Advancing Care in Epidermolysis Bullosa: Insights from Qualitative Research

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Epidermolysis bullosa (EB) concerns a heterogeneous group of rare genetic skin fragility disorders that result in chronic blistering and wounding (1). It is classified into four major types - EB simplex (EBS), junctional EB (JEB), dystrophic EB (DEB), and Kindler EB (KEB) - and further subdivided into 30 subtypes with varying severity. EB profoundly impacts the lives of patients and their families (2, 3). While

advancements in EB diagnostics, such as next-generation sequencing, have led to faster and more precise diagnoses, they also raise questions about the experiences of patients and their family throughout the diagnostic journey.

In this issue of the BJD Korte et al (4) report on the parent and patient perspective regarding the EB diagnostic trajectory. They interviewed 26 participants: 18 parents of 13 paediatric patients and eight adult patients, covering the major EB types except for Kindler EB. Korte et al show that the EB diagnostic process elicits a diversity of emotions, varying from desperation and uncertainty about the future, to clarification and confirmation. Interviewees and particularly parents of children with extensive presentation, emphasized the need for a timely and precise prognosis, underscoring the emotional toll of diagnostic delays. Moreover, both parents and patients with localized forms felt that severity ratings used in EB disease terminology made them feel less deserving of care and attention and inhibited them from seeking medical care or engaging in research or peer support.

By using qualitative research Korte et al gained an in-depth understanding of patients' and parents' lived experiences and needs in a way that was not restricted by predefined answers, revealing insights that may have otherwise been overlooked. Giving voice to those affected by a disease is crucial in understanding illness, offering key areas for improving care. This is particularly relevant in dermatology where most diseases profoundly affect quality of live, without most of the times, being directly life-threatening (5). Fortunately, we see an increasing number of qualitative articles being published in dermatology, with the BJD as the leading platform. Alongside this, a growing body of literature is available to help researchers understand and conduct high-quality qualitative research (5,6,7).

The study of Korte et al highlights that healthcare providers should avoid value-connotated disease terminology including clinical severity labelling of sub-types. EB appears to have unique individual implications that often do not correspond with the currently applied severity labels. The authors also emphasize that EB should not be treated as one entity and call for a distinct approach clustered by subtypes for clinical practice and research. While we acknowledge the differences between EB subtypes, we believe it is also important and potentially more feasible to focus on underlying experiences and needs that transcend individual subtypes and use these insights as a basis for further improving EB care. This approach can reduce black-and-white-thinking and allows for greater personalization of care to individual preferences, including tailored communication about diagnoses. This

may contribute to further advancing EB diagnostic care.

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