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Case report

Dorsal Pancreatic Agenesis: Case Presentation and Literature Review

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Abstract

Dorsal pancreatic agenesis (DPA) is an exceptionally rare congenital malformation resulting from the partial or complete absence of the dorsal pancreatic bud and accounts for fewer than 200 cases reported worldwide. We present the case of a 62-year-old man with insulin-requiring type 2 diabetes mellitus who presented with postprandial abdominal pain and obstructive jaundice. Initial tests showed a distended gallbladder without lithiasis and biochemical cholestasis. Magnetic resonance cholangiopancreatography (MRCP) revealed absence of the pancreatic neck, body, and tail, confirming DPA and ruling out surgically amenable biliary obstruction. Management was conservative, including hydration, analgesia, optimization of insulin therapy, and close monitoring. The patient showed clinical improvement, normalization of liver enzymes, and acceptable glycemic control, and was discharged with multidisciplinary follow-up to monitor long-term metabolic and pancreatic complications. This case highlights the importance of including DPA in the diagnostic algorithm of non-lithiasic obstructive jaundice, as well as the pivotal role of MRCP and endoscopic ultrasound in delineating ductal anatomy and preventing unnecessary interventions. It also underscores the need for a comprehensive approach that considers the increased risk of chronic pancreatitis, exocrine insufficiency, and potential neoplastic transformation of the ventral remnant.

Keywords

Pancreas; congenital abnormalities; magnetic resonance cholangiopancreatography; type 2 diabetes mellitus; obstructive jaundice.

INTRODUCTION

Dorsal pancreatic agenesis (DPA) is a very rare congenital malformation characterized by the complete or partial absence of the portions derived from the dorsal pancreatic bud, namely the neck, body, tail, minor papilla, and accessory duct of Santorini, resulting from a developmental error occurring between the fourth and seventh weeks of gestation, when the dorsal bud either fails to form or does not fuse adequately with the ventral bud[(1)]. This embryological defect is usually attributed to alterations in Hedgehog signaling, the retinoid gradient, and, more recently, to pathogenic variants in genes regulating pancreatic organogenesis, such as GATA6, HNF1B, PDX1, and PTF1A, among others[(2,3)].

Since the first anatomical description made by Daly and colleagues in 1911, the number of confirmed cases has increased from 53 in 2008 to approximately 134 in 2021,

and continues to rise thanks to the dissemination of magnetic resonance cholangiopancreatography and the greater availability of endoscopic ultrasound (EUS); the most recent series estimate that more than 160 cases had been published by 2025[2,4].

DPA may coexist with other midline malformations, including polysplenia, intestinal malrotation, biliary and renal anomalies, as well as various mucinous and solid-cystic pancreatic neoplasms, suggesting a shared genetic and embryological background(2,4–6).

Initial suspicion may arise from the absence of retrogastric pancreatic tissue on different diagnostic imaging studies (computed tomography or ultrasonography); however, definitive ductal characterization is obtained through secretin-stimulated magnetic resonance cholangiopancreatography (MRCP) or endoscopic retrograde cholangiopancreatography (ERCP). EUS provides high-resolution imaging to rule out associated masses and evaluate glandular reserve[1,3–5].

The case of a patient with DPA is presented as a starting point for a narrative review focused on its clinical presentation and its inclusion within the differential diagnosis of abdominal pain and obstructive jaundice, with emphasis on embryogenetic advances, high-precision imaging modalities, and endocrine-metabolic follow-up strategies.

CLINICAL CASE

A 62-year-old male patient with a 10-year history of insulin-requiring type 2 diabetes *mellitus* and well-controlled arterial hypertension presented to the emergency department with an eight-day history of postprandial abdominal pain in the upper abdomen, maximal in the right hypochondrium, colicky in nature, radiating from the flank to the ipsilateral upper lumbar region, with no relief from rest or antispasmodic agents. The pain was accompanied by multiple episodes of vomiting, initially consisting of food contents and later becoming bilious. The symptoms worsened on the day prior to presentation. The patient denied fever, malaise, acholic stools, choluria, or self-reported weight loss.

Physical examination revealed pain on deep palpation of the right hypochondrium, without signs of peritoneal irritation, masses, or organomegaly; renal percussion tenderness was negative. Of note, jaundice of the skin and mucous membranes was observed upon admission, which had not been previously perceived by the patient. Hemodynamic parameters were within normal limits. Admission laboratory tests showed leukocytosis with neutrophilia, hyperbilirubinemia predominantly involving the direct fraction, and a cholestatic pattern with marked elevation of alkaline phosphatase and γ-glutamyl transferase, in addition to glycemic decompensation and mild renal dysfunction (**Table 1**). Initial abdominal ultrasonography reported a distended gallbladder (105 × 40 mm) with a 4-mm wall thickness and no visible gallstones despite an adequate acoustic window.

With these findings, the condition was initially interpreted as a cholestatic jaundice syndrome under investigation. Although the presence of a distended gallbladder without gallstones transiently raised the possibility of acalculous cholecystitis or functional gallbladder distension, the severity of the cholestatic pattern and the presence of overt jaundice primarily suggested obstructive jaundice syndrome. Consequently, conservative management was initiated with hydration, analgesia, metabolic optimization, and broad-spectrum antibiotics while an infectious etiology was being ruled out. Serologic testing for hepatotropic viruses and leptospira, as well as other infectious and immunologic etiologic studies, yielded negative results.

During hospitalization, the pain progressively subsided; however, glycemic lability was documented, with blood glucose levels >300 mg/dL despite adjustments to the insulin regimen every 48 hours, along with persistence of clinical jaundice and cholestatic abnormalities on serial follow-up tests, without complete normalization of bilirubin levels during the hospital stay. Given the combination of jaundice, a distended gallbladder, and biochemical abnormalities, the general surgery service considered the possibility of obstructive jaundice syndrome of probable periampullary origin, raising the hypothesis of a neoplastic proliferative process (Courvoisier-Terrier sign). Upper gastrointestinal endoscopy revealed no lesions in the duodenal bulb or second portion of the duodenum, nor any signs of a periampullary tumor.

Magnetic resonance cholangiopancreatography (MRCP) was requested as the imaging modality of choice for characterization of the biliary tract and pancreas. This examination confirmed gallbladder distension without gallstones or dilation of the main biliary tract, mild hepatomegaly, a small volume of free fluid, and absence of the pancreatic body and tail, findings consistent with congenital dorsal pancreatic agenesis (**Figure 1**). Surgical obstructive pathology was therefore ruled out, and the hypothesis of periampullary neoplasia initially suggested by the surgical team was reconsidered. Endoscopic retrograde cholangiopancreatography (ERCP) was evaluated as a complementary diagnostic procedure; however, it was deferred because of the absence of evidence of mechanical obstruction on MRCP, the patient's clinical stability, and the non-negligible risk of post-procedural pancreatitis. The treating center does not have access to secretin-enhanced magnetic resonance imaging or pancreatobiliary endoscopic ultrasonography. Due to limitations in availability and insurance coverage, these studies could not be performed,

Table 1. Follow-up Laboratory Findings of the Patient

Laboratory Test	Patient Value	Reference Range
Hemoglobin	14 g/dL	Men: 13,5-17.5 g/dL Women: 12,0-15.5 g/dL
Hematocrit	42%	Men: 41%-53% Women: 36%-46%
Leukocytes	13,470 × 10^3/mm^3	4.5-11 × 10^3/mm^3
Neutrophils	12,160 × 10^3/mm^3	1.5-8.0 × 10^3/mm^3
Lymphocytes	610 × 10^3/mm^3	1.0-4.0 × 10^3/mm^3
Platelets	329,000 × 10^3/mm^3	150-400 × 10^3/mm^3
Sodium	131 mmol/L	136-145 mmol/L
Potassium	4.73 mmol/L	3.6-5.5 mmol/L
Chloride	96 mmol/L	95-108 mmol/L
Amylase	44 U/L	25-115 U/L
Lipase	118 U/L	73-393 U/L
Alkaline phosphatase	766 U/L	46-116 U/L
AST (SGOT)	184 U/L	15-37 U/L
ALT (SGPT)	253 U/L	16-63 U/L
γ-glutamyl transferase	1,878 U/L	15-85 U/L
LDH	178 U/L	85-227 U/L
Creatinine	1.70 mg/dL	0.70-1.30 mg/dL
BUN	24 mg/dL	7-18 mg/dL
Glucose	452 mg/dL	74-106 mg/dL
Total bilirubin	5.7 mg/dL	0.2-1.0 mg/dL
Direct bilirubin	5.5 mg/dL	0.0-0.2 mg/dL
Indirect bilirubin	0.2 mg/dL	≤0.76 mg/dL
C-reactive protein	31.4 mg/dL	<0.3 mg/dL
HbA_{1c}	11.3 %	Diabetes ≥6.5 %
Anti-hepatitis C	Negative	Negative
Leptospira IgG/IgM	Negative	Negative
Hepatitis A IgG	>100 mIU/mL	Reactive >20 mIU/mL
Hepatitis A IgM	0.25 UI	Reactive >1.2 UI
HBsAg	Negative	Negative
HIV	Negative	Negative
CMV IgM	0.27 (UI)	Negative <0.90
CMV IgG	9.55 (UI)	Positive >1.1
Free T_4	1.27 ng/dL	0.8-2.2 ng/dL
Total T_3	3.81 ng/mL	0.8-2.0 ng/mL
TSH	2.47 µIU/mL	0.4-4.0 µIU/mL
ANA	Negative	Negative
Total IgG	1,116 mg/dL	650-1,600 mg/dL
PT (patient/control)	9.5 s/11.4 s	11-13.5 s
INR	0.9	0.8-1.2
aPTT (patient/control)	26.6 s/27.3 s	25-35 s
Urine pH	6.0	4.6-8.0
Urine specific gravity	1.015	1.005-1.030
Glycosuria	1,000 mg/dL	Negative
Proteinuria	30 mg/dL	Negative
Urinary leukocyte sediment	0-2 × field	≤5 × field
Urinary red blood cell sediment	0-2 × field	≤3 × field
Bacterias en orina	Positive (+)	Negative

ALT (SGPT): alanine aminotransferase; ANA: antinuclear antibodies; aPTT: activated partial thromboplastin time; AST (SGOT): aspartate aminotransferase; BUN: blood urea nitrogen; CMV: cytomegalovirus; HBsAg: hepatitis B surface antigen; HbA_{1c}: glycated hemoglobin; HIV: human immunodeficiency virus; IgG: immunoglobulin G; IgM: immunoglobulin M; INR: international normalized ratio; LDH: lactate dehydrogenase; PT: prothrombin time; TSH: thyroid-stimulating hormone. Author's own research.

although they were recommended as complementary evaluations during outpatient follow-up, in accordance with the literature regarding detailed functional and anatomical assessment of the pancreatic duct in dorsal agenesis.

The endocrinology service documented a glycated hemoglobin level of 11.3% and adjusted the insulin regimen in conjunction with intensive education on self-monitoring and home glucose monitoring. No specific genetic studies were performed to rule out variants associated with congenital pancreatic malformations, and consideration of such testing was deferred to referral centers for long-term follow-up. The patient evolved with complete resolution of pain, progressive decline in liver enzyme levels, and improvement in glycemic control following optimization

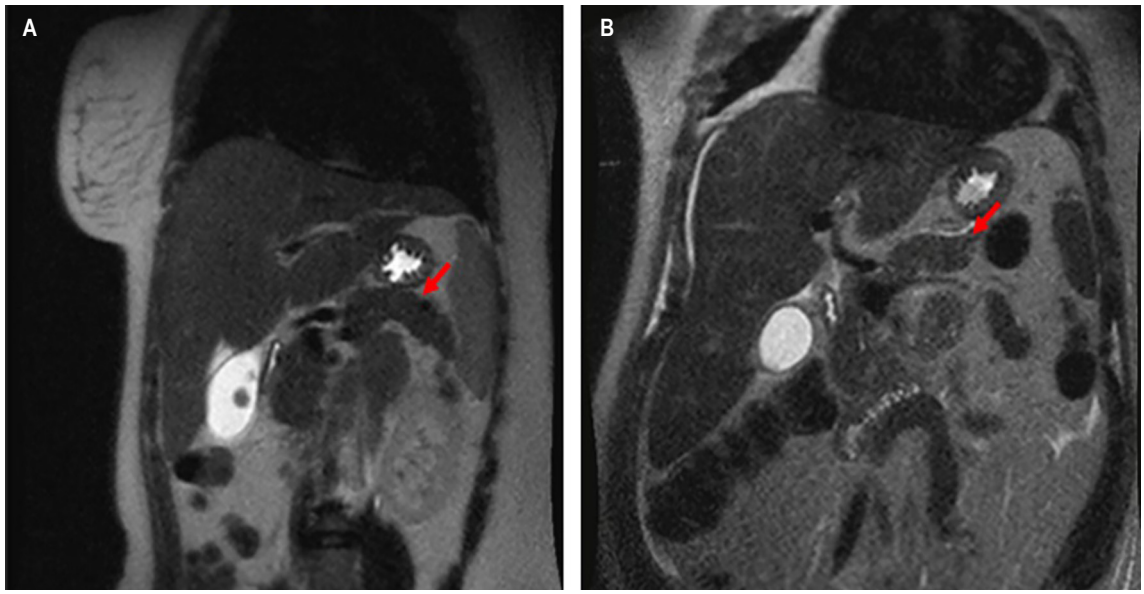


Figure 1. ERCP examination from the clinical case. **A.** Coronal T2-weighted magnetic resonance cholangiography image demonstrating normal pancreatic anatomy (arrow). **B.** Axial T2-weighted magnetic resonance cholangiography image from the patient showing evidence of pancreatic tail agenesis (arrow). Author's file.

of treatment. He was discharged on hospital day 12 with combined follow-up by gastroenterology, endocrinology, and surgery, aimed at monitoring pancreatic exocrine and endocrine function, as well as excluding long-term biliary or pancreatic complications.

DISCUSSION

Definition and Classification

Dorsal pancreatic agenesis (DPA) is an extremely rare congenital anomaly in which the dorsal pancreatic bud fails to develop or undergoes partial or complete involution[1,4]. Two morphological phenotypes are distinguished: complete DPA, characterized by absence of the neck, body, tail, minor papilla, and accessory duct of Santorini; and partial DPA, in which variable portions of the body, minor papilla, or duct of Santorini persist[1,4,6].

The terms *short pancreas* and *pancreatic hypoplasia* have also been described in the literature and correspond to incomplete expressions of the same embryological alteration[1].

Epidemiology

Since the first case identified at autopsy in 1911, reports have increased with the dissemination of MRCP and EUS[2–4]. The most recent systematic reviews include between 68 and 106 published cases from 1913 to 2021, although case series and isolated reports raise the number to approximately 150–160 patients by 2025[7]. The true population prevalence remains uncertain; data from radiologic series place it at 1–2 per 10,000 abdominal examinations[2,4,7].

Embryogenesis and Etiopathogenesis

Pancreatic organogenesis occurs between the fourth and seventh weeks of gestation with the formation of two endodermal buds: the ventral bud, which gives rise to the future uncinate process and the caudal half of the pancreatic head, and the dorsal bud, which corresponds to the cranial half of the head, neck, body, and tail. Between weeks 6 and 7, the duodenum rotates and both buds fuse, giving rise to the main duct of Wirsung and, secondarily, the accessory duct of Santorini (**Figure 2**)[1,4].

Alterations in the Hedgehog pathway, the retinoid gradient, and, most notably, pathogenic variants in genes regulating pancreatic organogenesis listed in **Table 2** have been implicated[2,9–11].

Most cases continue to be considered sporadic; however, autosomal dominant patterns and X-linked inheritance have been described in families with heterotaxy and polysplenia[1].

Clinical Manifestations

Sixty percent of patients with DPA remain asymptomatic, and the diagnosis is made incidentally. In symptomatic patients, the predominant manifestations are chronic or recurrent epigastric pain (approximately 90%), proba-

bly secondary to sphincter of Oddi dysfunction or ductal hypertension of the ventral remnant. Diabetes *mellitus* may also occur (40%–60%), usually requiring insulin therapy, and is attributed to the loss of β-islet cells in the body and tail. Acute or chronic pancreatitis, steatorrhea, and weight loss have been reported in a small percentage of cases associated with overt exocrine insufficiency in <10%[1,4,12]. Other findings include obstructive jaundice syndrome, biliary complications, and associated cystic nephropathy.

Imaging Diagnosis

Contrast-enhanced computed tomography suggests the anomaly by demonstrating absence of pancreatic parenchyma anterior to the splenic vein; gastric or jejunal interposition produces the "dependent stomach/intestine sign"[4,7,12].

Abdominal magnetic resonance imaging with T2-weighted sequences and secretin-stimulated MRCP constitute the diagnostic modalities of choice because they delineate the ductal tree, confirm the absence of the dorsal duct, and rule out pseudoagenesis or atrophy secondary to pancreatitis. Three-dimensional reconstruction increases sensitivity without generating radiation exposure or risk of procedure-induced pancreatitis[1,3,4].

EUS provides the best parenchymal resolution and allows characterization and sampling of incidental space-occupying lesions. ERCP is reserved for cases in which therapeutic intervention with sphincterotomy or stent placement is being considered[13].

Associated Diseases and Malformations

DPA is associated with midline anomalies such as polysplenia, heterotaxy, wandering spleen, and intestinal malrotation; urologic abnormalities such as horseshoe kidney and renal dysplasia; cardiac abnormalities such as tetralogy of Fallot and coarctation; and gynecologic abnormalities such as uterus didelphys[9–12].

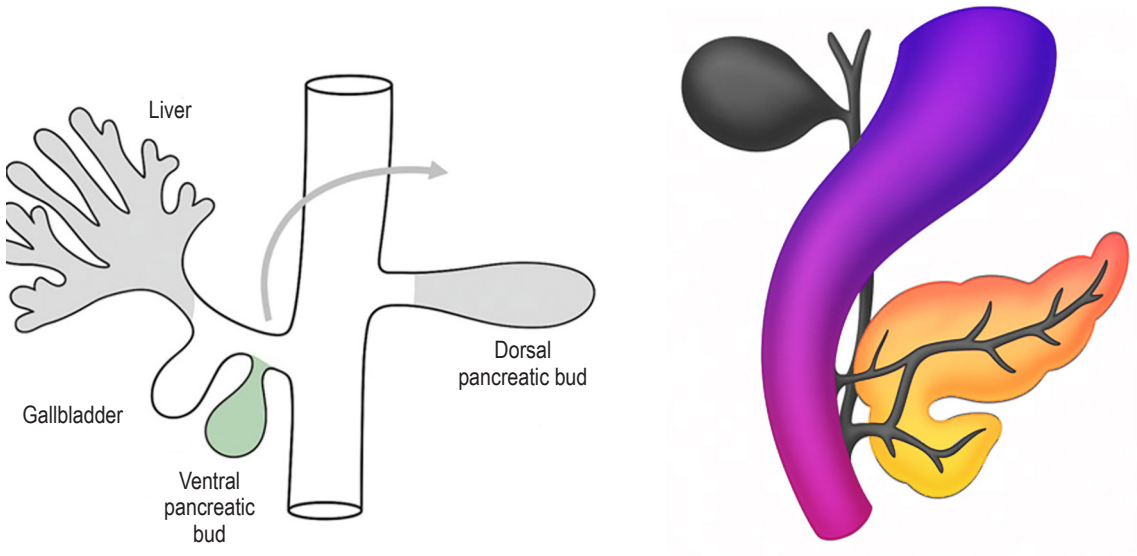


Figure 2. Embryologic development of the pancreas. Adapted from: Suckale J and colleagues. Front Biosci. 2008;13:7156-71[8].

Table 2. Genes Implicated in Dorsal Pancreatic Agenesis

Gene	Proposed Mechanism	Clinical Evidence
HNF1B	Early regulation of dorsal progenitor cells	Heterozygous mutations associated with DPA, diabetes, and cystic nephropathy
GATA6	Essential for proliferation and branching of pancreatic endoderm	Haploinsufficiency associated with agenesis and neonatal diabetes
PDX1 (MODY-4)	Transcription of islet-cell genes	Partial DPA associated with juvenile-onset diabetes

Author's own research.

Over the last 15 years, multiple pancreatic neoplasms associated with DPA have been described, including neuroendocrine tumors, mucinous tumors, and adenocarcinomas. Although causality has not been established, underlying chronic pancreatitis and regenerative stimulation of the ventral remnant could constitute a risk setting because of the unregulated inflammatory response[1–3].

Differential Diagnosis

Differential diagnoses that should be considered include pseudoagenesis secondary to pancreatic atrophy following chronic pancreatitis; distal pancreatic lipomatosis, in which the parenchyma is replaced by fat while the dorsal duct remains intact; pancreas divisum, in which the body and tail are normal and dual Wirsung-Santorini drainage is preserved; and finally, carcinoma of the pancreatic head with retrograde atrophy and cystic tumors of the body and tail[6,7,12].

Treatment and Follow-up

Given its rarity, there is no established treatment for DPA[1]. Patients with DPA are commonly treated according to their symptoms. Based on the clinical manifestations reported in DPA cases, recommendations are focused on low-fat dietary modifications and diabetes control as the primary treatment measures[3,4]. The use of analgesics is recommended for pain management, as well as pancreatic enzymes, which decrease pancreatic secretion and relieve pain. Patients with DPA associated with pancreatitis and pancreatic duct abnormalities may benefit from sphincterotomy. Endoscopic treatment is presented as an alternative to overcome ductal resistance to pancreatic flow[4,6,13].

Prognosis

The prognosis is generally favorable if pain and metabolic complications are adequately controlled. Nevertheless, the coexistence of chronic pancreatitis or neoplasia requires prolonged multidisciplinary follow-up.

CONCLUSIONS

Dorsal pancreatic agenesis, although exceptional, should be systematically incorporated into the differential diagnosis of recurrent abdominal pain and obstructive jaundice, especially when initial studies rule out evident lithiasic or neoplastic causes. This case highlights three cardinal points. First, the importance of considering congenital pancreatic malformations when initial diagnostic imaging is inconclusive; MRCP and EUS have become indispensable techniques for delineating ductal anatomy and avoiding unnecessary surgical interventions. Second, the relevance of a multidisciplinary approach: close collaboration among gastroenterology, surgery, radiology, and endocrinology enables timely diagnosis, optimizes glycemic control, and identifies associated complications such as pancreatitis or exocrine insufficiency. Third, the need for long-term follow-up because of the increasing risk of chronic pancreatitis and possible neoplastic transformation in the ventral remnant. In summary, DPA, although uncommon, should be considered in the diagnostic algorithm for abdominal pain and jaundice; early detection and a structured surveillance plan are essential to improve clinical outcomes and patient quality of life.

Conflict of Interest

The authors declare no conflicts of interest regarding this clinical case.

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