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Research Institution:	Bambino Gesù Children’s Hospital, IRCCS, Rome, Italy
Project Title:	Diagnosis and management of Oesophageal strictures in EB patient
Research Area:	Clinical Practice Guidelines
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Project Abstract

Oesophageal strictures are one of most common and serious complications of inherited epidermolysis bullosa (EB), in particular the severe, inverse and intermediate subtypes of recessive dystrophic epidermolysis bullosa (RDEB) and Kindler EB. Oesophageal strictures cause dysphagia and odynophagia, leading to restricted food and drug intake and thus contributing to poor nutritional status. Both oesophageal symptoms and complications significantly worsen the quality of life of EB patients. Strictures develop more frequently in the cervical oesophagus (the upper third part of the oesophagus), and may be solitary or multiple. The incidence of strictures increases with age, with major differences among various EB subtypes (e.g. they develop by age 10 years in severe RDEB, and later in Kindler EB). Treatment options include at first dietary recommendations (e.g. avoiding hard and hot food) and medical therapy for pain and gastroesophageal reflux disease, but are usually insufficient to counteract disease progression. Subsequent treatments comprise oesophageal dilation, gastrostomy, or, very rarely, surgery: the choice depends on patient’s conditions and on country/center specificities.

Oesophageal dilation is at present considered the first line treatment for strictures unresponsive to medical therapy. Recent retrospective studies have compared the outcome of different dilation techniques, their specific complications, and the recurrence rate in EB patients. Together with the existing general guidelines on oesophageal dilation and previous literature in the field, this evidence is crucial to support the development of a clinical practice guideline (CPG) for diagnosis and management of oesophageal strictures in paediatric and adult EB patients (all @subtypes).

This guideline will not specifically address management of gastroesophageal reflux disease (GERD). In the literature, there are a number of regularly updated guidelines from National/International societies on GERD management both in children and adults (some are listed at point 1b of the application), which can be tailored to EB patients. A short chapter on GERD in EB will be included in this CPG.

This CPG will represent a useful tool for health care professionals involved in EB care, optimizing the management of oesophageal strictures. The expected outcome of this CPG is to reduce complications and recurrence rate of oesophageal stenosis and to improve nutritional status, and overall patients' quality of life.

Blog post written about project for website

Clinical practice guidelines (CPGs) are recommendations which have been developed by experts in a disease area to guide clinicians in the treatment of specific conditions. CPGs aim to improve the quality of care received by patients and ensure they are treated appropriately. EB is a rare disease and therefore can be unknown to medical staff, as they may not have come across it before. It is important that CPG's are developed for various procedures and treatments associated with EB to ensure clinicians do not unintentionally cause discomfort or harm to the individual.

DEBRA Ireland recognises the importance of CPGs and has previously collaborated with many international colleagues to fund the creation of CPGs in many indications related to EB. DEBRA Ireland is currently co-funding the creation of two CPGs, one for the treatment of oesophageal strictures and the other for eye care in EB. Below you will find descriptions of both CPGs, which DEBRA Ireland is hopeful will improve the quality of life and level of care the EB community currently receive in these areas, both in Ireland and worldwide. A full list of CPGs for EB is on the [DEBRA International website](#).

Oesophageal Strictures CPG

An oesophageal stricture is an abnormal tightening or narrowing of the oesophagus. This makes carrying food and liquid to the stomach difficult and increases the risk of choking. Oesophageal strictures are one of the most common and serious complications of EB. Development of this guideline is being led by Dr El Hachem and Dr DeAngelis, in Bambino Gesù Children's Hospital, IRCCS, Rome, Italy. It is being designed to outline the most effective ways to diagnose and treat oesophageal strictures in EB.

Quotes we have from Researchers

None

Researcher (s) Bio

None
