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Case Reoprt

An undetected Cornelia De Lange Syndrome: A rare cause of erosive oesophagitis in an adult

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Abstract: Cornelia De Lange Syndrome is a rare syndrome with multiple congenital abnormalities, characteristic facial appearance, prenatal and postnatal growth delay, behavioral changes and numerous gastrointestinal manifestations, out of which gastroesophagial reflux disease (GERD) contributes to high degree of disease burden. The syndrome rarely persists into adulthood. Though isolated diagnosis of GERD in a young adult in a general medical ward is a common encounter, arriving at a syndromic diagnosis considering the detailed history, clinical features and endoscopic evidence of severe reflux oesophagitis, in a 26 year old male who presented with recurrent bouts of haemetemesis, highlights the importance of pattern recognition; an important tool in clinical diagnosis even in modern day practice.
Keywords: Cornelia De Lange Syndrome, Gastroesophagial reflux disease,

INTRODUCTION

Erosive oesophagitis is a common clinical entity in a general medical set up, which is most often caused by reflux oesophagitis due to loss of lower oesophagial sphincter tone. However diagnosis of infectious oesophagitis such as fungal oesophagitis, herpes oesophagitis is particularly in rising trend among elderly population due to age related immune suppression, chronic medical illnesses as well as due to iatrogenic reasons. Though oesophagitis due to systemic illnesses like scleroderma, Behcet disease, inflammatory bowel disease and graft versus host disease record few cases, such autoimmune and systemic vasculitic conditions become important in the differential diagnosis among the young age group with severe refractory oesophagitis. We describe on a 26 year old male patient who presented with recurrent episodes of haemetemesis and symptomatic anaemia. An upper gastrointestinal endoscopy confirmed erosive oesophagitis, on detailed clinical evaluation found to have multiple developmental abnormalities with characteristic facial appearance which fits into the clinical diagnosis of Cornelia De Lange Syndrome.

CASE PRESENTATION

A 26 year old male, who had marked psychomotor retardation and speech deficit since childhood was transferred from a District General Hospital for further evaluation of recurrent haemetemesis and symptomatic microcytic anaemia. He was the 4th child of a non-consanguinous marriage and has had an uneventful birth and perinatal history. Apart from left sided inguinal hernia repair which he underwent at the age of 2 years he had an uneventful past medical and surgical history.

His current presentation was due to recurrent bouts of large volume haemetemesis, malena with associated dyspeptic symptoms. Clinical examination on admission was significant for severe pallor, glossitis and angular stomatitis suggestive of iron deficiency anaemia, however apart from anaemia he did not had any peripheral stigmata of chronic liver cell disease or any other features to suggest a cause for haemetemesis. His abdominal examination was unremarkable. However on detailed examination he was found to be having short stature and very thick sharply defined eye brows which meet in the midline, low anterior and posterior hairlines, micrognathia together with pectus excavatum. On admission to the local hospital he was in a hemodynamically compromised state with a PR of 96/min and BP-100/70mmHg. Preliminary laboratory results recorded a hemoglobin value of 5.4g/dl, mean corpuscular volume of 73fl with normal platelet count and coagulation profile. His baseline liver biochemistry and renal profile were well within normal limits. In an acute setting patient was resuscitated with multiple blood transfusions and managed with intravenous proton pump inhibitors (PPI) and his hemoglobin was optimized to 8g/dl. Subsequently he underwent an

urgent upper gastrointestinal endoscopy (UGIE). The findings of UGIE were consistent with severe reflux oesophagitis with linear ulcerations (Figure.1), and patient was continued with PPI therapy.

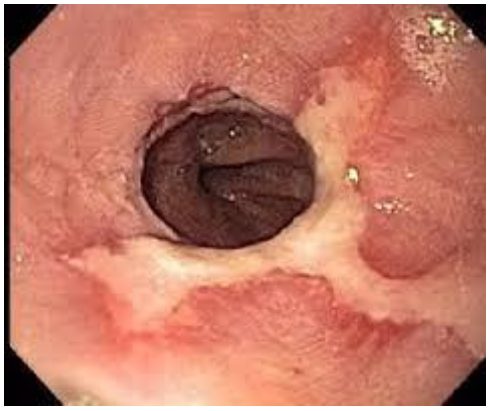

Fig-1: Endoscopic view of severe reflux oesophagitis with linear ulcerations

DISCUSSION

Cornelia De Lange syndrome is a syndrome with multiple congenital abnormalities characterized by a distinct facial appearance, prenatal and postnatal growth deficiency, feeding difficulties, psychomotor delay, behavioral problems and associated abnormalities that primarily involve upper extremities [1]. Most often it occurs in sporadic pattern however occasionally it is transmitted as an autosomal dominant as well as an autosomal recessive pattern [1]. Though heterozygous mutations in a gene named NIPBL, the human homolog of the Drosophila melanogaster Nipped- B gene, have been identified in 50% of cases the exact pathophysiological role of this altered protein is not well understood.

Gastrointestinal manifestations in Cornelia De Lange Syndrome (CdLS) are very common and include anatomical abnormalities, gastroesophagial reflux, constipation and feeding difficulties [2]. Gastroesophagial reflux disease is a very common gastrointestinal problem in CdLS observed frequently in early childhood [3]. The clinical presentation can range from typical features of GERD to atypical presentations like severe behavioral disturbances due to inability to express their discomfort to others verbally. The above clinical presentation elaborates on a patient who presented in compensated shock state following recurrent bouts of haemetemesis due to severe reflux oesophagitis. However the features of chronic iron deficiency anemia can be explained by chronic upper gastrointestinal blood loss due to asymptomatic reflux oesophagitis which is common in patients with CdLS as well as due to nutritional deficiency and chronic worm infestation which is a common occurrence in this part of the world. Hence silent reflux is more common in patients with CdLS and can lead to Barrets oesophagus due to chronic exposure to acid reflux, increasing the risk of adenocarcinoma of the oesophagus. Thus patients with CdLS needs careful monitoring for GERD. Patients diagnosed with GERD need regular medical therapy and continuous surveillance for occurrence of this pre-malignant condition.

CONCLUSION

Cornelia De Lange Syndrome is a rare syndrome with multiple congenital malformations, perinatal and postnatal developmental delay, characteristic facial appearance, behavioural abnormalities and multiple gastrointestinal manifestations detected usually in paediatric practice. It rarely presents in an adult with acute upper gastrointestinal bleeding. The application of basic stepwise approach which includes detailed history, temporal flow, thorough examination and correlating the findings to form a clinical pattern lead to a syndromic diagnosis of this rare entity in an adult.

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