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Case Study

A Case Report on Hereditary Pancreatitis

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ABSTRACT

Hereditary pancreatitis is an autosomal dominant condition that causes recurrent attacks of acute pancreatitis which can progress to chronic pancreatitis often at young stage. It was first described in the year 1952 by multiple genetic defects that affect the action of digestive enzyme in pancreas. Most of the patients with hereditary pancreatitis express the two mutations [R122H or N291] in cationic trypsinogen gene [PRSS1 gene]. A 17-year-old female patient was admitted to the emergency department with rapid onset of abdominal pain of one day duration. Pain was moderately severe and associated with nausea. Patient had previous history of acute pancreatitis once in the past. Her father was also having similar history of pancreatitis. The laboratory findings show an elevated amylase [519] and lipase [4653] levels. She was treated with Emeset 4 mg, pantoprazole 40 mg and panlipase during that period. Clinical manifestation includes recurrent attacks of severe abdominal pain. In addition to positive family history patients with hereditary pancreatitis tend to develop chronic pancreatitis such as strictures and fluid collections as well as exocrine and endocrine insufficiency. This case points out affected patients need to be individualized with an emphasis on early diagnosis and multidisciplinary involvement to develop a comprehensive treatment strategy.

INTRODUCTION

Hereditary pancreatitis is a rare cause of chronic pancreatitis, acute and recurrent acute pancreatitis. It was first described by comfort and Steinberg in 1952. It occurs similarly to other causes of pancreatitis such as alcohol, gallstones, and autoimmune causes. Symptoms usually develop in childhood or adolescence stage with time. Acute attacks of abdominal pain occur early in life and the disease often progress to chronic pancreatitis, in addition to positive family history [1]. The diagnosis of hereditary pancreatitis is usually delayed as many individuals labeled as having an idiopathic condition. Advance in genetic testing have enhanced the ability to detect inherited mutations that allows more precise characterization of hereditary pancreatitis [2].

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Hereditary pancreatitis is due to mutations in PRSS1 gene on long arm of chromosome 7 that encodes for cationic trypsinogen. R122H, N291 and A16V are the three more common mutations. One of the protective mechanisms includes the existence of arginine residue at 122 [R122] position in PRSS1 gene that allows autolysis of trypsin and prevent pancreas auto digestion[3]. A recurrent pancreatic injury and inflammation increases the risk of more adverse late complications of hereditary pancreatitis such as pancreatic cancer, pancreatic fibrosis, pancreatic exocrine insufficiency and pancreatic duct carcinoma[3,4]. The risk of pancreatic cancer increases by 40% by age of 70. Management should be focused on counseling patients to refrain from alcohol consumption and use of medications associated with pancreatitis[4]. Antioxidant therapies prone to reduce frequency of attacks. The patient who develops pancreatic insufficiency should receive pancreatic replacement and supplementation with fat soluble vitamins[5]. Chronic pain is difficult to manage and often requires the long term use of analgesic. Psuedocyst may either resolve spontaneously or may require certain interventions such as drainage procedures[5,6]. Pancreatic duct strictures and stones can be effectively managed by balloon dilation and stenting during ERCP[6]. For certain cases surgical interventions such as modified puestow procedure were used to maintain pain.

Case Report

The reports investigate a case of hereditary pancreatitis. A 17 year old female patient was admitted to the emergency department with rapid onset of abdominal pain of one day duration. Pain was moderately severe and associated with nausea. Patient had previous history of acute pancreatitis once in the past. Her father was also having similar history of pancreatitis. The laboratory findings show an elevated amylase [519] and lipase [4653] levels. LFT, TRG and calcium levels were normal. The patients USG abdomen and pelvis shows features of interstitial pancreatitis with minimal ascites and traces of bilateral pleural effusion. These findings supported the diagnosis of hereditary pancreatitis. She was treated with Emeset 4 mg, pantoprazole 40 mg and panlipase during that period. Her vitals was stable with heart rate of 78bpm, a temp 98 °F, BP of 130/70 mm Hg and respiratory rate of 20 Breaths/min on physical examination. After resolving the acute episodes the patient was discharged and monitored closely.

DISCUSSION

Hereditary pancreatitis is an autosomal dominant disease. In hereditary pancreatitis the acute attacks usually begins in childhood, but the age of onset usually ranges from infancy to fifth or sixth decades of life[7]. Chronic pancreatitis involves recurrent attacks of acute pancreatitis which can lead to certain complications such as pancreatic cancer, pancreatic fibrosis, pancreatic duct carcinoma and maldigestion[8]. Over the past decades many case reports have highlighted the genetic, clinical and diagnostic variability of hereditary pancreatitis, providing important insights into progression, treatment approaches and long term outcomes[9]. For instance, a case reported by *Rebours et al* described a 7-year-old patient with recurrent abdominal pain who was eventually diagnosed with HP due to a PRSS1 gene mutation. Similarly, a case by *Howes et al* documented pancreatitis beginning before the age of 10 in multiple members of the same family. Several case reports stress that the absence of a family history often leads to delayed or missed diagnosis, while its presence can be an early diagnostic clue. For example, a report by *Chen et al* described a 13-year-old boy with recurrent acute pancreatitis who was misdiagnosed for years until

a detailed family history revealed that his father and paternal aunt had similar symptoms in adolescence. Genetic testing then confirmed a PRSS1 R122H mutation. This underscores the diagnostic value of meticulous pedigree analysis in young patients presenting with unexplained recurrent abdominal pain or pancreatitis[10]. In fact, across many reported cases, symptoms begin in the second decade of life, with no clear precipitating factors like alcohol or gallstones. This was exemplified in a report by *Joergensen et al.* (2010), where a 16-year-old female experienced multiple episodes of pancreatitis, and only after probing further into family history was it revealed that her grandfather had undergone a pancreatectomy for chronic pancreatitis in his 40s—information that ultimately led to targeted genetic testing and confirmation of HP. In the 10–20 age group, patients often experience a high burden of recurrent acute episodes, which may transition to chronic pancreatitis within a few years if not correctly managed[11]. A multicase study by *Weiss et al* followed adolescent patients diagnosed with HP due to PRSS1 mutations and found that over 60% developed exocrine or endocrine insufficiency before age 25. Early diagnosis, enabled largely by recognition of familial patterns, was associated with better long-term outcomes through early interventions such as lifestyle changes, enzyme supplementation, and pain management. Therefore, in adolescents and young adults with recurrent pancreatitis, especially those without common risk factors, detailed family history should be considered as vital as lab and imaging studies[12]. This not only aids diagnosis but also prompts genetic counseling and early surveillance in asymptomatic family members. Early identification and intervention can potentially delay the onset of complications, reduce hospitalizations, and improve quality of life during crucial developmental years[13, 14].

CONCLUSION

The case report highlights the importance of genetic testing. Hereditary pancreatitis is an autosomal dominant condition associated with genetic risk factors[15]. Mutations in the cationic trypsinogen gene are the most frequently observed characteristics in this condition[15]. Mutations in cationic trypsinogen should be considered in individuals with presenting with repeated episodes of acute or chronic pancreatitis particularly when there is a family history of pancreatic disease[16]. Genetic counseling and early detection of pancreatic cancer are critical for managing these patients along with close monitoring[17].

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