et al.* Clinical practice guidelines for laboratory diagnosis of epidermolysis bullosa. *Br J Dermatol* 2020; **182**(3): 574–592.
- Arksey H, O'Malley L. Scoping studies: Towards a methodological framework. *International Journal of Social Research Methodology: Theory & Practice* 2005; **8**(1): 19–32.
- Tricco AC, Lillie E, Zarin W, *et al.* PRISMA extension for scoping reviews (PRISMA-ScR): Checklist and explanation. *Ann Intern Med* 2018; **169**(7): 467–473.
- Sampogna F, Finlay AY, Salek SS, *et al.* Measuring the impact of dermatological conditions on family and caregivers: A review of dermatology-specific instruments. *J Eur Acad Dermatol Venereol* 2017; **31**(9): 1429–1439.
- Sampogna F, Tabolli S, Di Pietro C, *et al.* The evaluation of family impact of recessive dystrophic epidermolysis bullosa using the Italian version of the family dermatology life quality index. *J Eur Acad Dermatol Venereol* 2013; **27**(9): 1151–1155.
- Frew JW, Martin LK, Nijsten T, *et al.* Quality of life evaluation in epidermolysis bullosa (EB) through the development of the QOLEB questionnaire: An EB-specific quality of life instrument. *Br J Dermatol* 2009; **161**(6): 1323–1330.
- Dufresne H, Hadj-Rabia S, Taieb C, *et al.* Development and validation of an epidermolysis bullosa family/parental burden score. *Br J Dermatol* 2015; **173**(6): 1405–1410.
- Tabolli S, Pagliarello C, Uras C, *et al.* Family burden in epidermolysis bullosa is high independent of disease type/subtype. *Acta Derm Venereol* 2010; **90**(6): 607–611.
- Fine JD, Johnson LB, Weiner M, *et al.* Impact of inherited epidermolysis bullosa on parental interpersonal relationships, marital status and family size. *Br J Dermatol* 2005; **152**(5): 1009–1014.
- Mącik D, Kowalska-Dąbrowska M. The need of social support, life attitudes and life satisfaction among parents of children suffering from epidermolysis bullosa. *Przegląd Dermatologiczny* 2015; **102**(3): 211–220.
- Wu YH, Sun FK, Lee PY. Family caregivers' lived experiences of caring for epidermolysis bullosa patients: A phenomenological study. *J Clin Nurs* 2020; **29**(9–10): 1552–1560.
- van Scheppingen C, Lettinga AT, Duipmans JC, *et al.* The main problems of parents of a child with epidermolysis bullosa. *Qual Health Res* 2008; **18**(4): 545–556.

- 15 Kearney S, Donohoe A, McAuliffe E. Living with epidermolysis bullosa: Daily challenges and health-care needs. *Health Expect* 2020; **23**(2): 368–376.
- 16 Bruckner AL, Losow M, Wisk J, *et al*. The challenges of living with and managing epidermolysis bullosa: Insights from patients and caregivers. *Orphanet J Rare Dis* 2020; **15**(1): 1.
- 17 Pelentsov LJ, Laws TA, Esterman AJ. The supportive care needs of parents caring for a child with a rare disease: a scoping review. *Disabil Health J* 2015; **8**(4): 475–491.
- 18 Kahraman S. Determination of caregiving burden of parents providing care to their children with epidermolysis bullosa. *Egypt J Dermatol Venerol* 2017; **37**: 1.
- 19 Martin K, Geuens S, Asche JK, *et al*. Psychosocial recommendations for the care of children and adults with epidermolysis bullosa and their family: Evidence based guidelines. *Orphanet J Rare Dis* 2019; **14**(1): 133.
- 20 Lansdown R, Atherton D, Dale A, *et al*. Practical and psychological problems for parents of children with epidermolysis bullosa. *Child Care Health Dev* 1986; **12**(4): 251–256.
- 21 Mauritz P, Jonkman MF, Visser SS, *et al*. Impact of painful wound Care in Epidermolysis Bullosa during Childhood: an interview study with adult patients and parents. *Acta Derm Venereol* 2019; **99**(9): 783–788.
- 22 Wen C-C, Chu S-Y. Parenting stress and depressive symptoms in the family caregivers of children with genetic or rare diseases: the mediation effects of coping strategies and self-esteem. *Ci Ji Yi Xue Za Zhi* 2019; **32**(2): 181–185.
- 23 Dufresne H, Hadj-Rabia S, Bodemer C. Impact of a rare chronic genodermatosis on family daily life: The example of epidermolysis bullosa. *Br J Dermatol* 2018; **179**(5): 1177–1178.
- 24 deSante-Bertkau JE, Shubkin CD, Nelson WA, *et al*. When specialty care is unavailable to rural families. *Pediatrics* 2019; **144**(6): e20191130.
- 25 Yuen WY, Duipmans JC, Jonkman MF. The needs of parents with children suffering from lethal epidermolysis bullosa. *Br J Dermatol* 2012; **167**(3): 613–618.
- 26 Hilkner S, Beck A, Tanaka E, *et al*. Perceptions of children's siblings hospitalized for chronic disease. *Revista de Enfermagem Referência* 2019; **IV Série**: 77–86.
- 27 Plumridge G, Metcalfe A, Coad J, *et al*. Parents' communication with siblings of children affected by an inherited genetic condition. *J Genet Couns* 2011; **20**(4): 374–383.
- 28 Nabors L, Cunningham J, Lang M, *et al*. Family coping during hospitalization of children with chronic illnesses. *J Child Fam Stud* 2018; **27**: 1482–1491.
- 29 Dures E, Morris M, Gleeson K, *et al*. You're whatever the patient needs at the time': The impact on health and social care professionals of supporting people with epidermolysis bullosa. *Chronic Illn* 2010; **6**(3): 215–227.
- 30 Norup M. Treatment of severely diseased newborns: a survey of attitudes among Danish physicians. *Acta Paediatrica* 1999; **88**(4): 438–444.
- 31 Budych K, Helms TM, Schultz C. How do patients with rare diseases experience the medical encounter? Exploring role behavior and its impact on patient-physician interaction. *Health Policy* 2012; **105**(2–3): 154–164.
- 32 Basra MK, Finlay AY. The family impact of skin diseases: the greater patient concept. *Br J Dermatol* 2007; **156**(5): 929–937.
- 33 Sorensen R, Iedema R. Emotional labour: clinicians' attitudes to death and dying. *J Health Organ Manag* 2009; **23**(1): 5–22.
- 34 Kearney MK, Weininger RB, Vachon ML, *et al*. Self-care of physicians caring for patients at the end of life: "being connected...A key to my survival". *JAMA* 2009; **301**(11): 1155–1164.