Making rare skin diseases visible: Lessons from epidermolysis bullosa for patient-centered care in Malaysia

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ABSTRACT

Rare skin diseases are often overlooked in health planning, despite the heavy and lifelong burden they place on patients and families. These are chronic conditions that impact far more than health alone but affecting eating, mobility, education, social participation, and mental wellbeing. Families frequently spend hours each day managing care while facing financial and emotional strain. In Malaysia, people living with rare skin diseases face common challenges: limited access to genetic testing (often only available overseas), high costs of specialised treatments and wound dressings, fragmented care pathways, and low awareness among health professionals. These systemic gaps delay diagnosis, restrict access to appropriate care, and leave families feeling unsupported. Epidermolysis Bullosa (EB), for example, illustrates these realities. EB is sometimes described as a "rare blistering skin disease," where fragile skin and painful wounds dominate daily life. But the struggles of EB patients and caregivers are shared by many families across the spectrum of rare skin conditions. Addressing these needs requires convergence and alignment. Initiatives such as newborn screening, national registries, and clinical protocols must be developed as part of a comprehensive rare skin disease framework. EB offers one example, but the solutions must be integrated, patient-centered, and inclusive for all rare skin disease communities. This presentation will share lessons from EB in Malaysia within the wider rare skin disease context, highlighting patient and caregiver realities and underscoring why rare skin diseases must be recognised in national health strategies to ensure sustainable care, equity, and dignity for families who have long been invisible.

Keywords: Epidermolysis Bullosa, Rare Skin Diseases, Patient Centered Care, Advocacy and Policy, Treatment and Management