ORIGINAL RESEARCH PAPER
Volume - 13 | Issue - 12 | December - 2024 | PRINT ISSN No. 2277 - 8179 | DOI : 10.36106/ijsr
INTERNATIONAL JOURNAL OF SCIENTIFIC RESEARCH

A CASE REPORT OF EPIDERMOLYSIS BULLOSA PRURIGINOSA (EBP) OF ESOPHAGUS IN A YOUNG BOY – RARE CASE REPORT

Gastroenterology

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ABSTRACT

Introduction: Epidermolysis bullosa is a genetically inherited disorder which can be autosomal dominant or recessive in inheritance. It is characterized by skin blistering and scarring after a minor trauma. It affects around 0.2 cases per 1 million people. Epidermolysis bullosa pruriginosa (EBP) is even rare. It can involve ears, eyes, nose, genitalia or gastrointestinal tract (GIT). GIT insult results from the trauma caused by the passage of solid or hot foods. And when the lesions heal, results in scarring with segmental stenosis. **Case:** A 15-year-old boy presented with history of difficulty in swallowing for 6 months, initially for solids followed by difficulty to swallow liquids also. He complained a weight loss of 5kgs in 6 months. Parents gave the history of blistering skin disease since the age of 1 year. Barium swallow was done and showed a long segment stricture of mid and distal esophagus. Skin biopsy and genetic testing were done which were s/o epidermolysis bullosa pruriginosa. Endoscopic CRE balloon dilatation was done and oral budesonide therapy was given after which he showed significant symptomatic improvement. **Conclusion:** In conclusion, we report a very rare case of Epidermolysis bullosa pruriginosa (EBP) presented with mid esophageal stricture treated by CRE balloon dilatation and oral Budesonide therapy. Patient is doing well after 8 months.

KEYWORDS

Epidermolysis bullosa pruriginosa (EBP), Endoscopic CRE balloon dilatation, oral budesonide therapy

INTRODUCTION:

Epidermolysis bullosa (EB) is a group of very rare, genetically and clinically heterogeneous disorders with an estimated half million cases worldwide[1]. Epidermolysis bullosa is caused by mutations in genes coding for components of the cytoskeletal keratin intermediate filaments, cell junctions such as, desmosomes and hemidesmosomes. This contributes to intraepidermal adhesion and dermo.epidermal anchorage of skin and mucous membranes[2][3]. As they involve several components, even a trivial trauma can lead to skin blistering, erosions, and ulceration[4]. This results in patients suffering from many complications like recurrent infection of open blisters, scarring of ruptured blisters and may lead to skin cancers in certain subtypes of EB conferring high morbidity, with a risk of increased mortality due to multi-system pathology[5]. Four major groups are recognized based on the impact of defects caused at the level of involvement within the dermo-epidermal junction. They are 1) Epidermolysis bullosa simplex 2) Junctional epidermolysis bullosa 3) Dystrophic epidermolysis bullosa 4) Kindler epidermolysis bullosa. Inherited epidermolysis bullosa is distinct from epidermolysis bullosa aquisita, a separate, non-inherited, immunobullous disorder characterized by antibodies against type VII collagen[4].

Case Report:

A 15-year-old boy presented with complaints of difficulty in swallowing for 6 months, which was insidious in onset started with solid foods and progressed to liquids. At presentation to OPD, he was able to swallow only saliva (Saeed classification 4 to 1). He also presented with weight loss of 5kgs in 6 months. Parents gave the history of blistering skin disease since the age of 1 year, with blisters usually appearing at sites of excessive stretch and trivial trauma-like elbow, knee and ankle etc. The blisters healed with scarring. Child is born out of a consanguineous marriage he attained all milestones appropriate for his age. External examination of skin showed presence of mix of new blisters with old healed scars at various locations like hands (both front and back), knees, ankles and feet. Blood investigations were unremarkable for except for mild anemia with Hb of 9.1gm% and borderline low serum albumin of 3.3g/dl. Barium swallow (Fig-1) was done which showed a long segment stricture of esophagus in mid and distal esophagus. Provisional diagnosis of Epidermolysis bullosa with mid and distal esophageal stricture was made. Patient was subjected to skin biopsy from blister on thigh region and genetic testing. Methylene blue stained frozen sections of lesional skin showed a small subepidermal blister. A linear band of collagen IV was mapped to roof of the blister indicating the level of split below lamina densa which was suggestive of Dystrophic Epidermolysis Bullosa. Genetic testing was done and revealed homozygous autosomal recessively inherited COL7A1 gene mutation which is diagnostic of epidermolytic bullosa pruroginosa. For symptomatic improvement, patient was posted for controlled radial expansion balloon dilatation at low pressure.

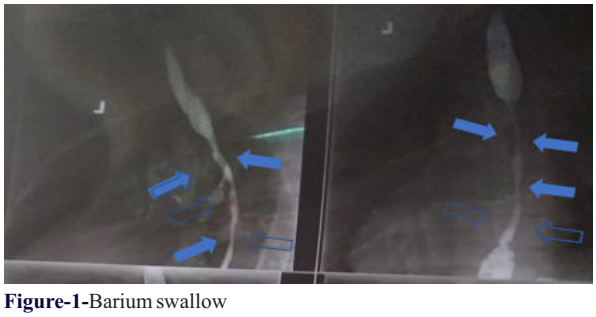


Figure-1-Barium swallow

This is considered as the preferred method of dilatation. Endoscopy is relatively contraindicated as this can result in trauma to mucosa resulting in scar/stricture[6]. Although previously considered as treatment modality for EB-related strictures, linear bougienage dilation is contraindicated in EB – due to risk of causing more mucosal injury and leading to new strictures[6]. Under gastroscopic (Fig-2) and fluoroscopic guidance (Fig-3 to 6) guidewire passed across stricture into the stomach and using 10 mm CRE balloon, serial dilation was

done from distal to proximal. More than two sessions of endoscopic CRE balloon dilatation were done at intervals of 3 months. Post procedure, patient was put on oral steroid (budesonide) and given for 21 days post procedure[7]. Further patient was managed with topical antibiotics, oral budesonide solution[7], vitamin E and iron supplements. He was advised to wear soft padded footwear. Patient on follow up reported a weight gain of 3 kgs over 8 months and required no further sessions of dilatation. Patient is on regular follow up now.

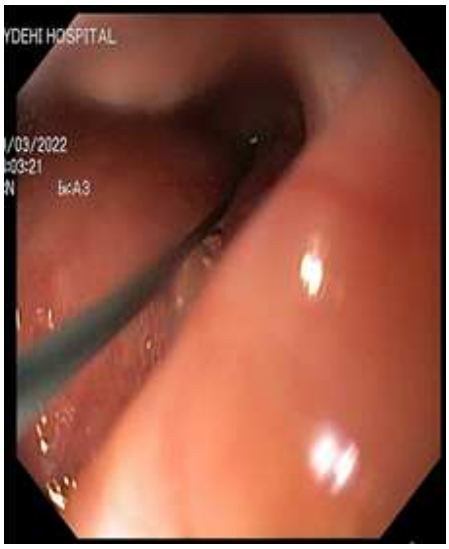
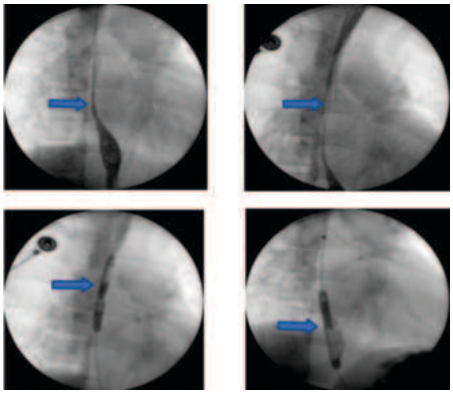


Figure-2 Gastroscopy



Figures-3,4,5,6- Fluoroscopy and CRE Dilatation

DISCUSSION:

Epidermolysis Bullosa Pruriginosa (EBP) is a very rare, distinctive clinical subtype of Dystrophic Epidermolysis Bullosa (DEB) which is included in four distinctive groups of epidermolysis bullosa[8]. Fewer than 100 cases have been reported in the literature. It is caused by mutations in the COL7A1 gene[9] [10] which encodes type VII collagen. Esophageal involvement was described for the first time in 1929. Esophageal and pharyngeal strictures occur in > 65% of patients with the autosomal recessive form of dystrophic epidermolysis bullosa (RDEB) causing severe dysphagia. Dysphagia is first reversible when caused by bullae or webs. Dysphagia later becomes permanent when the inflammatory lesions are replaced by permanent scars and strictures. The treatment of choice for esophageal stricture is the endoscopic balloon dilatation.

Epidermolysis bullosa can be both autosomal dominant or recessive in inheritance majorly affecting the structural part of skin namely dermis, epidermis and basement membrane. Different type of EB will have different proteins involved in the pathogenesis like keratin 5/14, pectin, bullous pemphigoid antigen and plakophilin-1, Desmo plakin, plakoglobin, laminin 332, collagen XVII, Specifically Collagen VII for Dystrophic variant of EB(DEB) as in case of our present patient and Kindlin-I for Kindler type of EB (KEB).

Clinical Features:

EBS is divided into cutaneous and extra cutaneous manifestations. Cutaneous manifestations are in the form of blisters (Fig-7) erosions, milia, violaceous scarring (Fig-8,9), dystrophy of nails, lichenified plaques or prurigo like lesions. Few subtypes have pathognomic findings like the JEB type which has exuberant granulation tissue as pathognomic finding. Some other types of findings like albopapuloid lesions or aplasia cutis is not restricted to one subtype[11]. Here the subtype of interest, the dystrophic variety is divided based on type of inheritance into two sub variants- dominant and recessive variety[12]. Recurrent blisters and erosion leading to esophageal scarring and dysphagia is common in this dominant variant[13]. The subtype of interest, the dystrophic EB pruriginosa is a more generalized disease with intense pruritus. Recessive variant is usually severe in presentation with generalized blistering leading to scarring of skin with retardation of growth and blistering and scarring of cornea, which can also present with anemia, failure to thrive and esophageal strictures[13].

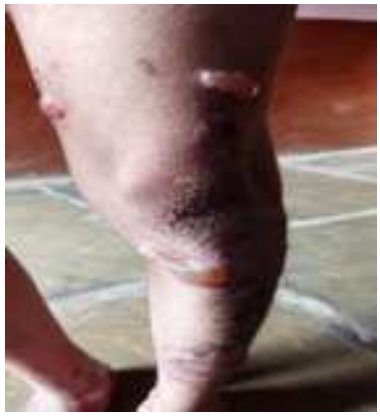


Figure-7- Blisters

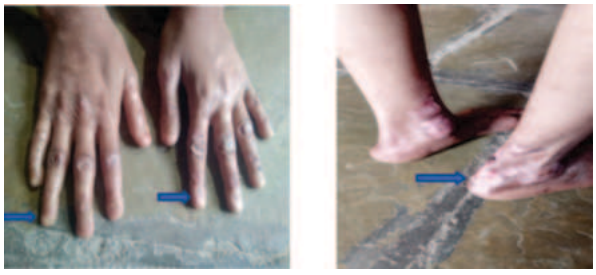


Figure-8,9- Scarring

Diagnosis :

Subtypes of EB are diagnosed based on their type of inheritance, electronic microscopic findings, immunohistochemistry and presenting phenotypic findings[12]. Subclassification of EB is done by mutational analysis of DNA mainly reserved for prenatal diagnosis. Prenatal diagnosis is done by either transmission electron microscopy (TEM) or immunofluorescence antigenic mapping (IAM) of skin sampling with help of ultrasound guided technique which is done either on or after 17th gestational week[14].

Management:

Management mainly focuses on prevention and early recognition along with prompt treatment of symptoms. This is done in the form of offering protective padding and avoiding trauma. Early recognition of complications like corneal disease and corneal scarring by regular ophthalmology follow up. Esophageal strictures are recognized and treated early by dilatation or gastrostomy feeding to maintain good nutrition[15].

New therapeutic approaches are being explored like gene replacement mainly for recessive variant[16], allogenic fibroblast transplant[17]. Various therapies like downregulation of negative genes are being explored for dominant variety.

Abbrevations:

Epidermolysis Bullosa: EB; EB simplex: EBS; ; EBS; Junctional EB: JEB; JEB; Dystrophic EB: DEB; Dominant dystrophic EB: DDEB; Dystrophic EB; Recessive dystrophic EB: RDEB; RDEB; Transmission electron microscopy: TEM; Immunofluorescence antigenic mapping: IAM;

REFERENCES

1. Fine JD, Bruckner-Tuderman L, Eady RAJ, Bauer EA, Bauer JW, Has C, et al. Inherited epidermolysis bullosa: Updated recommendations on diagnosis and classification. J Am Acad Dermatol. 2014 Jun;70(6):1103–26.
2. Bruckner-Tuderman L, McGrath JA, Robinson EC, Uitto J. Progress in Epidermolysis Bullosa Research: Summary of DEBRA International Research Conference 2012. J Invest Dermatol. 2013 Sep;133(9):2121–6.
3. Uitto J, Bruckner-Tuderman L, McGrath JA, Riedl R, Robinson C. EB2017—Progress in Epidermolysis Bullosa Research toward Treatment and Cure. J Invest Dermatol. 2018 May;138(5):1010–6.
4. Bardhan A, Bruckner-Tuderman L, Chapple ILC, Fine JD, Harper N, Has C, et al. Epidermolysis bullosa. Nat Rev Dis Primer. 2020 Sep 24;6(1):1–27.
5. Fine JD, Mellerio JE. Extracutaneous manifestations and complications of inherited epidermolysis bullosa: Part II. Other organs. J Am Acad Dermatol. 2009 Sep 1;61(3):387–402.
6. Anderson BT, Feinstein JA, Kramer RE, Narkewicz MR, Bruckner AL, Brumbaugh DE. The Approach and Safety of Esophageal Dilation for Treatment of Strictures in Children with Epidermolysis Bullosa. J Pediatr Gastroenterol Nutr. 2018 Dec;67(6):701–5.
7. Kianifar H, Talebi S. The effect of oral budesonide liquid therapy to alleviate clinical symptoms in patients with epidermolysis bullosa. Rev Clin Med. 2022 Dec 1;9(4):156–8.
8. Fine JD, Bruckner-Tuderman L, Eady RAJ, Bauer EA, Bauer JW, Has C, et al. Inherited epidermolysis bullosa: Updated recommendations on diagnosis and classification. J Am Acad Dermatol. 2014 Jun 1;70(6):1103–26.
9. Yang CS, Lu Y, Farhi A, Nelson-Williams C, Kashgarian M, Glusac EJ, et al. An Incompletely Penetrant Novel Mutation in COL7A1 Causes Epidermolysis Bullosa Pruriginosa and Dominant Dystrophic Epidermolysis Bullosa Phenotypes in an Extended Kindred. Pediatr Dermatol. 2012 Nov;29(6):725–31.
10. Shi BJ, Feng J. A novel missense mutation in the COL7A1 gene causes epidermolysis bullosa pruriginosa. Clin Exp Dermatol. 2009 Dec;34(8):e975-978.
11. Wong WL, Pemberton J. The musculoskeletal manifestations of epidermolysis bullosa: an analysis of 19 cases with a review of the literature. Br J Radiol. 1992 Jun;65(774):480–4.
12. Fine JD, Eady RAJ, Bauer EA, Bauer JW, Bruckner-Tuderman L, Heagerty A, et al. The classification of inherited epidermolysis bullosa (EB): Report of the Third International Consensus Meeting on Diagnosis and Classification of EB. J Am Acad Dermatol. 2008 Jun;58(6):931–50.
13. Fine JD, Johnson LB, Weiner M, Suchindran C. Gastrointestinal Complications of Inherited Epidermolysis Bullosa: Cumulative Experience of the National Epidermolysis Bullosa Registry. J Pediatr Gastroenterol Nutr. 2008;46(2):147–58.
14. Eady RA, Gunner DB, Tidman MJ, Nicolaides KH, Rodeck CH. Rapid processing of fetal skin for prenatal diagnosis by light and electron microscopy. J Clin Pathol. 1984 Jun;37(6):633–8.
15. Haynes L. Nutritional support for children with epidermolysis bullosa. Br J Nurs [Internet]. 2013 Sep 27 [cited 2024 Dec 1]; Available from: https://www.magonlinelibrary.com/doi/10.12968/bjon.2006.15.20.22292
16. Mavilio F, Pellegrini G, Ferrari S, Di Nunzio F, Di Iorio E, Recchia A, et al. Correction of junctional epidermolysis bullosa by transplantation of genetically modified epidermal stem cells. Nat Med. 2006 Dec;12(12):1397–402.
17. Tolar J, Ishida-Yamamoto A, Riddle M, McElmurry RT, Osborn M, Xia L, et al. Amelioration of epidermolysis bullosa by transfer of wild-type bone marrow cells. Blood. 2009 Jan 29;113(5):1167–74.