Genetics in idiopathic pancreatitis and acute recurrent pancreatitis

Key words: Genetics. Acute pancreatitis. Recurrent pancreatitis. Chronic pancreatitis. CFTR. PRSS1. SPINK1.


Dear Editor,

Acute recurrent pancreatitis (ARP) occurs in 10-35% of children presenting idiopathic acute pancreatitis (IAP) and can evolve to chronic pancreatitis (CP), especially if genetic mutations are present (1).

Case report

A 14-year-old male with a history of three episodes of AP at the ages of 10, 11 and 12 presented with a new episode. Previous episodes were diagnosed as IAP after performing a complete medical history and imaging tests. No history of drugs or toxin consumption was reported.

Physical examination revealed nothing but epigastric pain. Laboratory findings were: elevated pancreatic amylase (1,345 U/l) and lipase (2,300 U/l) serum values. Sweat test, viral serology, metabolic and auto-immunity tests were negative. An obstructive cause was discarded with magnetic resonance cholangio-pancreatography (Fig. 1). Fasting, fluid resuscitation and analgesics were initiated favorably.

Routine tests during follow-up included monitoring of exocrine and endocrine pancreatic function (fecal-elastase-1 and glycosylated hemoglobin serum levels respectively).

At month 18, the patient presented with a mild episode not requiring hospitalization. At month 22 the laboratory findings revealed a decreased value of fecal elastase (85 μg/g: normal ranges > 200 μg/g stool), without steatorrhea. The findings suggested a mild exocrine pancreatic insufficiency (EPI). Ultrasoundography revealed pancreatic atrophy and endoscopic ultrasound confirmed CP. No pharmacological treatment was initiated as the patient had an adequate nutritional status and remained asymptomatic.

Due to the presence of recurrent episodes of IAP and the evolution towards CP, genetic testing of CFTR, PRSS1 and SPINK1 genes was performed. A homozygous mutation (N34S) in exon 3 of the SPINK1 gene was identified. Currently other family members are being tested. No family history of pancreatitis has been reported.

Discussion

We present the case of a child with a homozygous mutation N34S in the SPINK1 gene, leading to ARP and ultimately to CP with EPI.

Several pediatric series have described the high prevalence of genetic mutations in IAP (1-5), the most common involving PRSS1, CFTR and SPINK1 genes.
Genetics involving SPINK1 are complex; heterozygous mutations act as a disease modifier lowering the threshold for developing pancreatitis from other genetic/structural abnormalities (2-4), but in the rare cases of homozygous inheritance (1), as in our case, SPINK1 mutations may be the sole cause for unleashing the event.

Children with acute and ARP at an early age should be closely followed up as they are more likely to progress towards chronic disease, thus genetic testing should be considered in patients younger than 30 years with IAP and AP.

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References