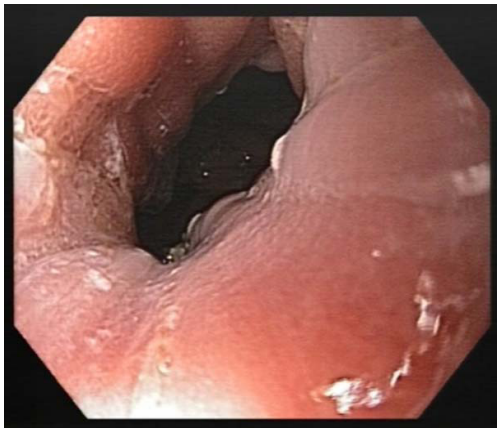

Figure 4A: Healed leak site

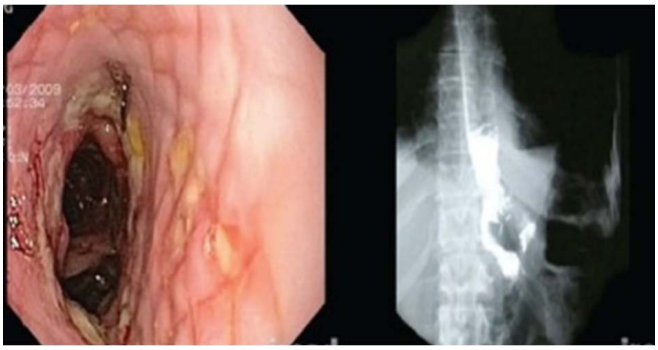

Figure 4B: C-CEMS removal,& Healed leak site on radiography

Double-type 12 mm x24 mm from Niti's, Taewoong^{R} was placed (**Figures 3A and B**). After twenty-seven days the patient was followed up and C-SEMS was removed.Endoscopy and esophagogram showed healing of the leak site (**Figures 4A and B**).

Discussion

Oesophageal or gastric anastomotic leakage, perforation, staple line dehiscence or trauma can be a life-threatening situation. There is a lack of standard protocol for their treatment. The spectrum of treatments suggested span from aggressive surgical re-exploration and repair or even disassembly of the anastomosisto conservative treatment using total parenteral nutrition, peri-anastomotic drainage with chest or abdominal image-guided percutaneous drainage and broad spectrum antibiotics in selected patients. Re-operation is often not easy and associated with high morbidity and mortality. Enteral feeding started early during the treatment plays a significant role in leak closure.In case of post-surgical leaks, with the surgical sutures holding the partially opened anastomosis or staple line, we believe that the SEMS should be left in place for a minimum possible duration, but at least for 2-3 weeks, because of the potential threat of SEMS putting some tension on the suture anastomosis, thus compromising wound healing.Stenting with SEMS seems to be a feasible option as a primary care modality for patients with post-operative foregut leaks.

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Ellis Van Creveld syndrome with mesenteroaxial volvulus – expanding the spectrum of endodermal involvement

Introduction

Ellis-van Creveld is a rare autosomal recessive disorder with

ecto and mesodermal defects.[1] The extent of endodermal involvement in this syndrome is not well established. However there have been reported instances of endodermal involvement in the liver, lung, pancreas and central nervous system.[2-4] The aetiology of gastric volvulus is thought mainly idiopathic, but congenital and acquired associations have also been implicated.[5] Given the strong idiopathic preponderance, and genetic associations; gastric volvulus could occur because of aberrations during endoderm organogenesis. Hedgehog (Hh) signalling plays several important roles during endoderm organogenesis.[6] Hh signalling is one of the critical pathways involved in the development and homeostasis of the mammalian gastrointestinal tract.[7,8] Hh signal is transduced by Smoothened (Smo) forming a complex with EVC/EVC2.[9] Recent study has shown that EVC2 also acts as a positive regulator of the Hh signalling pathway.[10] Furthermore, the genetic and cellular mechanisms involved in the regulation of Hh signalling in the development of mammalian gastrointestinal tract are emerging.[11] This case represents the first instance of gastrointestinal involvement and may thus represent the expanding spectrum of endodermal involvement in EVC.

Case report

A 14-year-old Asian girl with Ellis Van Creveld (EVC) syndrome presented to our hospital for correction of her musculoskeletal deformities. She was the only living child of a healthy non-consanguineous couple. She was born through normal vaginal delivery at term with birth weight 3.25 kg. Limb anomalies were identified at birth. Another pair of twins with similar musculoskeletal deformities was born to the same couple but died at 4 and 50 days of life.

She achieved normal neuropsycomotor milestones. She attended school till a year ago when the progressive genu valgum causing gait disturbance prevented her from doing the same. She presented to our hospital for treatment of her musculoskeletal anomalies. Physical examination was as follows: height 154 cm and weight 60 kg. Sparce scalp and body hair with hirsuitism. High arched palate with a bifid uvula and conical sharp peg shaped teeth. Bilateral cubitus valgus with broad hands, brachydactyly, polydactyly and syndactyly. Bilateral genu valgum with flat feet. The diagnosis of Ellis Van Creveld syndrome was made based on the above features. Echocardiogram and ultrasound abdomen were normal.

She underwent corrective osteotomies for bilateral genu valgum. In the post operative period she developed non bilious vomiting with upper abdominal pain. She was initially treated conservatively, but the episode did not resolve. She had history of similar episodes in the past. Upper gastrointestinal series findings demonstrated a mesenteroaxial volvulus of the stomach (**Figure 1**). The stomach was decompressed using a nasogastric tube and after gaining an informed consent she underwent a laparoscopic gastropexy. The stomach was found folded below the left lobe of the liver with the distal half congested. The absence of diaphragmatic hernia, splenic anomalies, hiatus hernia and malrotation were confirmed. An anterior gastropexy was performed by placing three interrupted non absorbable sutures along the greater curvature of the stomach. At discharge she was pain free and tolerating solids orally.

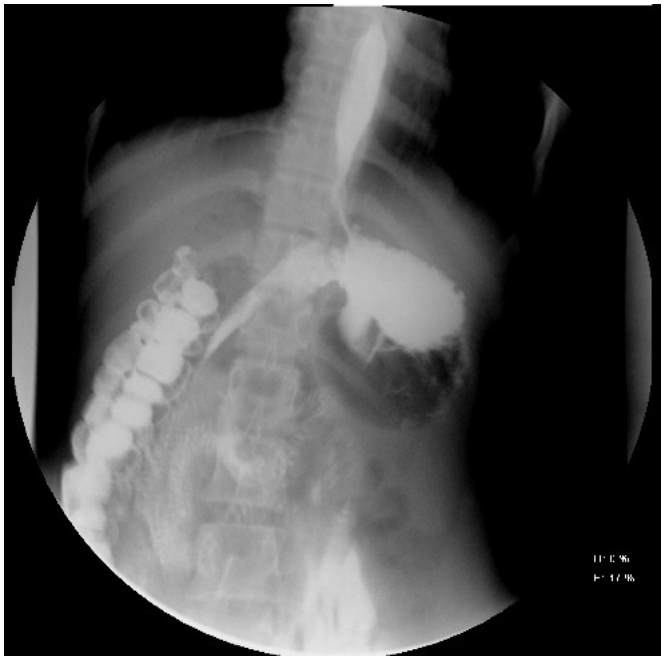

Figure 1: Barium study showing a mesenteroaxial volvulus

Discussion

EVC syndrome is a rare disorder with an incidence of 1 in 60,000 live births.[1] It is an autosomal recessive disorder due to mutations of the EVC and EVC2 genes on chromosome 4p16.[12-13] Till date, more than 41 independent EVC mutations have been described.[14] The features of the syndrome include chondrodystrophy, polydactyly, ectodermal dysplasia, and cardiac defects. However, the clinical presentation is variable, the full spectrum is not necessarily seen in every patient.[15] Rare associated anomalies involving the pulmonary, renal,

hepatic, pancreatic, and central nervous systems have been reported previously which underscore the definite endodermal involvement.[2-4] However, the extent of endodermal involvement in this syndrome is not well established.

To the best of our knowledge this is the first reported case of EVC syndrome to include gastric volvulus. Considering other rare anomalies associated with EVC involving the endoderm, gastric volvulus seen in this patient may represent an extension of endodermal involvement. We believe that the accretion of evidence for the endodermal involvement in EVC syndrome may have a mechanistic basis. The aetiology of gastric volvulus has a strong idiopathic predominance.[5] Hedgehog (Hh) signalling plays several specialized roles during endoderm organogenesis.[6] In particular, Hh signalling is also pivotal for the embryonic development and homeostasis of the mammalian gastrointestinal tract.[7,8] A recent study has shown that EVC/EVC2 acts downstream of Smoothened (Smo) and transduces signal to Gli by antagonizing Sufu.[9] EVC2 also acts as a positive regulator of Hh signalling pathway.[10] In addition, Hh signalling has various specialized paracrine functions during embryogenesis of stomach and intestine.[7] Considering the ubiquity of Hh signalling in embryogenesis, the mutated EVC/EVC2 protein in EVC syndrome could lead to wider dysregulation of Hh signalling and its effects may not be restricted entirely to chondrocytes. Therefore, genetic and functional dissection of EVC syndrome with endodermal involvement may provide novel insights into molecular mechanisms to elucidate the aetiology of gastrointestinal diseases with idiopathic preponderance.

In conclusion, gastric volvulus may be a coincidental occurrence or may represent an unusual EVC syndrome association. Hence, this work may serve as a hypothesis generating paper to ascertain the influence of EVC protein mediated regulation of Hh signalling in endodermal involvement, particularly in gastrointestinal diseases with idiopathic preponderance.

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