

INHERITED EPITHELIAL ADHESION DISEASES: ASSESSMENT OF THE QUALITY OF LIFE IN EPIDERMOLYSIS BULLOSA



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INTRODUCTION. The epithelial adhesion diseases are a rare and genetically heterogeneous group of inherited disorders characterized by skin and mucous membrane fragility, in which blisters and erosions develop in response to minor injury. The quality of life (QoL) of people with epidermolysis bullosa (EB) was assessed (children and adults) taking into account disease severity, extension and localisation, in order to describe physical, social and psychosocial suffering of such patients. The family burden of the disease was also assessed with the involvement of the principal family caregiver.

METHODS. A cross-sectional observational study, with postal questionnaires. 185 patients from IDI-IRCCS Institute (Rome) and Debra ONG databases were contacted (February-March 2008). Disease severity was assessed by patients, using the Patient's Global Assessment (PGA) index (from 0 to 4, corresponding to "very mild", "mild", "moderate", "severe", and "very severe"). Children severity was evaluated by their mothers. Adult patients were asked to complete the Medical Outcome Study 36-item short-form questionnaire (SF-36), the Skindex-29, and the 12-item General Health Questionnaire (GHQ-12). Children (7-14 years) completed the EQ5D questionnaire.

The SF-36 includes 36 items in a Likert-type or forced-choice format, intended to measure the following eight dimensions: physical functioning (PF), role-physical (RP), bodily pain (BP), general health (GH), vitality (VT), social functioning (SF), role-emotional (RE), and mental health (MH). Scores for each domain range from 0 to 100, with higher scores indicating better health. Two additional summary measures, the physical (PCS) and mental component scores (MCS), were also obtained.

The Skindex-29 consists of three scales assessing areas considered essential in any tool intended to evaluate QoL: symptom burden, functioning, and emotional state. Higher values reflect a worse QoL.

The GHQ-12 is a self-administered 12-item questionnaire designed to measure psychological distress and to detect current non-psychotic psychiatric disorders, such as depression and anxiety. Answers are given on a four-point scale; when scored with the binary method (0-0-1-1) the GHQ-12 can be used as a screening tool to detect minor non-psychotic psychiatric disorders, yielding final scores that range from 0 to 12. Operationally, patients scoring 4 or more were considered as "GHQ-positive" (GHQ+).

EQ-5D has a descriptive system and a visual analogue scale (VAS). The EQ-5D descriptive system includes the following 5 dimensions: mobility, self care, usual activities, pain/discomfort and anxiety/depression. Each dimension has 3 levels: no problems, some problems, severe problems. The EQ-5D VAS records the respondent's self rate health on a vertical scale.

All statistical analyses were performed using the STATA statistical package, release 9 (STATA, College Station, TX, U.S.A.).

RESULTS. 185 patients were invited to participate, data were available for 125 patients: 27 (0-7 yrs), 18 (8-14 yrs) and 80 (>14 yrs). Mean ages were 3.1±1.7, 10.2±3 and 33.5±13 respectively. Adult women had lower scores on all SF-36 scales compared to men and all EB patients had lower SF-36 values than normal subjects.

In **FIGURE 1** the comparison of the SF-36 scores of EB patients with the normative score for the general Italian population shows a strong impact of the disease on all scales ($p<0.05$), and differences between males and females. The worst health status was observed for patients who perceived the disease as more severe (**FIGURE 2**) and for those who reported a larger skin involvement.

In **FIGURE 3** we reported values for Skindex-29 (specific dermatologic questionnaire) in males and females: a worse QoL was observed in women. Symptoms referred "all the times or often" were skin sensitivity (58%), itch (51%), burning and stinging (40%); Emotions were to be annoyed (48%), angry (34%) and worried (32%); for Social functioning: the disease makes hard to work 43%, tiredness (31%) and (27%) problems in sex life.

In patients aged 14 or more, 48% of females and 16% of males were GHQ+.

No significant differences were observed for different histological types of EB (**FIGURE 4**).

At EQ-5D children reported a worse mobility and self care compared to adults.

Referring to mothers with children with EB, we identified 19% and 29% of GHQ+ in the groups aged 0-7 and 8-14 years, respectively.

The family burden was higher for emotional aspects, but in comparison to other diseases (e.g. oncological, nephrological, mental, vascular leg ulcers) was lower than expected.

FIGURE 1 SF-36 mean values for EB patients

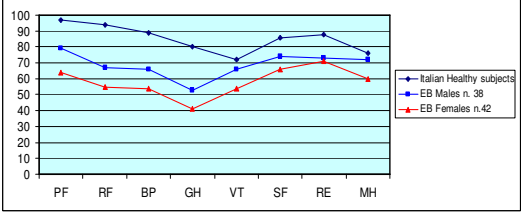


FIGURE 2 SF-36 mean values according to self-evaluated disease severity (adults)

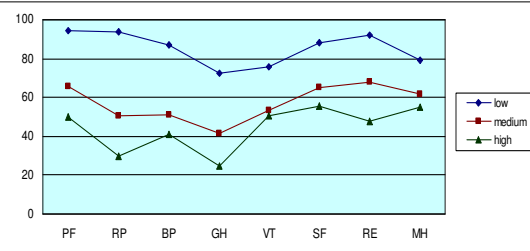


FIGURE 3 Skindex-29 mean values for each scale (adults)

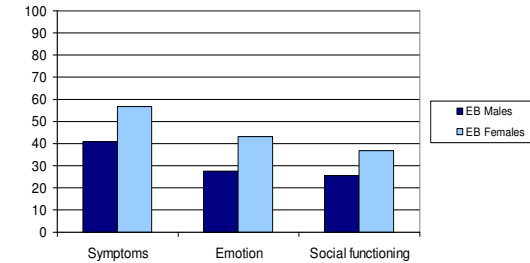
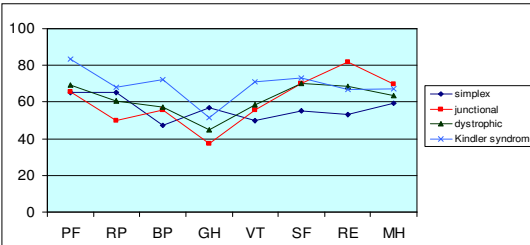


FIGURE 4 SF-36 mean values in different EB histological types (adults)



CONCLUSIONS. We described a strong impact of EB on patients' QoL both for dermatology-specific (Skindex-29) and for general health aspects (SF-36). Both physical and psychosocial aspects of QoL were more impaired in women than in men.

A set of tools with the characteristics outlined above, exhaustively evaluating a number of parameters, would thus provide important additional information for the global assessment of patients' health status. The introduction of QoL evaluation in clinical practice, using a minimum set of generic and specific tools, as proposed here, could help clinicians to select patients who would most benefit from the addition of psychological support to their standard treatment, and to monitor the patients in their care.

These validated, easy to manage and simple to use tools are already widely employed by dermatologists and other physicians. The high percentage of patients at risk of minor psychiatric non psychotic disease strongly support the idea to involve a psychology expert to assist patients and the EB families.

More studies are necessary to identify why the family burden is not high and to study the coping effect in such disease that usually has a neonatal onset.