Quality of life of patients with epidermolysis bullosa

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- Having a vulnerable skin causes many limitations in everyday life and numerous psychological and sociological problems for the patients and their families

- There is a growing need to investigate the patients perspective of the impact of the disease on their lives

HRQoL

- health-related **quality of life** (HRQoL): is an individual perception of how her/his wellbeing is affected over time by a disease, disability, or a disorder and how it affects emotional, social and physical, cognitive, work-related aspects of the individual’s life

- Determination of HRQoL gives information of an individual's physical, emotional health, psychological, social wellbeing, economic burden, fulfilment of ones personal expectations, and functional capacity to carry out daily routines

Studies of QoL in EB

- Published studies are scarce
- usually involve a relatively small number of patients
- variable disease severity
- studies are: often descriptive, use different tools

Evaluation of HR QoL

- **different questionnaires/tools**
  - ✓ Generic instruments
  - ✓ Skin-specific questionnaires: DLQI, Skindex-29 and 17 ...
  - ✓ Disease specific QoL instruments

- **Studies are often descriptive (interview, semi structured interview)**

- **Some studies investigate psychological, functional health status, economical effects of the disease and/or the effect on family**

QoL for EB questionnaires

- Quality of life evaluation questionnaire in EB (QOLEB)- Br J Dermatol 2009;161:1323-30. (translated and validated to Dutch, Brazilian Portuguese, Hispanic)

- EB family/parental burden score questionnaire (EB-BoD)- Br J Dermatol 2015; 173:1405-10.

Evaluation of HR QoL

- **different questionnaires/tools**
  - ✓ Generic instruments: SF-36,VAS....
  - ✓ Skin-specific questionnaires: DLQI, Skindex-29 and 17 ...
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- **Studies are often descriptive (interview, semi structured interview)**

- **Some studies investigate psychological, functional health status (mobility, hand function), economical effects of the disease or the effect on family**

What have we learned

- Similar issues as other chronic conditions–coping with symptom management, disability, emotional impact, need for easy access to good medical care

- Similar issues as other skin diseases–visible and has an influence on social and interpersonal relations

- Specific for EB–is rare, life long and hereditary nature, often disfiguring and painful

QoL of adult/children with EB

- QoL comparable to moderate and severe psoriasis and AD
- The perceived disease severity and skin area involved are relevant for QoL in all EB subtypes
- Greatest scores RDEB-HS
- QoL is more severely impaired in children and women

Clin Experimental Dermatol 2002;27:707-10.
Clin Experimental Dermatol 2004;29:122-7.
Br J Dermatol 2009;161:869-77.
Ann Dermatol 2016;28:6-14.

QoL of adult/children with EB

- Very few patients are free of pain, it varies depending on extent of skin surface involved and depth of wounds
- patients with all subtypes of EB are likely, to at least partially, be dependant on others
- restriction of physical, social and sporting activities which influences QoL
- impact of the disease on employment and education

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Impact of EB on psychological distress (depression/anxiety)

- the physical burden placed on EB patients can lead to depression/anxiety
- psychological suffering and severity of the disease did not correlate in all studies
- Possible explanation: patients get accustomed to living uncomplainingly with the disease and valuate areas of life which remain uncompromised by EB (school, family, friends)

Br J Dermatol 2009;161:869-77.
J Clin Psychol Med Settings 2010;17:333-9.
J Dermatol 2016;43:70-8.

Patients’ perspective

- importance of having comprehensive information about the disease, the exact diagnosis, mode of inheritance
- want to be „in charge” of everyday life
- want to be acknowledged as “partners” in the care process by health care professionals
- problem of interaction with health care professionals who are not well informed about EB
- need to interact with those who understand them – other people with EB

Qualitative Health Res 2011;21(6):771-82.

Childrens’ perception

- Itch (new wounds, feeling of helplessness, impact on sleep)
- constant pain and fear of pain
- difficulties to join in activities with others due to physical limitations
- feelings of being left out and loss of self-esteem
- Stigmatisation – often bothered by the visibility of the disease (annoyed by staring and teasing of others)
- the feeling of being different – wish to be seen as „normal“ and to have an identity unrelated to their skin disease

Acta Derm Venereol 2008;88:143-50.

Implications for support provision

- Informational needs (patients, family, layman, health care providers)
- Need for extensive practical, social and emotional/psychological support
- Availability of multidisciplinary, specialized medical centres
- Need for assistance of parents in seeking support and counselling to prevent destruction of the family unit

Conclusions and aims

- Investigation of the HRQoL is important in care and management of patients with EB – holistic approach
- Can help in assessing new therapies in clinical trials
- Determination of HRQoL and comparison with other diseases can help in fundraising

- Aims

  -enrolment of a greater number of patients, with clinically different disease severity

  – in quantitative studies use disease specific questionnaires

  -development of paediatric QoL tools