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Johanson-Blizzard syndrome presenting as chronic diarrhoea

Introduction

Johanson Blizzard syndrome (JBS) was described in 1971 by Johanson & Blizzard.¹ It is an autosomal recessive disorder characterized by exocrine pancreatic insufficiency, absent or hypoplastic alae nasi, ectodermal scalp defect, microcephaly, congenital deafness and growth retardation.^{2,3} Only two cases have been reported in the literature from India till date.^{4,5} We here report a rare case of JBS who presented to us with chronic diarrhoea and failure to thrive.

Case History

A 5-months old female child presented to us with a history of not gaining weight accompanied by diarrhoea since the neonatal period. She also had history of pedal edema over the past two months. The child was born out of consanguineous marriage and previously male child with dysmorphism had expired during the neonatal period. The birth history was uneventful, patient cried immediately after birth and her birth weight was 2.8 kg. She had global developmental delay.

On physical examination, the child was lethargic with poor muscle tone and had severe pallor. Her weight was 2.9 kg (< 3rd percentile), length was 49 cm (< 3rd percentile) and head circumference was 34 cm (< 3rd percentile) suggestive of microcephaly. Her anterior fontanel was wide open. There was absence of alae nasi, parrot beak nose, low set ear, patchy alopecia with irregular coarse hair over scalp (**Figure 1**). Pitting edema was also detected feet on physical examination.

Investigations revealed Hb 4.2 g/dL, WBC $17.1 \times 10^3/\mu\text{L}$,



Figure 1: Absence of alae nasia and irregular coarse hair over scalp in child with JBS

platelets $208 \times 10^3/\mu\text{L}$, CRP 11.4 mg/L, ALT 45U/L, AST 32U/L, bilirubin 0.8 mg/dL, protein 4.8 g/dL, albumin 2.3 g/dL, urea 11 mg/dL, creatinine 1.1 mg/dL, Na 149 mEq/L, K 3.1mEq/L and Ca 7.5 mg/dL. Serum amylase and lipase were low. Thyroid function test showed low T_4 and elevated TSH suggestive of hypothyroidism. Whole body X-ray, USG abdomen, CT scan brain and echocardiography were normal.

Genetic study and BERA were not performed, as the parents were neither willing nor could afford the same. She was treated with antibiotics, and thyroxin supplementation, with plan to start pancreatic supplementation. The patient was discharged against medical advice on the 3rd day of admission and died at home as confirmed telephonically.

Discussion

The most common complication of JBS is malabsorption syndrome due to exocrine pancreatic insufficiency, which may lead to chronic diarrhoea, anaemia, hypoproteinemia, oedema and infection, ultimately causing death.⁶ There is no definitive treatment for JBS. Management of JBS is symptomatic and supportive.

JBS can be diagnosed early by antenatal USG detecting aplasia of alae nasi in suspected couples of consanguineous marriage.⁷ Early diagnosis helps in timely management of pancreatic insufficiency and hypothyroidism. The present case highlights the importance of genetic counselling and antenatal diagnosis as this syndrome is autosomal recessive and chances

of recurrence are 25% in subsequent offerings.

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Endoscopic ultrasound detection of a parathyroid adenoma in a patient with chronic pancreatitis due to hyperparathyroidism

Introduction

A proportion of patients will have no identifiable cause of their chronic pancreatitis and are diagnosed with idiopathic chronic

pancreatitis. In this report, we discuss a case of primary hyperparathyroidism presenting as chronic pancreatitis to the gastroenterologist.

Case report

A 27-year-old male was referred to a tertiary gastrointestinal hospital for the complaint of recurrent epigastric pain of 4 months duration. He had been evaluated at another hospital and was diagnosed with chronic pancreatitis based on a CT scan. He had elevated serum amylase and lipase at the time of initial presentation. The cause of his chronic pancreatitis was presumed idiopathic, as he was a teetotaler and did not have gallstones or hypertriglyceridemia.

We evaluated the pancreas and biliary tree with endoscopic ultrasound. A radial echoendoscope (GF-UE160, Olympus Optical Co, Ltd, Tokyo, Japan) was inserted with the patient under moderate sedation. The pancreas showed diffuse hypoechoic strands, echogenic foci, honeycombing and lobularity (**Figure 1**). The pancreatic duct was irregular, but not dilated. There were no pancreatic ductal stones or pseudocysts. The stack sign was observed from the duodenal bulb, suggesting that pancreas divisum was unlikely. Examination of the biliary tree and gallbladder did not reveal any microlithiasis. These features were consistent with a diagnosis of chronic pancreatitis.

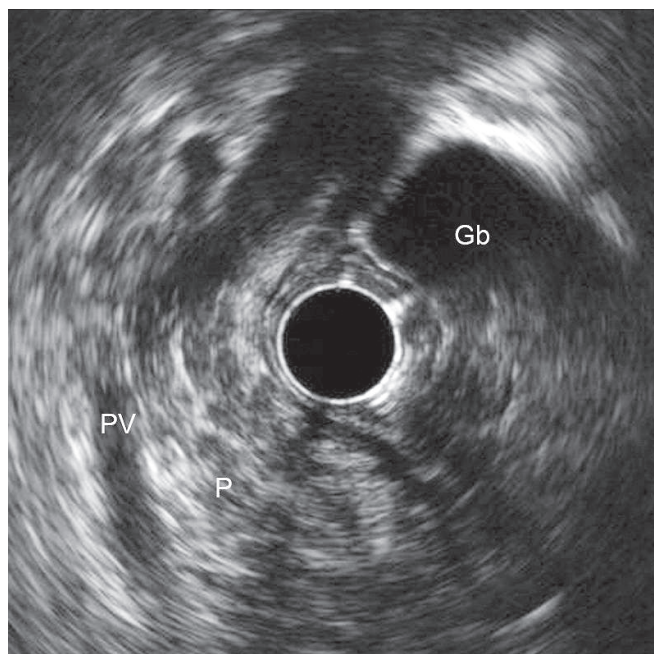


Figure 1: Radial EUS image from the duodenal bulb. The pancreatic head (P) shows lobularity and echogenic stranding suggestive of chronic pancreatitis. The portal vein (PV) and gallbladder (GB) are normal.