Case Report

Complete agenesis of dorsal pancreas: a rare cause of diabetes mellitus

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ABSTRACT

Agenesis of dorsal pancreas is a rare congenital anomaly that results from defective development of pancreas with less than 100 cases described in the literature. Patients may present with epigastric pain, acute or chronic pancreatitis and various disorders of glucose metabolism, can even be asymptomatic. About half of affected individuals develop diabetes mellitus, resulting from reduced islet cell mass. Being very rare, it is generally not kept in mind while dealing these cases and not suspected until imaging investigations are done. In our case study, 14-years-old twin brothers presented with polyuria and polydipsia for seven days. During evaluation diabetes mellitus with dorsal pancreatic agenesis was detected in both brothers.

Key words: *agenesis of dorsal pancreas, diabetes mellitus.*

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INTRODUCTION

Agenesis of the dorsal pancreas (ADP) is an exceedingly rare anomaly that is caused by a defective formation of the pancreas.[1] The dorsal pancreatic bud fails to develop properly during embryonic development. As a consequence, the body and tail of the pancreas are absent, leaving only a small, round pancreatic head adjacent to the duodenum. The size of the remaining pancreas varies and there is a small duodenal papilla and a remnant of the duct of Santorini. Patients may present with epigastric pain, acute or chronic pancreatitis and various disorders of glucose metabolism, even can be asymptomatic. About half of affected individuals develop diabetes mellitus (DM), resulting from reduced islet cell mass.[2] Since its initial documentation in 1911, only a few cases have been documented in published literature. Here we report a case of twin brothers who presented with DM at 14 years of age. Agenesis of the dorsal pancreas was discovered during investigation.

CASE REPORT

Fourteen-year-old twin brothers, 2nd issue of their nonconsanguineous parents presented with the complaints of generalized weakness, polyuria and polydipsia for 7 days. They had no history of fever, vomiting, diarrhea, abdominal pain, weight loss, burning sensation during micturition, loss of appetite, respiratory distress or trauma. They were born at term with normal birth weight and their postnatal period and development were normal. There was no positive family history of DM in their family.

On examination they were well alert, vitals were normal, dehydration was absent, anthropometry was age appropriate, hyperglycaemia, acetonuria and glycosuria were present in both the children. Systemic examination revealed no abnormality. On investigations there was high HbA1c low C-peptide and there was no metabolic acidosis. Serum electrolytes, urea and creatinine were normal in both patients.

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Table 1. Investigation profile of the cases

Investigations	Twin 1	Twin 2
CBG	20 mmol/L	22 mmol/L
Urine for sugar	++	++
Urine for acetone	+	+
Blood gas		
pH	7.35	7.38
PCO2	40.9 mmHg	42.9mmHg
PO2	39 mmHg	40mmHg
HCO3	22.1 mmol/L	23mmo/L
BE	-2.7 mmo/L	-2.9 mmo/L
HbA1 (<6.5%)	18.1 %	19.7%
C-peptide (0.7-3.6ng/ml)	0.42 ng/ml	0.45 ng/ml
S. Amylase (upto 100U/L)	31 U/L	43U/L
S. Lipid profile		
Cholesterol (<200mg/dl)	218 mg/dl	250 mg/dl
HDL(>45mg/dl)	52mg/dl	56 mg/dl
LDL(<100mg/dl)	139mg/dl	168mg/dl
TG(50-150mg/dl)	143mg/dl	138mg/dl
S.TSH (0.36-6.0uIU/ml)	2.2 uIU/ml	2.8 uIU/ml
FT4 (9.4-23.18pmol/L)	13.7 pmol/L	12.22pmol/L
USG of whole abdomen	Suggestive of pancreatic agenesis	Suggestive of pancreatic agenesis Well defined anechoic cystic area (46x36mm) behind stomach
MRI of Abdomen with MRCP	Suggestive of Pancreatic agenesis	Suggestive of Pancreatic agenesis Septated cystic lesion possibly nuroenteric / duplication cyst

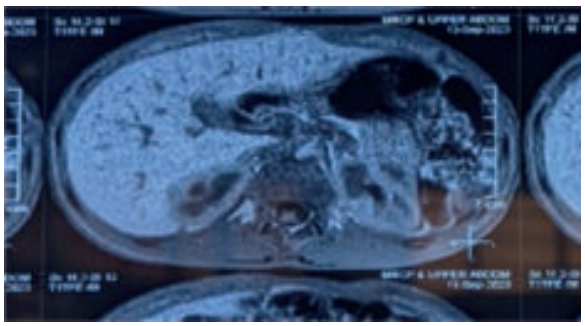

Figure 1. Pancreatic agenesis

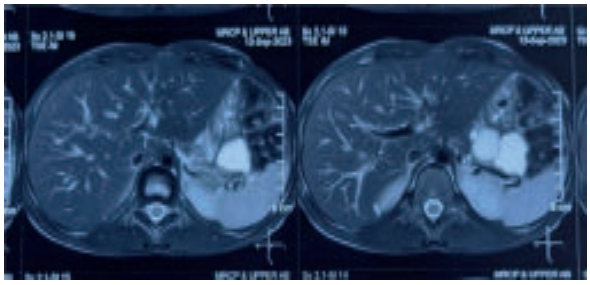

Figure 2. Pancreatic agenesis with Septated cystic lesion possibly nuroenteric /duplication cyst

DISCUSSION

The pancreas develops from the fusion of the ventral and dorsal buds during the seventh week of gestation. The ventral bud is responsible for the development of the Wirsung duct, the most inferior portion of the head and the uncinate process. On the other hand, the dorsal bud, which is connected to the minor papilla through the Santorini duct, gives rise to the upper head, body and tail of the pancreas.[1, 2] Agenesis of the ventral pancreas and complete agenesis of the pancreas are both fatal and not compatible with life.[3]

The exact cause of dorsal pancreatic agenesis is not yet under stood. The mode of inheritance may be autosomal dominant, autosomal recessive or X-linked dominant. Several genes are identified to be reportedly associated with dorsal agenesis of the pancreas.

Pancreatic and duodenal homeobox 1 (*PDX1*) is found in the pancreatic endodermal buds. A defect in this gene will result in insulin requiring hyperglycemia secondary to exocrine pancreatic insufficiency. Mutation involving transcription factor 1A (*PTF1A*) is associated with pancreatic agenesis as well as cerebellar agenesis.[4] Insufficiency in the GATA-binding protein 6 *(GATA6*) is also linked to pancreatic agenesis. Of note, the *GATA6* gene mutation has also been associated with pancreatic hypoplasia associated with neonatal DM and other structural cardiac defects.[5] The *HNF1B* gene is also known to regulate pancreatic development, some studies have found that ADP and pancreatic exocrine dysfunction are parts of the phenotype in *HNF1B* mutation carriers;[6] animal experiments have confirmed that retinaldehydrogenase 2 (*Raldh2)*[7] and gene *H1xb9*[8] mutations can cause ADP in mice. In our case we could not do genetic study due to economical strain.

Dorsal agenesis of the pancreas can remain asymptomatic or present with a range of conditions, including recurrent abdominal pain, bloating, acute or chronic pancreatitis, hyperglycemia, polysplenia, congenital heart defects, tetralogy of Fallot, or pulmonary artery stenosis.[9] In our study patients were asymptomatic and developed features of hyperglycaemia only.

As most of the islet β cells are located in the tail of the pancreas, patients with ADP may have diabetes due to insufficient insulin secretion but not necessarily abdominal symptoms. Approximately half of these patients need insulin therapy.[10] Patients may remain asymptomatic and present at young adulthood with complication. A young woman with ADP was reported to have asymptomatic diabetes mellitus and already had severe retinal lesions at the time of presentation.[11]

Other common presentations of ADP are abdominal pain and bloating, which may result from autonomic neuropathy causing dysfunction of the sphincter of Oddi.This noted dysfunction in the sphincter of Oddi may also be linked with acute pancreatitis, pancreatic duct hypertension, increased secretion of pancreatic juice, and compensatory pancreatic head hypertrophy. ADP has been linked to pancreatic neoplasia, such as cystic lesions, malignant intraductal papillary mucinous neoplasia (IPMN), and adenocarcinoma, which some studies have attributed to chronic pancreatitis.[11,12] In our study one of twin had pancreatic cyst.

Previously, diagnosis of ADP was on autopsy or laparotomy; however, advancing medical imaging, including the use of ultrasound, CT, magnetic resonance cholangiopancreatography (MRCP), and endoscopic retrograde cholangiopancreatography (ERCP), has helped identification of this congenital abnormality.

Ultrasonography, although fast and readily available but may fail to identify this ADP, as bowel gas may obscure image acquisition.CT can detect the pancreatic head; however, MRCP or ERCP is able to visualize the pancreatic ductal anatomy adequately and therefore confirm the diagnosis of partial or complete ADP.[13]

Treatment and prognosis depends on the remaining function of pancreas and presence of other associated anomalies. Diabetes mellitus can be managed with insulin. Surgical support may also be needed.

Conclusion

Hyperglycaemia and DM are the common presentation of ADP. Though it is a very rare condition, ADP should also be considered in patients who present with a glucose metabolism disorder along with abdominal pain, pancreatitis, or steatorrhea and with other congenital anomalies. This rare clinical entity was detected by routine ultrasonography of whole abdomen which highlights the importance of imaging while evaluating a newly detected diabetic patient.

Authors' contribution: SJ reviewed the literatures and write the paper; FM reviewed the literatures, supervised the study and reviewed the manuscript. Authors have read and approved the manuscript.

Consent: Consent was taken from parents.

Conflicts of interest: Nothing to declare.

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