

Management of oesophageal strictures in inherited epidermolysis bullosa: a clinical practice guideline.

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T.C., E.P., R.J., M.L.B., J.H., A.C.F.R., C.B., C.R., P.T., L.M.B., S.P.M., A.D., J.A.F., G.Z. and P.D.A. state no conflicts of interest. M.E.H. is a member of the advisory board of Krystal Biotech and a consultant for Chiesi Global Rare Diseases. I.L.-C. is consultant for Abeona Therapeutics and a board member of DEBRA Canada. A.E.M. is a consultant for Amryt Pharma and Krystal Biotech. K.M.-C. claims no financial conflicts of interest; she declared a potential conflict from her professional work coordinating guidelines for DEBRA International between 1 February 2016 and 6 February 2023; therefore, she was not involved in the writing subgroups.

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<https://creativecommons.org/licenses/by-nc/4.0/> This is an Open Access article distributed under the terms of the Creative Commons Attribution-NonCommercial License (<https://creativecommons.org/licenses/by-nc/4.0/>), which permits non-commercial re-use, distribution, and reproduction in any medium, provided the original work is properly cited. For commercial re-use, please contact reprints@oup.com for reprints and translation rights for reprints. All other permissions can be obtained through our RightsLink service via the Permissions link on the article page on our site—for further information please contact journals.permissions@oup.com. Inherited epidermolysis bullosa (EB) is a group of rare and complex genetic disorders characterized by fragility of the skin and mucous membranes. Specifically, the gastrointestinal tract is commonly involved with a range of complications, one of the most disabling being oesophageal strictures (OS). OS manifest with progressive dysphagia, which in turn contributes to malnutrition, chronic anaemia and growth delay, with high impact on the quality of life of patients and their families. DEBRA International has supported the development of clinical practice guidelines (CPGs) for different aspects of EB care. The present CPG aims to provide healthcare professionals and affected individuals and their caregivers with recommendations on diagnostic procedures, preventive measures and treatment of OS. An international multidisciplinary panel comprising clinical experts and patient and public involvement (PPI) representatives developed the CPG, following an international PPI survey and literature review. The GRADE methodology was adopted to define PICO (population, intervention, comparison, outcome) questions, implement a literature appraisal process and prepare recommendations. Three recommendations are focused on OS diagnosis, and seven on preventive nonpharmacological (diet, oral care and therapeutic education) and pharmacological measures (topical corticosteroids) and treatment procedures. Treatment focuses particularly on oesophageal dilatation and how to delay and manage disease relapses. It is expected that this CPG will contribute to improving the quality and equity of care for individuals affected with EB, and will hopefully foster clinical research to increase evidence, in particular on noninvasive OS treatment to prevent complications and delay relapses.

Inherited epidermolysis bullosa (EB) is a group of rare genetic conditions. They present as fragility of the skin and formation of blisters. Oesophageal strictures (OS) are a painful, common and disabling complication in EB. These are narrowing of the food pipe, causing difficulties with swallowing. They contribute to malnutrition and anaemia. Early diagnosis and treatment of OS are essential in people with EB. Early treatment can improve the health and quality of life of people with EB and their families.

DEBRA International is a global association of patients with EB. They appointed 19 experts from seven countries to develop these guidelines. These included EB representatives, clinicians, dietitians and specialized nurses. The aim was to develop guidelines for the diagnosis, prevention and treatment of OS.

The group first prepared an online survey for people with EB and their caregivers. They then analysed the survey findings. Finally, they searched and reviewed the related scientific literature. With this information, the panel identified 10 questions important in the management of OS. Using these questions the panel agreed on the recommendations. These recommendations are for healthcare professionals, people with EB and caregivers.

Three of the recommendations focus on diagnosis. The other seven focus on prevention and treatment. Prevention includes diet, oral care and medications. Treatment includes widening the

gullet and delaying and managing relapses.

We expect that this guideline will improve the quality and equity of care for people living with EB.