http://www.periodicos.uem.br/ojs/index.php/CiencCuidSaude
ISSN on-line1984-7513
DOI: 10.4025/cienccuidsaude.v19i0.50427

ORIGINAL ARTICLE

CHALLENGES OF FAMILY CAREGIVERS OF CHILDREN AND ADOLESCENTS WITH EPIDERMOLYSIS BULLOSA[1]

Kelly Cristina Soares de Azevedo Silva*
Leiliane Teixeira Bento Fernandes**
Mônica Valéria Morais de Oliveira***
Thalita Correia Braga****
Kenya de Lima Silva*****

ABSTRACT

Objective: to understand the challenges faced by family caregivers of children and adolescents with Epidermolysis Bullosa in the search for assistance in the Health Care Network. **Method**: exploratory-descriptive study, with a qualitative approach, carried out in an outpatient clinic of a University Hospital in Paraíba that accompanies 11 children/adolescents with EB. Data were collected through semi-structured interviews with five main caregivers and the closure was based on sufficiency criteria. For data analysis, the Thematic Analysis, proposed by Minayo, was followed. **Results:** the main challenges identified were the difficulty in defining the diagnosis and early initiation of adequate treatment; the onerous financial reality that permeates the deficiency in the supply of inputs and continuity of care; the gaps in the organization of the Health Care Network and the coordination of care by Primary Health Care that culminate in the search for the social support network due to the deficitin resolving issues regarding the rights of the child/adolescent. **Final considerations:** alert to the development of assistance directed to the family, in addition to the reorganization of health network services aiming at comprehensive care for these individuals through intersectoral action in order to reduce obstacles in carrying out specific care, continuous and individualized.

Keywords: Rare diseases. Epidermolysis bullosa. Family. Child health. Adolescent health.

INTRODUCTION

Rare genetic diseases imply a considerable problem that has not yet been addressed from the perspective of public health, as evidenced by a simple search in the Scientific Electronic Library Online(SciELO) carried out with the descriptors "rare genetic disease" or "rare diseases", in which only six publications related to this field[1].

In Brazil, in 2014, the *Política Nacional de Atenção Integral às Pessoas com Doenças Raras* (PNAIPDR, National Policy for Comprehensive Care for People with Rare Diseases) was instituted to reduce the morbidity and mortality of these individuals, including by expanding access in time to the diagnosis and available specific therapies[2]. However, the country still lacks greater investments in research in this area.

Among these rare diseases, there are Epidermolysis Bullosa, or Bullous Epidermolysis, (EB). This has four subtypes: EB Simple (EBS), EB Junctional (EBJ), EB Dystrophic - dominant (EBDD) and recessive - (EBDR) and mixed EB or Kindler Syndrome (KS), being characterized by chronicity and heredity through mutations in various structural proteins of the skin and mucous membrane. This division is useful for the establishment of general guidelines, however, in each subtype there are great genetic variability and phenotypic influence that determine the severity of clinical conditions[3].

The worldwide incidence of EB is not fully known due to the few published epidemiological studies[4]. In Brazil, according to DEBRA (National Medical Research Association dedicated to curing EB), it is estimated that

[1]Article extracted from: Graduation Course Conclusion Thesis of the Graduation in Nursing, from the Federal University of Paraíba. Developed alongside members of Group of Studies and Research in Children and Adolescent Health Care, attached to the program of Post-Graduation in Nursing of the UFPB (Federal University of Paraíba).

*Nurse. Member of the Grupo de Estudos e Pesquisa em Saúde da Criança e do Adolescente – GEPSCA (Group of Studies and Research in Children and Adolescent Health Care) of the Universidade Federal da Paraíba - UFPB (Federal University of Paraíba). E-mail: kellyazevedo.17@hotmail.com. ORCID iD: https://orcid.org/0000-0002-6029-9935.
**Nurse. Doctorby the Post-Graduation Program in Nursing of the UFPB (Federal University of Paraíba). E-mail: leilianeufpb@gmail.com. ORCID iD: https://orcid.org/0000-0003-2643-5638.
***Nurse. Hospital Universitário Lauro Wanderley (University Hospital Lauro Wanderley). Member of the Debra Brasil EB. E-mail: monicavaleria2012@bol.com.br. ORCID iD: https://orcid.org/0000-0003-3226-3828.
****Nurse. E-mail: thalitacorreia96@gmail.com. ORCID iD: http://orcid.org/0000-0002-4916-6875.
*****Nurse. Professor of the Universidade da Paraíba (Federal University of Paraíba). Doctor by the Escola de Enfermagem de Ribeirão Preto/Universidade de São Paulo- EERP/USP (School of Nursing of Ribeirão Preto/University of São Paulo). E-mail: kenya.cateq@gmail.com. ORCID iD:0000-0002-7955-2531

8:1,000,000 inhabitants have EB in the world and, in the country, approximately 1,600 people[5].

According to the National Registry of People with EB of DEBRA Brasil started in 2014, there are 994 individuals registered and of these there have been 147 deaths so far. As for the classification, including deaths, there are 328 people with EBDR, 325 with EBS, 53 with EBDD, 27 with EBJ and 261 with a subtype still undefined. No individuals with KS were identified and there was no gender predominance. Of the total registered, there are 406 alive aged between 0 and 15 years old, with a predominance of 145 cases of EBS, 109 with EBDR, 22 with EBDD, 4 with EBJ and 126 without defined subtype[6].

With regard to the general clinical picture, it results from congenital flaws in intraepidermal or dermoepidermal adhesion that lead to the formation of vesicles on the skin and mucous membranes that can be spontaneous or due to minimal trauma[4]. Pain and itching are recurrent symptoms and in individuals with EBDR they are worse. They also have more severe conditions, followed by EBJ, EBDD and EBS[7]. As for the most severe cases, some have mutations in genes expressed in other organs, in addition to the skin, resulting in EB associated with interstitial lung disease, congenital nephrotic syndrome, cardiomyopathy and pyloric atresia[3].

In addition, the fragility of the skin causes several injuries, generating chronic pain, especially when handling dressings and general care[8]. Family members refer that the treatment of wounds can last up to more than four hours a day[7].

Thus, there is great repercussion in the life of the individual and family members, both for the aspects experienced by the EB patient, such as skin appearance, physical pain, psychological suffering, and for the family members with the financial and psychological impacts on health care[9]. In addition, there is difficulty in diagnosis, which is characteristic of rare genetic diseases, since the signs and symptoms can be similar to those of common diseases[1].

Families of children with EB also need to deal with the negative impact of social stigma around the child, which experiences change to their self-image, which can result, over time, into depression[8], being of importance the existence of a strengthened support network for the quality of life of these individuals.

Given the relevance of the theme in public health, the importance of knowing the complex health needs experienced by these patients and caregivers and how they perceive their therapeutic process in the face of obstacles in accessing diagnosis and treatment[1] is evident. Thus, the present study is justified by the importance of recognizing the challenges and ways of coping with caregivers of children/adolescents with EB, when they exercise the role as care managers in the search for guarantee of assistance to their loved ones in the *Rede de Atenção à Saúde*(RAS, Health Care Network, HCN).

Thus, the following question arose: what are the challenges faced by family caregivers of children and adolescents with EB in carrying out daily care? To answer this question, the objective was: to understand the challenges faced by family caregivers of children and adolescents with EB in the search for assistance in the HCN.

METHODOLOGY

Exploratory-descriptive study with a qualitative approach, carried out between June and August 2019 with five main caregivers of children/adolescents with EB, at the dermatology clinic of a University Hospital in Paraíba, which is the outpatient referral service for individuals with EB in the state. This currently accompanies 11 children and adolescents with EB. Adolescence was understood until the age of 19[10].

Caregivers who met the following inclusion criteria were included in the study: being a caregiver/guardian of a child/adolescent diagnosed with EB; be over 18 years old; attend the clinic during the data collection period. And as exclusion criteria: not being in physical or psychological conditions at the time of data collection; present difficulties in understanding the questions of the research instrument. All the caregivers with whom they had contact met the inclusion criteria, however, due to the individuals' absences from appointments

scheduled by the service, for financial and transportation reasons, six participants were excluded.

Data collection was mediated by the indication of the service nurse who listed and informed the dates and times when the children/adolescents would be seen. Thus, the researcher came to the place to conduct a semi-structured interview with the following guiding questions: what is the experience of family members who care for children/adolescents with EB? How has the family lived since the child/adolescent was born and there was a diagnosis of EB? What challenges have you faced in caring for the child/adolescent with EB? Are you inserted in any group or Non-Governmental Organization (NGO) to support children/adolescents with EB? How have you done so that the child/adolescent is monitored in health services? What are the difficulties faced in the treatment?

The interviews were recorded in audio and transcribed in full, with duration varying between 15 and 52 minutes, with an average of 25 minutes. The closure of data collection was based on a sufficiency criterion , understood as the moment when the data begins to recur and it is already possible to draw a comprehensive picture that responds to the objective of the study[11]. The material obtained from the testimonies transcription was composed of 20 pages, written in Times New Roman font, size 12 and 1.5 spacing.

Data were submitted to analysis THEME p or through the exhaustive reading of the transcripts; exploration of the material and establishment of the central thematic units; and elaboration of an interpretative synthesis with the researcher's inferences from the literature inherent to the theme[12]. In this way, the following nuclei of meaning were constructed: The challenges faced in obtaining the diagnosis and carrying out the treatment of EB and Health care for children/adolescents with EB and the influence of support networks.

The study complied with the ethical guidelines for research with human beings and was approved by the Research Ethics Committee of *Hospital Universitário Lauro Wanderley* (University Hospital Lauro Wanderley), under opinion No. 3.333.184. All participants signed the Free and Informed Consent Form (ICF) and, to guarantee confidentiality, they were identified by the letter F, for "Family member", followed by the number of order of the interview and the degree of kinship with the child/adolescent.

RESULTS AND DISCUSSION

The five main caregivers of children/adolescents with EB were four biological mothers and an aunt, who is a foster mother. Regarding the number of children, two had two children; two had one child; and one had four children. They were aged between 24 and 40 years old and all lived outside the state capital, in cities in the interior regions of the sertão, agreste and the Paraíba forest zone. As for education, two had incomplete primary education; one an incomplete high school; another had complete high school; and one did not attend school. Regarding employment, four caregivers were unemployed and one was away from work due to depression due to the difficulty in living with the condition of the child.

With regard to the age of children and adolescents, one was 1 year and 9 months old (F3), two were 7 years old (F1 and F4), one was 12 (F5) and the other 14 years old (F2).

The challenges faced in obtaining EB diagnosis and of the treatment

Families of children/adolescents with rare diseases go through challenges in their experience of caring for these individuals who need multiple and continuous care at home. This confrontation requires time, dedication and reorganization of tasks based on changes demanded by the disease[1].

In addition, one of the biggest challenges faced by families is the difficulty in obtaining the diagnosis of EB. The participants' statements demonstrated that this process permeates several uncertainties, such as the mistake with other diagnoses, thus increasing the time for its definition:

> Before I took him to the doctor, I spent four months with him treating Impetigo, [...] then I took him to the pediatrician and he said: - This is a contagious bacterial disease **(F1, mother of the child).**

> The doctor said it was skin syphilis, gave her a medicine and an ointment, and she spent 13 years taking this medicine and using this ointment [...] they came to discover this disease {EB} of her recently, [...] in fact it's been over a year **(F2, aunt of the teenager).**

A study carried out with 210 individuals with EB demonstrated that in 119 cases the symptoms were observed at birth and 85 individuals obtained the diagnosis before the first year of life[7]. However, it should be noted that the clinical presentation may be uncertain in early childhood, either due to the lack of standardized clinical data or the rarity and variability of the disease, which contribute to incorrect diagnoses, resulting in inadequate treatments[13], as evidenced in this study.

Given this process, uncertainties arising from the clarity deficitand ambiguity of the information provided by professional cause a state of unpredictability in the caregivers in regards to the diagnosis:

> [...] I had faith that I would find out what that was. [...] until I had the knowledge of what it was, we {family} suffered a lot **(F1, mother of the child).**

This reality of the interviewees generated difficulties to manage care in the absence of appropriate drugs and adequate covers/dressing for the realization of the skin care:

> [...] I would buy socks and cut them underneath, and then mend the socks, as I still do today [...] as he did not have access to the material, so I would prefer not to put the dressing on so as not to pass that pain again, because every time I went to touch him, I pulled more, I hurt more, so I preferred to clean and leave it open [...] There were injuries that would take 30 days to heal **(F1, mother of the child).**

> [...] I used the ointment and put cotton, but it was too much suffering, it stuck too much and the other day it was suffering [...] today there is the appropriate material for the dressing, I do it myself, I learned from the girls here {referral clinic for EB} **(F3, mother of the child).**

After the child/adolescent receives the diagnosis, the families face several difficulties in obtaining the necessary follow-up for the treatment of the disease, since the referral clinic is located in the state capital, making it difficult to travel and increasing the costs of families with long-term care with the long outbound trips. The referenced family income shows the low socio-economic level of the participants, in which becomes difficult the access to key inputs for nutrition and treatment, barriers that compromise the continuity of care:

> [...] at the beginning my husband rented a car and spent the whole day here {capital} and sometimes I needed to return during the week, it was difficult, it was a lot spent, often even without having it, it is not easy. [...] today we come more through SESAI {Special Secretariat for Indigenous Health} when there is no vacancy, we come through the city hall, and when there is no vacancy in one or the other, we have been paying [...] three years ago he used a supplement that was enriched milk, [...] the nutritionist spent eight cans a month, I only bought four because the cost is also high, and it was the people who bought it, [.. .] today we only supplement when he has the flu or when he has no appetite **(F1, mother of the child).**

> [...] the food is by us {family}, there are days, sometimes we come with a penny so that she can eat and I stay hungry (F2, aunt of the teenager).

> Because that way she can't travel in the sun, it has to be an air-conditioned car, but sometimes they put me on the bus, so I prefer to travel at night because it's not so hot, but for that I need to come first. [...] I prefer to travel from 1:00 am that we arrive here at dawn and don't need to stay in the support house (F3, mother of the child).

In addition to the difficulty in obtaining transportation, increasing the costs of these families with long commuting trips, they still face financial difficulties to meet the high cost of treatment. The care directed to these patients, regardless of the type of EB, is nonspecific, consisting of nutritional support, use of topical dressings and local care of the vesicles with essential fatty acid oils and antibiotics, in cases of secondary infection, in addition to measures of being careful, especially with children, in order to avoid trauma. In addition, it is recommended that a trained health team carry out a semiannual assessment to analyze skin and extracutaneous involvement[8].

However, so that the family have access to inputs provided by the public health system, diagnostic confirmation is required through medical report to formally request the responsible agencies:

> The doctor {doctor at the referral clinic} gave me

> the dressing report [...] gave me some samples and explained how I used it **(F1, mother of the child).**
>
> She doesn't receive the dressings [...] that's what I want to try too, for her to be entitled to these dressings. [...] **(F2, the teenager's aunt).**
>
> Because even today she has nothing, the dressing is only done when she comes here {to the referral clinic}, at home I have nothing **(F4, mother of the child).**

However, even with the report, other barriers need to be overcome for the acquisition of these inputs, such as the transfer of responsibility between the various spheres of government, culminating, sometimes, in lawsuits:

> [...] there are also the milk cans [...] the health secretary from the city supplies it, six cans were supposed to come, but they only give two **(F2, the teenager's aunt).**
>
> [...] I went to the prosecutor's office and filed for justice, and I'm receiving the milk, they said it is six months from the state and six months from the municipality, then I get six cans **(F3, mother of the child).**
>
> [...] I received the dressings in court, every month I receive them, yesterday I went to get them, then every month the doctor {from the referral clinic} gives me the prescription and I will get it for both of us. [...] The doctor still passes the medicines out, when you can buy them, I buy them, when you can't, you run out (F5, mother of the teenager).

The health demands of these children/adolescents are burdensome, which leads the family to make a pilgrimage to obtain social security through the *Benefício da Prestação Continuada* (BPC, Continuous Installment Benefit):

> So, the difficulties are mainly financial, the doctor {from the referral clinic} gave me the report, I filed for the benefit, but it was denied. [...] I checked in again and it is in the hands of a lawyer here in the capital. Then she {child} already did the survey. I'm just waiting for a social survey to come home (F3, mother of the child).
>
> We have no benefit, it has been denied several times, now it has been in court for a year. [...] The biggest difficulty is financial **(F5, mother of the teenager).**

Despite the difficulty, a caregiver reported that she had already achieved this regularization:

> She receives a benefit from the INSS {National Institute of Social Security}, which I retired when she was one to two years old [...] it's been thirteen years since she received this benefit **(F2, aunt of the teenager).**

In this context necessary daily care to these subjects, emphasizes the relevance of the BPC as essential for families, since most of the participants do not have the resources for lack of employment bond. Ratifying research[7], in which 52% of participants, including patients and caregivers, reported that EB causes a high financial burden.

However, coping with families goes beyond financial difficulties, since, once the uncertainties with the diagnosis are overcome, there are those related to the prognosis, since it is a disease with little information about it:

> When the doctor {from the referral clinic} said Epidermolysis Bullosa, I said: - My God, this is a monster. [...] without knowing how you are going to solve it, without knowing if it will work, if there will be a doctor who will actually follow up [...] the expectation I was given was 15 years **(F1, mother of the child).**

This difficulty in relation to the availability of information about the disease causes the social stigma resulting from the characteristic of the skin:

> Then the last straw is prejudice, it's a sadness, once I arrived at the post {Family Health Strategy} [...] then the nurse took his little hand and [...] went straight to the alcohol gel [...] she thought it was that Bubbly Scab [...] then I said to her: - Don't worry, you will not touch the lady **(F1, mother of the child).**
>
> [...] when she was small, the school did not want to receive her, the people had prejudice, then she was unwilling to go to school **(F2, aunt of the teenager).**

For the family, the care to be provided to children/adolescents with EB is difficult, as the disease causes several scars of skin and mucosa resulting from injuries that compromise their physical appearance and trigger prejudice in social environments, including at school. In addition, there are other challenging factors for children/adolescents, since they are unable to participate in common activities for their age group due to pain, itching or to prevent injuries, which contributes to making them feel different

and, coupled with negative comments from people, suffer psychological changes[14]. Studies stated that EB affects individuals' ability to socialize, including making new friends[7].

As a consequence, this emotional impact also affects people close to the patient, as is the case of one of the caregivers in the study, who reported having been diagnosed with depression for this reason, corroborating research that identified 57% of caregivers of people with EB were anxious or worried and 34.4% depressed[7].

The renounce from work is also a stressor that causes physical, emotional and financial repercussions for the whole family, reaching, on a larger scale, parents who need to appropriate the role of full-time caregivers deciding not to work or reduce their burden of work for this reason[7].

This emotional burden can manifest itself silently, requiring a sensitive look and attentive listening by the multiprofessional team to all the family's needs[15]. These situations make the whole family vulnerable, for this reason, this search in religious beliefs means to re-signify the meaning of life:

> [...] the psychological structure of us {family} is very shaken [...] I spent a period that I don't think I lived [...] I just focused on that, I didn't see my son, [...] before I only saw 'Epidermolysis bullosa', today I see that he can do a lot. [...] God has been giving me strength, a lot of strength **(F1, mother of the child).**

> When I had her I almost got depressed, because I cried all the time, I think it was because of her problem. [...] And the father doesn't say anything, just once she was a month and a half I went to bathe in front of him and she was crying a lot, he started to cry and said that if it were to be born like this, God had better take her **(F3, mother of the child).**

The family members cling to religion and the spirituality as sources of help and protection, as when they experience chronic illness they go through traumatic circumstances that arouse feelings of anxiety and insecurity before the unexpected[15].

It is evident the complexity of the path to obtain adequate care for children/adolescents with EB by family members, who still need to live with the social prejudice resulting from the ignorance of the population and even some health professionals about the disease.

Health care for children/adolescents with EB and the influence of support networks

The attention of public agencies, including professionals *Rede de Atenção à Saúde* (RAS, Health Care Network, HCN), the provision of assistance and supplies and home care supported by teams of Primary Care in Health Care (PHC) are very important factors in the lives of children in families/adolescents living with chronic diseases[16].

Managing the disease on a daily basis causes emotional and physical dysfunctions to caregivers. Thus, a well organized and interconnected health care network between different levels and services, in order to offer material resources, qualified assistance and territorial decentralization, can contribute to strengthen the family-service-community link and minimize these burdens. However, the difficulty in making access to health services effective has led the family to trace their own trajectory in the HCN:

> There was a period here at the hospital {reference} that the doctor would not see anymore without an appointment [...] I spent six months without seeing him, I would come, bring my son, but I could not speak to the doctor [...] I had time that he was in a deplorable situation and what could I do? [...] the doctor said: - Unfortunately, I can't do anything for your son [...] it has to be via PSF {Programa de Saúde da Família, Family Health Program} **(F1, mother of the child)**.

> All the assistance she has had until today is me who goes after it, and the treatment it's us who pay it on our own **(F4, the child's mother)**.

There are countless difficulties faced by family members related to health services, including the absence or insufficiency of information on the rights of children/adolescents and the distant relationship with professionals[15]. This fragmentation in the levels of care renders the system inefficient to meet the demands of care for children/adolescents with chronic illness and their family, as they need continuous care and commitment from the professionals in charge to plan health maintenance actions in all areas stages of the disease[17].

However, some positive changes can be seen

over time, for example, the articulation between the different sectors of the reference hospital, which has reduced the referral bureaucracy:

> Today, it is score 10/10 everything I dreamed of, the dermatologist accompanies me, the nurse does the dressings, there is a dentist, nutritionist, lung doctor, otorhinolaryngologist, these things we do here {Reference hospital}, because the doctor sends and we can already book here, without having to go to the PSF, which made people's lives much easier, which before it was very difficult to get an appointment **(F1, mother of the child).**

It is noteworthy that in spite of the specialized service have been cited as positive, it is required greater participation of teams from the Family Health Strategy (FHS) in the care, because it ends up transferring responsibility for the referral service, excluding construction of a Singular Therapeutic Project (STP) linked to other levels of assistance[17].

In this sense, the importance of social support is observed, since the interviewees reported that the resolution of day-to-day difficulties is primarily resolved by these networks:

> My friend brought a lot of content; she had internet access and I didn't, so she did a handout with information about the disease, [...] she was my gateway to knowledge, [...] after I started to understand, to know, I started taking care. [...] I did not imagine that there were so many people with this problem [...] and knowing that these people survived, for me it was very good. [...] then you will be comforted because you will see that you are not alone **(F1, mother of the child).**

It is noticed that the formal network organized to care for the health needs of children/adolescents with EB is deficient, with a greater concentration of care in the specialized reference service. This leads to a gap in the integrality and continuity of care due to the difficulty of communication between the other points of the HCN, thus interfering in the resolvability of care at home[18].

Such difficulties lead these families to seek assistance in specialized services, informally, through contacts with friends and family, demonstrating the fragmentation in the assistance network, since bonds are being built through families with secondary and tertiary services. From HCN, since the participants choose this level of care due to the multidisciplinary service and satisfactory social support.

Another strategy carried out by these families has been the support network between families or with religious entities, including the purchase of dressings:

> [...] she {adolescent with EB} participates in a group of online message exchanges [...] because when someone has a surplus of bandages, they donate **(F2, aunt of the teenager)**.

> [...] I receive help from the community, the people from church, when they see that we need it, they go and help, not every month, not every week, but they always help **(F5, mother of the teenager)**.

Non-Governmental Organizations (NGOs) have also played an important role in helping these individuals and they cross geographical barriers:

> [...] I started to accompany an NGO from Bahia that helps people with Epidermolysis Bullosa through social networks, then I saw her posts, I got in touch {with the social network administrator} and she gave me the office number, we made an appointment [...] they came to C.G. {city}, I went there to meet her. So she took my daughter's data, when she was about a month old, the first box of bandages arrived [...] it has been two months since I received it from the NGO **(F3, mother of the child).**

In this context, the reports show a weakening of the care process in the health network's points of care, causing the demand for this care to be found in philanthropic and religious entities and in strengthening the bonds between families. It is essential to support these caregiving families through discussions about this health condition, in order to enhance the living conditions of the entire family nucleus[19].

FINAL CONSIDERATIONS

The experience of the caregivers participating in this study is involved in daily challenges, amid feelings of anxiety and fear and the work overload caused by a continuous demand for child/adolescent care. There was, still, the financial impact and the difficulty of access to inputs necessary for the realization of care related to shortcomings in the organization of health care and coordination of care network.

In addition, caregivers face a deficit of preparation to perform home care; obstacles to access and monitoring in PHC services; little knowledge and resolution regarding the rights of the child/adolescent; as well as the limitation of support services and the ignorance of society to receive the child/adolescent in social life.

Regarding the limitations of the study, the results refer to the experience lived in a specific reality. In this context, one must caution in generalizations to other situations prevailing in the country, being necessary other studies involving this theme to deepen their knowledge and contribute to the continuity of care to children/adolescents with EB and their families. It is noteworthy that in the area of public health, especially for nursing, there is provided increased visibility of these individuals and their families in order to contribute to the rethinking of the implementation of HCN to improve service.

DESAFIOS DE CUIDADORES FAMILIARES DE CRIANÇAS E ADOLESCENTES COM EPIDERMÓLISE BOLHOSA

RESUMO

Objetivo: compreender os desafios enfrentados por cuidadores familiares de crianças e adolescentes com Epidermólise Bolhosa na busca pela assistência na Rede de Atenção à Saúde. **Método**: estudo exploratório-descritivo, de abordagem qualitativa, realizado em ambulatório de um Hospital Universitário na Paraíba que acompanha 11 crianças/adolescentes com EB. Realizou-se a coleta dos dados por meio de entrevista semiestruturada com cinco cuidadoras principais e o encerramento por critério de suficiência. Para análise dos dados, seguiu-se a Análise Temática, proposta por Minayo. **Resultados:** os principais desafios identificados foram a dificuldade na definição do diagnóstico e início precoce do tratamento adequado; a realidade financeira onerosa que permeia a deficiência na oferta de insumos e continuidade do cuidado; as lacunas na organização da Rede de Atenção à Saúde e coordenação do cuidado pela Atenção Primária em Saúde que culminam na busca pela rede de apoio social devido ao *deficit* na resolutividade em relação aos direitos da criança/adolescente. **Considerações finais:** alerta-se para o desenvolvimento de uma assistência direcionada à família, além da reorganização dos serviços da rede de saúde objetivando a integralidade do cuidado a esses indivíduos por meio da atuação intersetorial a fim de reduzir obstáculos na realização de um cuidado específico, contínuo e individualizado.

Palavras-chave: Doenças Raras. Epidermólise Bolhosa. Família. Saúde da Criança. Saúde do Adolescente.

DESAFÍOS DE LOS CUIDADORES FAMILIARES DE NIÑOS Y ADOLESCENTES CON EPIDERMÓLISIS AMPOLLOSA

RESUMEN

Objetivo: comprender los desafíos enfrentados por cuidadores familiares de niños y adolescentes con Epidermólisis Ampollosa (EB) en la busca por asistencia en la Red de Atención a la Salud. **Método**: estudio exploratorio-descriptivo, de abordaje cualitativo, realizado en ambulatorio de un Hospital Universitario en Paraíba-Brasil que acompaña a 11 niños/adolescentes con EB. Se realizó la recolección de los datos por medio de entrevista semiestructurada con cinco cuidadoras principales y el cierre por criterio de suficiencia. Para el análisis de los datos, fue seguido el Análisis Temático, propuesto por Minayo. **Resultados:** los principales desafíos identificados fueron la dificultad en la definición del diagnóstico e inicio precoz del tratamiento adecuado; la realidad financiera onerosa que raya el déficit en la oferta de insumos y continuidad del cuidado; las lagunas en la organización de la Red de Atención a la Salud y coordinación del cuidado por la Atención Primaria en Salud que culminan en la búsqueda por la red de apoyo social debido al *deficit* en la resolución respecto a los derechos del niño/adolescente. **Consideraciones finales:** es importante el desarrollo de una atención dirigida a la familia, además de la reorganización de los servicios de la red de salud, teniendo como objetivo la integralidad del cuidado a estos individuos por medio de la actuación intersectorial con la finalidad de reducir obstáculos en la realización de un cuidado específico, continuo e individualizado.

Palabras clave: Enfermedades raras. Epidermólisis ampollosa. Familia. Salud del niño. Salud del adolescente.

REFERENCES

1. Iriart JAB, Nucci MF, Muniz TP, Viana GB, Aureliano WA, Gibbon S. From the search for diagnosis to treatment uncertainties: challenges of care for rare genetic diseases in Brazil. Ciência & Saúde Coletiva. 2019; 24(10): 3637-3650. doi: https://doi.org/10.1590/1413-812320182410.01612019.
2. Brasil. Ministério da Saúde. Portaria n.199, de 30 de janeiro de 2014. Institui a Política Nacional de Atenção Integral às Pessoas com Doenças Raras, aprova as Diretrizes para Atenção Integral às Pessoas com Doenças Raras no âmbito do Sistema Único de Saúde (SUS) e institui incentivos financeiros de custeio. Diário Oficial da União [Internet]. Disponível em: https://bvsms.saude.gov.br/bvs/saudelegis/gm/2014/prt0199_30_01_2014.html. Acesso em: 07 jun. 2020.
3. Vahidnezhad H, Youssefian L, Saeidian AH, Uitto J. Phenotypic Spectrum of Epidermolysis Bullosa: The Paradigm of Syndromic versus Non-Syndromic Skin Fragility Disorders.

Journal of Investigative Dermatology. 2018; 139, 522-527. doi: https://doi.org/10.1016/j.jid.2018.10.017.

4. Koga H, Prost-Squarcioni C, Iwata H, Jonkman MF, Ludwig RJ, Bieber K. Epidermolysis Bullosa Acquisita: The 2019 Update. Front. Med. 2018; (5): 362. doi: https://doi.org/10.3389/fmed.2018.00362.

5. DEBRA Brasil. Quem somos – Cadastro [Internet]. 2019. Disponível em: debrabrasil.com.br/quem-somos/cadastro/. Acesso em: 09 jun. 2020.

6. DEBRA Brasil. Cadastro Nacional de Pessoas com Epidermólise Bolhosa. 2019.

7. Bruckner AL, Losow M, Wisk J, Patel N, Reha A, Lagast H, et al. The challenges of living with and managing epidermolysis bullosa: insights from patients and caregivers. Orphanet Journal of Rare Diseases. 2020; 15:1. doi: https://doi.org/10.1186/s13023-019-1279-y.

8. Lima LF, Vasconcelos PF. Epidermólise bolhosa: suas repercussões restritivas na vida diária do paciente. J. Health Biol Sci. 2019; 7(4): 423-428. doi: http://dx.doi.org/10.12662/2317-3076jhbs.v7i4.2657.p423-428.2019.

9. Benício CDAV, Carvalho NAR, Santos JDM, Nolêto IRSG, Luz MHBA. Epidermólise bolhosa: Foco na assistência de enfermagem. Revista Estima. 2016; 14(2): 91-8. doi: http://dx.doi.org/10.5327/Z1806-3144201600020007.

10. World Health Organization – WHO. Young People's Health – a challenge for society. Reporto of a WHO study group on young people and health for all. Technical Report Series 731. Geneva: WHO; 1986. Available from: http://apps.who.int/iris/bitstream/10665/41720/1/who_trs_731.pdf. Acess in: 09 out. 2019.

11. Minayo MCS, Assis SG, Souza ER. Avaliação por triangulação de métodos: abordagem de programas sociais. Rio de Janeiro: Editora Fiocruz; 2014.

12. Minayo MCS. Qualitative analysis: theory, steps and reliability. Ciência & Saúde Coletiva. 2012; 17(3): 621-26. doi: https://doi.org/10.1590/S1413-81232012000300007.

13. Laimer M, Pohla-Gubo G, Diem A, Prodinger C, Bauer JW, Hintner H. Epidermolysis bullosa House Austria and Epidermolysis bullosa clinical network. Wien Klin Wochenschr. 2017; 129:1–7. doi: https://doi.org/10.1007/s00508-016-1133-3.

14. Jain SV, Murrell DF. Psychosocial impact of inherited and autoimune blistering diseases. Intern J Wom Dermatol. 2018 Mar; 4(1): 49-53. doi: https://doi.org/10.1016/j.ijwd.2017.11.004.

15. Dias BC, Ichisato SM, Marchetti MA, Neves ET, Higarashi IH, Marcon SS. Challenges of family caregivers of children with special needs of multiple, complex and continuing care at home. Escola Anna Nery. 2019; 23(1). doi: https://doi.org/10.1590/2177-9465-ean-2018-0127.

16. Martins LA, Silva TCC, Santos HAS, Aguiar ACSA, Whitaker COM, Camargo CL. Therapeutical itinerary of quilombola children with falciforme disease. Cienc Cuid Saude. 2019; 18(2): e45177. doi: https://doi.org/10.4025/cienccuidsaude.v18i2.45177.

17. Vaz EMC, Collet N, Cursino EG, Forte FDS, Magalhães RKBP, Reichert APS. Care coordination in Health Care for the child/ adolescent in chronic condition. Rev Bras Enferm [Internet]. 2018; 71(6): 2612-9. doi: http://dx.doi.org/10.1590/0034-7167-2017-0787.

18. Silva MEA, Reichert APS, Souza SAF, Pimenta EAG, Collet N. Doença crônica na infância e adolescência: vínculos da família na rede de atenção à saúde. Texto Contexto Enferm. 2018; 27(2): e4460016. doi: http://dx.doi.org/10.1590/0104-070720180004460016.

19. Silveira A, Neves ET. The social network of adolescents who need special health care. Rev Bras Enferm. 2019; 72(2): 442-9. doi: http://dx.doi.org/10.1590/0034-7167-2018-0543.

Corresponding author: Kelly Cristina Soares de Azevedo Silva. Address: Street João Ferreira dos Santos, nº 496, centro, Marcação, Paraíba, Brasil. Telephone: (83) 99154-9667. E-mail: kellyazevedo.17@hotmail.com.

Submitted: 16/10/2019
Accepted: 06/07/2020