Rare disease

Abdominal pain in a 5-year-old boy

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Summary

A 5-year-old male presented with a 1-day history of vomiting, epigastric pain, loose stools and poor oral intake. The child had intermittent symptoms of abdominal pain and vomiting for the past 2 years and there was a strong family history of chronic hereditary pancreatitis. A random serum amylase was elevated at 2320 IU/l. Abdominal ultrasound scan revealed no abnormalities. On the basis of the family history, positive symptoms and elevated amylase, a diagnosis of hereditary pancreatitis was made. The child received symptomatic management and treatment for his acute illness. He is currently awaiting results from molecular testing.

BACKGROUND

When this child presented to the Paediatric ward, none of my colleagues were aware of another child with hereditary pancreatitis. I performed a literature search via Medline which revealed that this is a rare disorder with little published research available. There is a need for more research in this area to help understand the overall disease mechanism and to explain the incomplete penetrance. At present, there is no data regarding the incidence or prevalence of chronic pancreatitis in children.

CASE PRESENTATION

A 5-year-old male presented to the Accident and Emergency department with a 1-day history of vomiting, epigastric pain, loose stools and poor oral intake. The history further revealed that the child had been having intermittent symptoms of abdominal pain and vomiting for the past 2 years and had previously had a few admissions to the Paediatric ward for gastroenteritis type illnesses. His medical history and birth history were otherwise unremarkable and he was up-to-date with all of the recommended vaccinations. There was a strong family history of chronic hereditary pancreatitis. His mother was diagnosed with idiopathic familial pancreatitis at the age of 16 years, although she had been symptomatic since the age of 7 years; and had later gone on to develop insulin-dependent diabetes mellitus at age 20. His maternal grandfather had developed symptoms of pancreatitis as an adolescent and died at the age of 37 years from complications related to chronic pancreatitis.

On examination, the child was in obvious pain and looked distressed. The abdomen was soft but tender in the epigastric region with some guarding. Bedside observations were all within the normal limits. His height was between the 50th and 75th centile and weight was on the 9th centile for his age.

INVESTIGATIONS

Serum amylase was elevated at 2320 IU/l. Abdominal ultrasound scan showed no abnormalities. The results from molecular testing are currently being awaited.

DIFFERENTIAL DIAGNOSIS

On the basis of the family history, positive symptoms and elevated amylase, a diagnosis of hereditary pancreatitis was made.

TREATMENT

The child received symptomatic management and his acute illness was treated. The child is currently being managed symptomatically with Creon and dietary modification.

OUTCOME AND FOLLOW-UP

A referral was made to the regional Genetics Centre for further investigation and follow-up.

He is currently awaiting results from undergoing molecular testing for *PRSS1*, *SPINK1* and *Chromotrypsin C* genes to look for an underlying mutation.

DISCUSSION

A review article published by Rosendahl *et al* found that the diagnostic criteria for hereditary types of chronic pancreatitis has changed over the years and that there are different classifications depending on the family lineage of the disease.¹ Due to this ambiguity with the classification of chronic pancreatitis, the authors use the umbrella term hereditary chronic pancreatitis (HCP) to cover all forms of hereditary pancreatitis. HCP is defined as chronic pancreatitis with no detectable cause with the presence of one first or second degree relative with proven chronic pancreatitis.¹

First identified by Comfort and Steinberg in 1952²; HCP has been described as a very rare form of early onset chronic pancreatitis. Studies of affected families have demonstrated an autosomal dominant pattern of inheritance with a penetrance of 80%.¹⁻³ Genetic linkage studies by Le Bodic *et al*⁴ and later by Whitcomb *et al*,⁵ have found that the hereditary pancreatitis locus has been found to lie within the long arm of chromosome 7.³ The clinical course, morphological features and laboratory findings are similar to those of alcoholic chronic pancreatitis making hereditary pancreatitis indistinguishable from other causes of acute and chronic pancreatitis.¹⁻³ The defining feature

of hereditary pancreatitis is the early onset of the disease, with acute attacks of pancreatitis, usually beginning in childhood.³ In affected children, the cardinal symptom is that of recurring, sudden onset epigastric pain which may be accompanied by nausea, vomiting and abdominal pressure pain.¹ Children with hereditary pancreatitis may go on to develop insulin-dependent diabetes and pancreatic insufficiency.

Diagnosis is mainly from clinical findings and the presence of a positive family history of chronic pancreatitis. Pancreatic function tests can be used to aid diagnosis with invasive pancreatic function tests in combination with the finding of pancreatic calcification, is the current gold standard in diagnosis, although there is a call for direct DNA sequencing of both strands to identify the culprit gene mutation.¹

Several gene mutations have been identified as playing a causative role in the development of HCP, in particular, mutations of the *PRSS1* gene and protease inhibitor Kazal type 1 (*SPINK1*).⁶ Mutations in these genes have been found to contribute to the development of chronic pancreatitis by altering the balance of pancreatic proteases and inhibitors creating an excess of intrapancreatic trypsin, leading to autodigestion of the pancreas.

At present, the mainstay of management remains that of symptomatic control, achieved through dietary modification and pain management; and the treatment of disease complications such as endocrine failure, pseudocysts, duodenal obstruction, bile duct obstruction, diabetes and maldigestion.¹⁻³ Patients with chronic pain, secondary to persistent pancreatic duct dilatation may opt to have a longitudinal pancreaticojejunostomy, which has proved to have good early results. Patients with a diagnosis of HCP have a 50-fold elevated risk of pancreatic cancer in comparison to the general population thus patients should be advised to completely avoid smoking and keep alcohol intake to a minimum to limit disease progression.

At present, there is no data regarding the incidence or prevalence of chronic pancreatitis in children.¹ In short, more research needs to be performed to look at this rare disorder and help explain the incomplete penetrance and overall disease mechanism which is still not fully understood.

Learning points

- ▶ Pancreatitis should be considered as a differential diagnosis in children with unexplained recurrent abdominal pain
- ▶ More research needs to be conducted to the area of hereditary pancreatitis as the disease mechanism is not fully understood
- ▶ Patients diagnosed with hereditary pancreatitis need to be advised of lifestyle factors such as the avoidance of alcohol and smoking, due to the increased risk of pancreatic cancer.

Competing interests None.

Patient consent Obtained.

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