Published in final edited form as:

Pancreas. 2016 February ; 45(2): e1–e2. doi:10.1097/MPA.0000000000000506.

An Unusual Presentation of Pediatric Autoimmune Pancreatitis

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To The Editor

Autoimmune pancreatitis (AIP) is a form of acute and chronic pancreatitis with an overall incidence described as up to 0.71–0.82 per 100,000 persons.^{1,2} Two distinct phenotypes have been identified.³ Type 1 is more common in adult males and associated with serum IgG4 elevation and characteristic histological findings, whereas type 2 is more common in the pediatric population with a less frequent association with serum IgG4 elevation.^{1,3,4} No confirmed cases of type 1 AIP have been described in the pediatric population although there have been a few cases of probable type 1 AIP.^{4,5} We present a case of probable type 1 AIP in a child with a unique presentation of steatorrhea resulting from chronic pancreatitis.

Case Presentation

An 8-year old female presented to the outpatient clinic with a chief complaint of weight loss (11 lbs.; 17% of total body weight) and diarrhea over a four-month period. Diarrhea was described as “oily, yellow/orange” and foul smelling. There was a history of vague, intermittent abdominal pain but no prior episodes of documented acute pancreatitis.

Initial workup was revealing for amylase 46 u/L (< 105 u/L), lipase 40 u/L (48–199 u/L), fecal elastase 15 mcg/g (> 201 mcg/g), and sweat chloride 30 mEq Cl/L (0–39 mEq Cl/L for age range). Serum total IgG was elevated at 1400 mg/dL (598–1379 mg/dL) with a serum IgG4 of 320 mg/dL (<189 mg/dL). Direct endoscopic pancreatic function testing (ePFT) revealed insufficient levels of trypsin 5.9 and 7.4 nm/ml/min (>55.4 nm/ml/min), amylase 8 and 9.8 um/ml/min (>32 um/ml/min), lipase 7 and 4 um/ml/min (>146 um/ml/min) and chymotrypsin 0.9 and 0.9 um/ml/min (>2.5 um/ml/min) on two separate samples. Immunoreactive trypsinogen was < 1.2 ng/mL (10–57 ng/mL). Gene sequence analysis of *CFTR*, *PRSS1*, *SPINK1*, and *CTRC* revealed that the patient was heterozygous for the

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Conflict of Interest and financial disclosures: The authors of this manuscript do not have any conflicts of interest or financial disclosures to report.

p.R1070Q pathogenic mutation in the CFTR gene. Magnetic resonance cholangiopancreatography (MRCP) was consistent with chronic pancreatitis with a small volume pancreas and residual low-grade inflammation throughout with emphasis on the body and tail. The pancreatic duct was collapsed and not seen. (Figure 1)

Endoscopic ultrasound (EUS) with pancreatic biopsy using fine needle aspiration (FNA) yielded tissue that was insufficient for analysis. Given the elevated IgG4 and imaging findings, a diagnosis of probable type 1 AIP was given. Patient was started on prednisone 20 mg daily (0.7 mg/kg/day) for three weeks followed by a taper over two months. Patient is currently asymptomatic more than six months since prednisone therapy with complete resolution of diarrhea and abdominal pain. She remains on supplemental pancreatic enzymes and is gaining weight. Total serum IgG and IgG4 have declined to 1030 mg/dL and 167 mg/dL, respectively. Repeat immunoreactive trypsinogen remains undetectable at <1.2 ng/mL despite therapy.

Discussion

We describe a case of probable type 1 AIP in a child with a unique presentation. Most cases of type 1 AIP occur in adult males and present with obstructive jaundice.^{1,6} Additionally, organ involvement outside the pancreas is common with type 1 AIP and occurs in 50–70% of cases.¹ Our patient's presentation varied from these more commonly described features with an insidious onset of steatorrhea, vague abdominal pain and weight loss over several months without evidence for jaundice or other organ involvement.

According to the ICDC for AIP³, our patient met criteria for probable type 1 AIP with atypical imaging findings of pancreatic parenchymal atrophy, level 2 serology and resolution of pain and IgG4 elevation after steroid course. To date, she does not have any other organ involvement.

Several studies have illustrated that serum IgG4 can be elevated in various conditions in addition to AIP to include IgG4-associated cholangitis, pancreatic cancer, benign pancreatic tumors and even in a normal pancreas.^{6,7} However, ranges for each of these entities differ. A serum IgG4 level of 135–140 mg/dL has a sensitivity of 76–95% and a specificity of 86–97% for type 1 AIP.^{1,6,7} Specificity reached 99% in one study when a cutoff of > 280 mg/dL was implemented.⁶ Two cases of probable type 1 AIP in children have been described in the literature.⁵ Only one had documented serum IgG4 elevation to a level of 224 mg/dL. Our patient presented with an initial serum IgG4 level of 320 mg/dL, which is well above the thresholds discussed here and the highest reported level of serum IgG4 described in the pediatric autoimmune pancreatitis literature.

The CFTR gene mutation (p.R1070.Q, also known as c.3209G>A) found in our patient has been documented in the literature in subjects with pancreatic disease including chronic pancreatitis and likely is contributing to our patient's pancreatic dysfunction.^{8,9}

In conclusion, we have shared a case of probable type 1 AIP in an 8-year old child who presented with weight loss, steatorrhea and vague abdominal pain. Future studies in pediatric AIP are needed to further our understanding of this rare entity.

Acknowledgments

There were no sources of funding or financial disclosures for this manuscript.

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Figure 1. MRCP axial T2 images of the pancreas. These are several axial T2 images of the pancreas with fat saturation demonstrating a heterogeneous signal, particularly at the body (a) and tail region (b). Overall pancreatic volume is decreased. The pancreatic duct is collapsed and not seen.