CASE REPORT

Agenesis of the Dorsal Pancreas: An Uncommon Cause of Pancreatic Diabetes in Young Adults. A Case Report

Agénésie du Pancréas Dorsal : Une Cause Rare de Diabète Pancréatique chez les Jeunes Adultes. Rapport de Cas

Yosra Hasni [1,3], Wiem Saafi [1,3], Hamza EL Fekih[1,3], Salem Braham [2], Soumaiya Tahri [1,3], Amel Maaroufi[1,3]

1. Hospital Farhat Hached, endocrinology department, 4031, Sousse, Tunisia
2. Hospital Farhat Hached, radiology department, Sousse, Tunisia
3. Ibn El Jazzar Medical Faculty of Sousse, University of Sousse, Sousse, Tunisia

Abstract

Introduction: Agenesis of the dorsal pancreas (ADP) is a rare congenital abnormality characterized by the absence of the body and tail of the pancreas. This condition is often asymptomatic. While many patients exhibit nonspecific symptoms, some may experience more severe manifestations complicating the diagnostic process.

Observation: We presented the case of a 27-year-old man who was diagnosed with type 1 diabetes mellitus three years ago. He was experiencing considerable weight loss, abdominal pain, and diarrhea despite effectively managed insulin therapy. The patient appeared emaciated and malnourished. Investigations revealed a normal thyroid function, negative celiac disease serology, and negative pancreatic antibodies. Abdominal imaging demonstrated ADP. After treatment with insulin and pancreatic enzymes, we noted a significant improvement in the patient's condition.

Conclusion: This case emphasizes diagnostic challenges in young patients presenting with pancreatic diabetes. Early recognition and appropriate management are crucial to prevent complications of both exocrine and endocrine deficiencies.

Key words: Congenital Abnormalities, Endocrine glands, Magnetic resonance imaging, Pancreas, Pancreatic diseases

Résumé

Introduction: L'agénésie du pancréas dorsal (APD) est une anomalie congénitale rare caractérisée par l'absence du corps et de la queue du pancréas. Cette condition est souvent asymptomatique. Bien que de nombreux patients présentent des symptômes non spécifiques, certains peuvent avoir des manifestations plus sévères compliquant ainsi le processus de diagnostic.

Observation: Nous avons présenté le cas d'un jeune homme de 27 ans qui a été diagnostiqué avec un diabète sucré de type 1 il y a trois ans. Il souffrait d'un amaigrissement important, de douleurs abdominales et de diarrhée malgré une insulinothérapie intensifiée. Le patient apparaissait émacié et malnutri. Les investigations ont révélé une fonction thyroïdienne normale, une sérologie négative pour la maladie cœliaque et des anticorps pancréatiques négatifs. L'imagerie abdominale a montré une APD. Après un traitement par insuline et enzymes pancréatiques, nous avons noté une amélioration significative de l'état du patient.

Conclusion: Ce cas souligne les défis diagnostiques chez les jeunes patients présentant un diabète pancréatique. La reconnaissance précoce et la gestion appropriée sont cruciales pour prévenir les complications des déficiences exocrines et endocriniennes.

Mots clés: Glandes endocrines, imagerie par résonance magnétique, maladies du pancréas, malformations, Pancréas

Correspondance
Wiem Saafi
Hospital Farhat Hached, endocrinology department, 4031, Sousse, Tunisia
Email: wiem.saafi@gmail.com

LA TUNISIE MEDICALE-2025; Vol 103 (02): 290-293 DOI: 10.62438/tunismed.v103i2.5405

Introduction

Agenesis of the dorsal pancreas (ADP) is a rare congenital anomaly characterized by the absence of the dorsal portion of the pancreas, which includes the body and tail (1). This condition was first described in 1911 (1), and since then, approximately 100 cases have been documented in the medical literature (1), making it a significant medical curiosity.

Patients with ADP may exhibit a range of nonspecific symptoms, including epigastric discomfort, hyperglycemia, and episodes of acute or chronic pancreatitis (2). In many instances, individuals with ADP remain asymptomatic, and the condition is often discovered incidentally during imaging studies conducted for unrelated issues (3). However, some patients may present with more severe manifestations, such as recurrent abdominal pain, which can complicate the diagnostic process (4).

We presented a case of ADP in a young diabetic man.

Case report

We presented the case of a 27-year-old man of low socioeconomic status, who presented with recurrent abdominal pain, chronic diarrhea, and substantial weight loss. In August 2019, he was diagnosed with type 1 Diabetes Mellitus (T1DM) at age 24 following ketoacidosis presentation and significant weight loss. Pancreatic antibodies (Glutamic acid decarboxylase antibodies, Insulinoma-associated protein 2 antibodies and Zinc transporter 8 antibodies) were negative. The patient has been receiving insulin therapy since diagnosis. His T1DM was poorly controlled due to poor adherence to treatment. The patient had no family history of diabetes mellitus or autoimmune disease.

The patient was hospitalized in December 2021 in front of considerable weight loss, abdominal pain, and diarrhea despite effectively managed insulin therapy. On examination, the patient appeared severely malnourished with arterial hypotension (systolic pressure/diastolic pressure: 80/55 mmHg) and tachycardia at 98 bpm. He was cachectic with a body mass index of 14 kg/m². Superinfected ulcerative skin lesions were observed.

Initial investigations as detailed in table 1 confirmed poor controlled diabetes mellitus and malabsorption reveling a high fasting blood glucose, an elevated glycated hemoglobin with normocytic normochromic anemia. Serum creatinine was elevated. Thyroid-stimulating hormone levels were within the normal range, and coeliac disease antibodies were negative.

The skin lesions were suggestive of severe immunodepression leading to the practice of serological tests for viral hepatitis and Human Immunodeficiency Virus that were unremarkable. Pancreatic enzyme levels were normal, and Stool examinations did not reveal any infection. Gastrointestinal endoscopy revealed gastritis with Helicobacter pylori infection and showed no signs of intestinal disease. Colonoscopy was negative for inflammatory bowel disease.

Table 1. Biological investigation of the patient.

Variable (unit)	Value	Normal range
Hemoglobin (g/dL)	11.5	12-16
Fasting glycaemia (mg/dL)	270	70-99
Glycated hemoglobin (%)	16.2	
Total cholesterol (mg/dL)	92.8	<200
Triglycerides (mg/dL)	79.6	<150
High density lipoprotein cholesterol (mg/dL)	34.8	>40
Creatinine (mg/dL)	3.73	0.6 - 1.2
Lipase (UI/L)	25	22-55
Thyroid-stimulating hormone (mUI/l)	1.8	0.4-40

Given the persistence of steatorrhea and abdominal pain, abdominal computed tomography (CT) scan was practiced, showing only cephalic part of the pancreas (Figure 1).

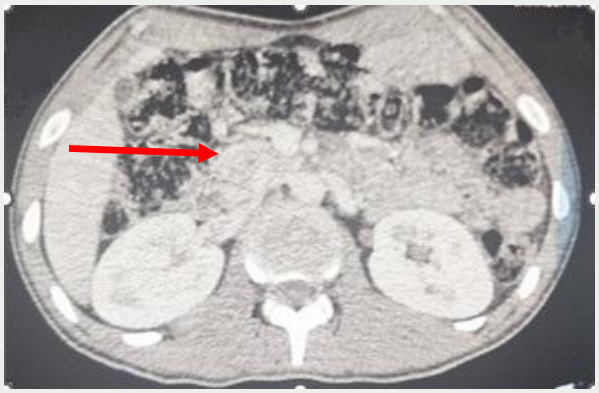

Figure 1. Axial slices of abdominal computed tomography showing only cephalic part of the pancreas measuring 3 cm.

Magnetic resonance imaging (MRI) confirmed the agenesia of pancreatic duct and corporeo-caudal parts of the pancreas (Figure 2).

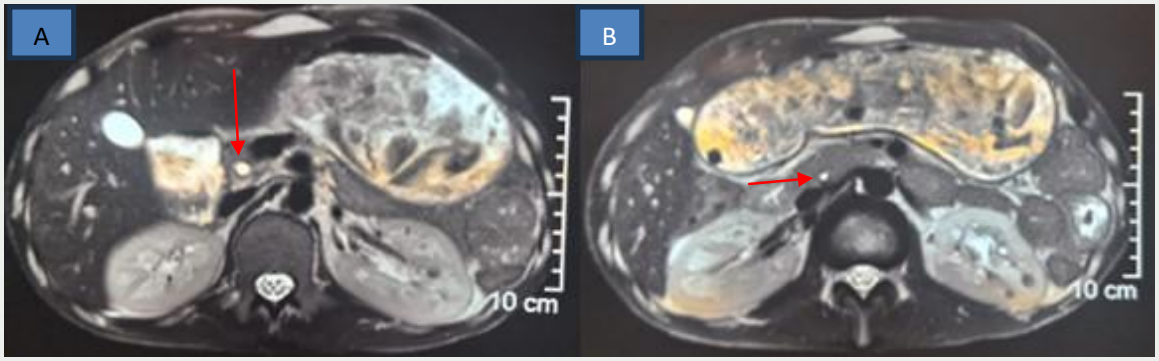

Figure 2. **A & B**. T2 Spectral Adiabatic Inversion Recovery axial slices of pancreatic magnetic resonance imaging confirmed the absence (agenesia) of pancreatic duct and corporeo-caudal parts of the pancreas.

The patient was treated with insulin therapy and pancreatic enzyme replacement therapy (PERT), leading to resolution of diarrhea, improved glycemic control, and a weight gain of 6 kilograms over three months.

Discussion

This case emphasizes the importance of identifying ADP as a potential differential diagnosis for patients initially misclassified as having T1DM with negative antibodies, thereby reinforcing the necessity for thorough diagnostic assessments.

ADP is a very rare congenital anomaly due to abnormal and relatively complex embryogenesis (1). The pancreas develops via ventral and dorsal endodermal buds (1). The upper part of the head, body and tail of the pancreas is formed by the dorsal bud which drains through the duct of Santorini, while the ventral bud gives rise to most of the head and the uncinate process which drains through the duct of Wirsung (1).

Till late 2024, the pathogenesis of ADP is not fully understood (2). The Hepatocyte Nuclear Factor-1 beta (HNF1B) gene is known to regulate pancreatic development, some studies have reported that ADP and pancreatic exocrine dysfunction are parts of the phenotype in HNF1B mutation carriers, and GATA binding protein 6 (GATA6) gene mutation (5).

In our patient's case, the diagnosis of pancreatic diabetes was not suspected since the patient presented mainly signs of insulin deficiency. His young age and the initial clinical picture made the diagnosis of T1DM with negative pancreatic antibodies more likely despite the absence of family history of autoimmune diseases. However, the subsequent clinical deterioration, significant weight loss, abdominal pain, and signs of exocrine pancreatic insufficiency (ie; steatorrhea, hypocholesterolemia) made us suspect pancreatic diabetes that was confirmed by pancreatic imaging. In fact most cases of ADP are asymptomatic and discovered incidentally (6).

A review of literature carried out between 1931 and 2021 collated 57 publications describing 68 cases, 38 of which were women, ranging in age from one month to 79 years (4). The reported manifestations in symptomatic forms of ADP were

i) Abdominal pain described as is a dull, intermittent pain in the epigastrium and right hypochondrium (4), which may radiate to the back and is often aggravated by eating (1).

ii) Pancreatitis: The most common form of pancreatitis is acute or recurrent, presenting with symptoms including nausea, vomiting and elevated pancreatic enzymes (4,6). The mechanism of which may be Oddi sphincter dysfunction, pancreatic head compensatory hypertrophy, increased pancreatic juice secretion, and pancreatic duct hypertension (7) .

iii) Hyperglycemia and diabetes mellitus are described in approximately 45% of patients with ADP (4). The reduced β-cell mass from the absent dorsal pancreas is the primary mechanism leading to insulin deficiency and hyperglycemia in these patients. On another hand, signs of exocrine pancreatic insufficiency are less frequent as only 10% of functional pancreatic tissue is required for adequate exocrine function. The relationship between these two conditions remains poorly understood, and there are limited case reports available in the literature (1).

Due to the deep-seated position of the pancreas, abdominal CT is commonly selected as the first imaging modality to assess for pancreatic abnormalities (9). Although abdominal ultrasound has been shown to be useful in diagnosing ADP (9), the superior visualization and three-dimensional reconstruction capabilities of modern CT scanners, as highlighted by Guimarães et al. (8) have solidified its role as the primary diagnostic tool. In fact, CT scanning offers detailed cross-sectional images of the abdomen, making it a valuable tool for evaluating the presence or absence of pancreatic tissue (10). By visualizing the absence of the pancreatic body and tail, CT can help distinguish dorsal pancreatic agenesis from other conditions like pancreatic lipomatosis and chronic pancreatitis (8). MRI provides a comprehensive view of pancreatic morphology and surrounding structures. Magnetic resonance cholangiopancreatography (MRCP), a non-invasive imaging technique, offers detailed visualization of the pancreatic ductal system without ionizing radiation (10). It is particularly effective in demonstrating the absence of the dorsal pancreatic duct and can confirm the diagnosis of dorsal pancreatic agenesis by visualizing the main and accessory pancreatic ducts, as well as endoscopic retrograde pancreatography (ERCP) (9). MRCP, a non-invasive imaging technique, is generally preferred over ERCP, which carries the risk of inducing acute pancreatitis and potentially exacerbating diabetes mellitus. While MRI or CT scans are typically sufficient for detecting ADP, ultrasound imaging may be limited due to interference from bowel gas, hindering visualization of the pancreatic body and tail (10).

In our case, despite intensive insulin therapy glycemic control was not achieved and the patient presented significant weight loss, malnutrition and signs of immunodepression. These manifestations regressed after the use of PERT. That emphasizes the need for adequate management for both endocrine and exocrine abnormalities.

The findings of our case report are subject to two limitations including the lack of genetic testing since the potential involvement of the HNF1B and GATA6 genes was not confirmed. In addition, the present study is a single-patient case report making the conclusions drawn from this isolated case not generalizable to all cases of ADP. These limitations restrict the broader applicability of our findings.

Conclusion

ADP is a rare congenital anomaly that can lead to pancreatic diabetes. This diagnosis can be mistaken for other types of diabetes mellitus since it is frequently asymptomatic. This diagnosis should be considered in young patients presenting with diabetes mellitus when pancreatic antibodies are negative even in the absence of signs of exocrine insufficiency. Imaging studies, including

CT and MRI, are important diagnostic exams. Treatment with insulin and pancreatic enzyme supplementation resulted in improved glycemic control and weight gain. Early recognition and appropriate management are crucial to prevent complications and improve patient outcomes.

References

1. Cienfuegos JA, Rotellar F, Salguero J, Benito A, Solórzano JL, Sangro B. Agenesis of the dorsal pancreas: systematic review of a clinical challenge. Rev Esp Enfermedades Dig. 2016;108(8):479-84.
2. Robert AP, Iqbal S, John M. Complete agenesis of the dorsal pancreas: a rare clinical entity. Int J Appl Basic Med Res. 2016;6(4):290.
3. Grey CA, Desai A, Nowicki MJ, Bhesania N. Agenesis of the dorsal pancreas: case report and review of age-related differences in presentation. JPGN Rep. 2023;4(3):e337.
4. Lalchandani A, Maurya A, Rizvi SFM, Yadav A. Agenesis of the dorsal pancreas: a rare cause of diabetes and recurrent upper abdominal pain. Cureus. 2023;15(2):e34689.
5. Chao CS, McKnight KD, Cox KL, Chang AL, Kim SK, Feldman BJ. Novel GATA6 mutations in patients with pancreatic agenesis and congenital heart malformations. Plos One. 2015;10(2):e0118449.
6. V SK, Sangu P, C K, R P, Chidambaranathan S, Obla Lakshmanamoorthy NB. Congenital anomalies of the pancreas: various clinical manifestations and their impact on pancreatic diseases and outcomes. Cureus. 2022;14(8):e27915.
7. Jain A, Singh M, Dey S, Kaura A, Diwakar G. A rare case of complete agenesis of dorsal pancreas. Euroasian J Hepato-Gastroenterol. 2017;7(2):183-4.
8. Guimarães AB, Guimarães CA, Manso JEF. Agenesis or pseudoagenesis of the dorsal pancreas. Rev Colégio Bras Cir. 2015;42:352-5.
9. Mohapatra M, Mishra S, Dalai PC, Acharya SD, Nahak B, Ibrarullah M, et al. Imaging findings in agenesis of the dorsal pancreas: report of three cases. JOP J Pancreas. 2012;13(1):108-14.
10. Jha N, Prasad U, Kumar D, Gupta R, Jha A, Suman SK. The role of cross-sectional imaging in the diagnosis of agenesis of the dorsal pancreas: a case series. Cureus. 2023;15(6):e40930.