Abstract

Epidermolysis bullosa is an inherited disease characterized by severe skin fragility and blister formation in response to minor trauma. Apart from the main features mentioned above, patients suffer from chronic pain, pruritus, skin infections, but also from more severe systemic symptoms and even mutilations. All these aspects can potentially influence the psychological well being of both patients and family members. This review article summarizes the information published in the literature regarding quality of life (QoL), generic and specific questionnaires and also psychological interventions available for epidermolysis bullosa patients and caregivers.

Cite this article

Rezumat

Epidermoliza buloasă este o afecțiune genetică caracterizată prin fragilitate cutanată marcată și formarea consecutivă de leziuni buloase ca răspuns la traumatisme minore. În afara trăsăturilor principale menționate anterior, pacienții sunt afectați de durere, prurit, infecții cutanate dar și de manifestări sistemice și chiar mutilări. Toate aceste aspecte au abilitatea de a influența starea psihică a pacienților dar și a familiilor acestora. Articolul de față rezumă informațiile existente în literatura de specialitate cu privire la calitatea vieții (QoL), chestionare generice sau specifice dar și intervențiile psihologice disponibile în cazul pacienților suferind de epidermoliză buloasă și a familiilor acestora.
Introduction:

Inherited epidermolysis bullosa is a genodermatosis characterized by skin and mucosa fragility with subsequent formation of blisters in response to minor trauma. Because of the skin fragility, children affected by the disease are also referred to as “butterfly children”. Epidermolysis bullosa subtypes vary depending on the distribution of the lesions, the level of blister formation, the underlying molecular defect, the internal organ involvement and the wound healing response (1).

Therefore, the spectrum of epidermolysis bullosa varies from minimal - or no internal organ involvement and a normal lifespan - to multiorgan disorders, severe mutilation, profound impact on the quality of life of both patients and caregivers, as well as a shorter expectancy of life (1).

Quality of life (QoL) is a complex concept designed to reflect the negative impact of an individual’s disease on a physical, psychological and social level. However, the burden of the disease on the individual is often extended to family members and caregivers, therefore a holistic approach in QoL assessment should also include the persons in close contact or who take care of the patient (2).

Quality of life measurements:

The assessment of QoL in epidermolysis bullosa has major benefits because the information provided by these measurements can be used in clinical setting, treatment management and also research (3).

A research conducted in Scotland in 2002 assessed QoL using the Dermatology Life Quality Index (DLQI) and the Children’s Dermatology Life Quality Index (CDLQI). The conclusion was that the effect of epidermolysis bullosa on the QoL was similar to that of atopic dermatitis and psoriasis (4).

An observational, cross-sectional postal survey study conducted by Tabolli et al. using different questionnaires (Short Form-36 SH-36, Skindex-29, General Health Questionnaire-12 GHQ-12, EuroQo15, Patient Global Assessment five-point scale and the Family Strain Questionnaire) aimed to assess the burden of different types of epidermolysis bullosa on a group of Italian patients and their families. The results highlighted the fact that the mental components were not significantly impaired, but the physical components of the SF-36 showed lower scores. Regarding the impact of different epidermolysis bullosa types, the researchers concluded that patients with severe generalized dystrophic and junctional forms reported lower values in the GHQ-12 scores than patients with epidermolysis bullosa simplex. No significant differences were observed regarding EB subtypes for Skindex-29, but women proved to be more affected than males in all Skindex-29 and SF-36 scales, suggesting a higher risk for anxiety and depression. Also, the QoL of both patients and caregivers decreased with increasing patient body surface involvement. The analysis of the differences in mobility and self-care in children versus adult patients revealed that anxiety and depression regarding the aspects mentioned above were higher among adults (5).

The first epidermolysis bullosa specific QoL questionnaire, QOLEB (Quality of Life Evaluation in Epidermolysis Bullosa), an English 17 item measurement tool, was developed in 2009. The QOLEB gives different QoL scores for different epidermolysis bullosa subtypes and was statistically proven to be valid and reliable in assessing the impact of epidermolysis bullosa on patients’ lives (6). It allows the stratification of QoL impairment into very mild, mild, moderate, severe and very severe, as well as monitoring the changes in QoL during the administration of new treatments in clinical trials (7).

Recently, the QOLEB was translated into Brazilian Portuguese (8) and Dutch. The latter translation was used together with other measurement tools to assess the impact of Epidermolysis bullosa on QoL in the Dutch population. The authors concluded that the results obtained in their study are similar to those of Tabolli et al. described above (9).

Many quality of life measurement tools, both quantitative and qualitative, have been used in the attempt to assess the impact of epidermolysis bullosa on the patients, as well as on their families. The quantitative tools prove to be the most statistically accurate and valid, but generic dermatology tools have been used with success in some circumstances (3).

Patients’ quality of life:

Patients’ lives are affected mainly by the clinical manifestations and the disease severity (10). Studies have shown that these patients also suffer from different psychological and psychiatric problems (11).

Adolescents have a need to be accepted and liked. According to Eric Erikson, the quest for identity is the principal developmental task confronting every individual throughout the lifespan. Body image affects our emotions, thoughts, and behaviours in everyday life. But, above all, body image influences our relationships. Furthermore, body image has the potential to dramatically influence our quality of life.

Being visibly different, as is the case in many children and adults with EB, can be the cause of dissatisfaction with appearance, psychological difficulties and social problems. The difficulties with physical appearance experienced by youngsters are also a cause of concern for the parents. Williams et al. (12) relate the experiences of young people with EB simplex. Even with a mild form of EB, the children interviewed felt visibly different to others and sometimes excluded by their peers. The subjects in this study also experienced others’ fear of contagion.

In another qualitative study, this time with adults, Dures et al. (13) look into the impact of the disease on the beholder. A continuum can be drawn with patients who found their skin condition to affect all
areas of their lives on one end, with those who had learned to live with it on the other end. Some of the participants interviewed expressed their desire to have more personalized therapy, a one-to-one support to help them cope with their emotional distress.

Recently, a group of researchers aimed to determine the relationship between hand function in epidermolysis bullosa (EB) and quality of life, conducting a cross-sectional study that included children suffering from EB. After measuring parent and patient-reported hand function with the ABILHAND-Kids questionnaire, they concluded that hand impairment has a negative effect on the quality of life, probably because it interferes with daily activities. The influence of pain due to skin, joint and bone lesions on the quality of life in young patients suffering from epidermolysis bullosa has been assessed during a study conducted between 2011-2014 in Southampton. The research highlighted the prevalence of pain in patients with EB and its negative effect on their own and their families’ lives. Patients’ QoL is impaired because of frequent pain but also due to fright of pain. The authors concluded that on-line materials about psychological interventions for pain management are necessary in order to help patients and caregivers with everyday challenges, which will contribute to improving QoL. A 3-year project, 2015-2018, is currently being conducted in Dublin. The epidemiological and pathophysiological mechanisms of pruritus in patients with epidermolysis Bullosa are being studied. Similar to pain, itch has a profound impact on the quality of life of both patients and their families, for which, up until now there is no effective therapy. The study aims to assess the impact of itch on the quality of life, as well as the pathophysiological mechanisms that lead to therapy-resistance itch in EB, in order to identify new treatment options for this troublesome symptom. Itch can be a source of social embarrassment due to the presence excoriated skin and social scratching. 

Families’ quality of life: 

Taking care of a chronically ill person poses a great deal of stress on family members, phenomenon referred to as the caregiver burden. The family burden in EB children is a heavy one. Caregiver burden can develop in many ways and affect parents both as individuals and as a couple. It can affect areas such as a couple’s marital relationship, their sense of ‘private life’, conversation, the ability to stay close (physically and emotionally), their sex life and it can influence the decision to have more children. Their children’s disease can be a major cause for divorce in some cases. According to a Polish Study, parents of children with EB seem to have a greater need of social support and a lower level of life satisfaction. Tabolli et al. used the Family Strain Questionnaire (FSQ), the Family Dermatology Life Quality Index (FDLQI) and the General Health Questionnaire (GHQ-12) to assess the burden of epidermolysis bullosa on patient’s caregivers. The study concludes that the family burden is associated with the clinical severity of the disease and with the body surface involved, similar to that observed in neurological or oncological diseases. Furthermore, the impact of the burden may vary depending on the EB subtype. Parents of children with junctional EB and recessive dystrophic EB reported a more profound impact on their lives than those of children with EB simplex and dominant dystrophic EB. In order to better assess the parental burden of epidermolysis bullosa regarding medical and socio-economic aspects, a specific 20 item questionnaire has been recently developed: the Epidermolysis Bullosa Burden of Disease (EB-BoD).

Psychological interventions:

It is important for health professionals to help patients define their problem and not to make assumptions about the nature of their problem and the way that it affects them. In doing this, there is a danger of over-emphasizing certain issues while ignoring others that may be more pertinent to the patient. In order to avoid making assumptions, the healthcare giver should allow the patient to explain what the issues are that are worrying him/her, and not assume that patients will be able to “open up” and discuss their feelings immediately; be patient and let them “set the pace”. The patient and his/her problem needs to be approached in such a way that takes into account the complete procedure of their “illness experience.”

Programs providing patients with detailed information about their skin disease, including etiology, therapeutic options, and prognosis, can be helpful in enhancing compliance with treatment regimen. Additionally, psychoeducation directed at educating the patient with regard to common emotional reactions to their skin disease can be helpful in reducing the patient’s sense of isolation. Stress can cause exacerbation of disease and this in turn can cause more stress, leading to a vicious cycle. Relaxation can be effective in reducing arousal levels and hence feelings of anxiety and stress. Relaxation training can be accomplished by numerous techniques, all mainly directed at minimizing sympathetic reactivity and enhancing parasympathetic function.

Included in this group of procedures are progressive muscle relaxation, autogenic training, guided imagery, as well as transcendental and other meditation techniques, such as mindfulness. Breathing exercises and self-talk have also shown to be helpful.

Psychological intervention should initially be targeted towards patients’ symptoms and areas of burden and distress, as for example: pain, itch, feelings of isolation, body image issues, emotional problems, and social difficulties. Some authors suggest that cognitive behavioural therapy (CBT) and hypnosis may prove helpful in controlling pain symptoms. Both are evi-
dence-based interventions and have proven to be effective in other pain related conditions. For the control of itch: hypnosis, biofeedback, meditation, eye movement desensitization and processing (EMDR), and CBT. Based on their findings, Williams et al. (12) suggest that psychological interventions focused on the patient may place the causes of their distress on them, while wider community-focused educational programs could be offered in order to address misinformation and negative attitudes and behaviors in their communities, for example, in their schools. Support groups may also prove helpful as subjects with EB can share experiences and feelings about their condition and thereby reduce their sense of loneliness. Parents can also benefit from support groups as many feel they lack information about their children’s condition. In a group setting, patients are encouraged to understand their own and one another’s difficulties. There are many different modalities of group therapy, including family therapy that can help families develop more functional patterns of organizing and interacting with each other (23).

Conclusions:

Patients with severe forms of epidermolysis bullosa suffer from chronic pain, mutilations, residual scarring and, sometimes, underlying systemic involvement. While some of them, despite severe manifestations, seem to find the resources to cope on their own with their disease, others are profoundly affected on a psychological level, leading to anxiety and depression. In addition, their families and caregivers are also subject of distress (11).

Quality of life evaluation is an important topic in epidermolysis bullosa since it can improve treatment management, as well as to assess new therapeutic modalities in order to help patients and their families to cope better with the disease (12).

A multidisciplinary approach is therefore mandatory, involving dermatologists, psychologists and psychiatrists, working together to support and provide both patients and caregivers with a holistic attention, identifying any sign of emotional distress or mental disorder and addressing it in the best possible manner. In addition, support groups, psychotherapy sessions and organizations such as Dystrophic Epidermolysis Bullosa Research Association (DEBRA) and Epidermolysis Bullosa Medical Research Foundation can help to provide patients and their families with information, psychoeducation and support (11).

Bibliography