

Reference models for children and young people with Epidermolysis bullosa: First case report on the challenges, rewards and limiting factors for successful academic and professional achievements[†]

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Abstract

Epidermolysis bullosa (EB), also known as Butterfly skin, is a term for a heterogeneous group of rare genodermatosis disorders resulting in painful mucocutaneous blisters and extreme fragility with minor trauma or friction. The low prevalence and heterogeneity of EB, clinically classified into four major groups and over 30 subtypes, lead to significant psychological and social research constraints. An inverse correlation between disease severity and quality of life in patients and their families has been described. Psycho-social aspects in people with more severe subtypes of EB and body dysmorphia refer to loss of self-esteem and coping skills, low self-control, intense fear, anxiety, depression, and social isolation. On the other hand, acute, chronic and procedural pain, common comorbidities in EB, can negatively affect cognition and interfere with academic and professional achievements. Here, we present the first case report of a current international project that searches for reference models for children and young people with EB and the creation of a guidebook with different life stories of strategies and skills proven to help achieve academic/professional success despite EB's condition. We analyze the semistructured interview with Sonia Ortiz Romero, alias Mariposa de cristal, an expert patient with dystrophic EB and co-founder of 'Son mis alas, I.A.P' ('They are my wings, NGO'), together with his brother Marcos Humberto. Her storytelling illustrates challenges, rewards, and limiting factors that make their Mexican association a successful case in helping other EB patients in Santiago Tianguistenco with their physical, psychological, and social well-being, as well as raising social awareness.

Keywords

Reference models; Epidermolysis Bullosa (EB); rare diseases; expert patient; dystrophic epidermolysis bullosa (DEB); storytelling; successful cases; education; coping strategies; family and social support.