The first hereditary pancreatitis family in Hungary

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INTRODUCTION

Determination of the etiology of recurrent acute pancreatitis is often difficult.

ETIOLOGY

Children

- Idiopathic 26.9%
- Positive family history/positive genetics/altered sweat test 42.3%
  - CFTR 39.6%
  - SPINK1 7.1%
  - **PRSS1 4.5%**
- Structural abnormalities 19.2%
- Biliary lithiasis 6.5%
- Others 5.1% (polycystosis, drug, dyslipidemia)

V Lucidi et al. The etiology of acute recurrent pancreatitis in children: a challenge for pediatricians. *Pancreas* 2011; 40:517-521.

Adults

- Choledocholithiasis, Alcohol abuse } 70%
- Autoimmune disease
- Congenital anomaly
- Duodenal obstruction
- Genetic
- Idiopathic
- Infection
- Medication
- Metabolic
- Neoplasm
- Sphincter of Oddi dysfunction
- Tropical
- Vasculitis

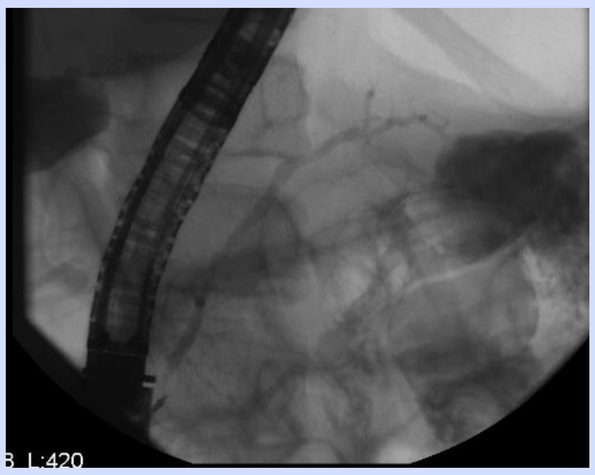
LF Lara et al. Idiopathic Recurrent Acute Pancreatitis. *MedGenMed*. 2004; 6:10.

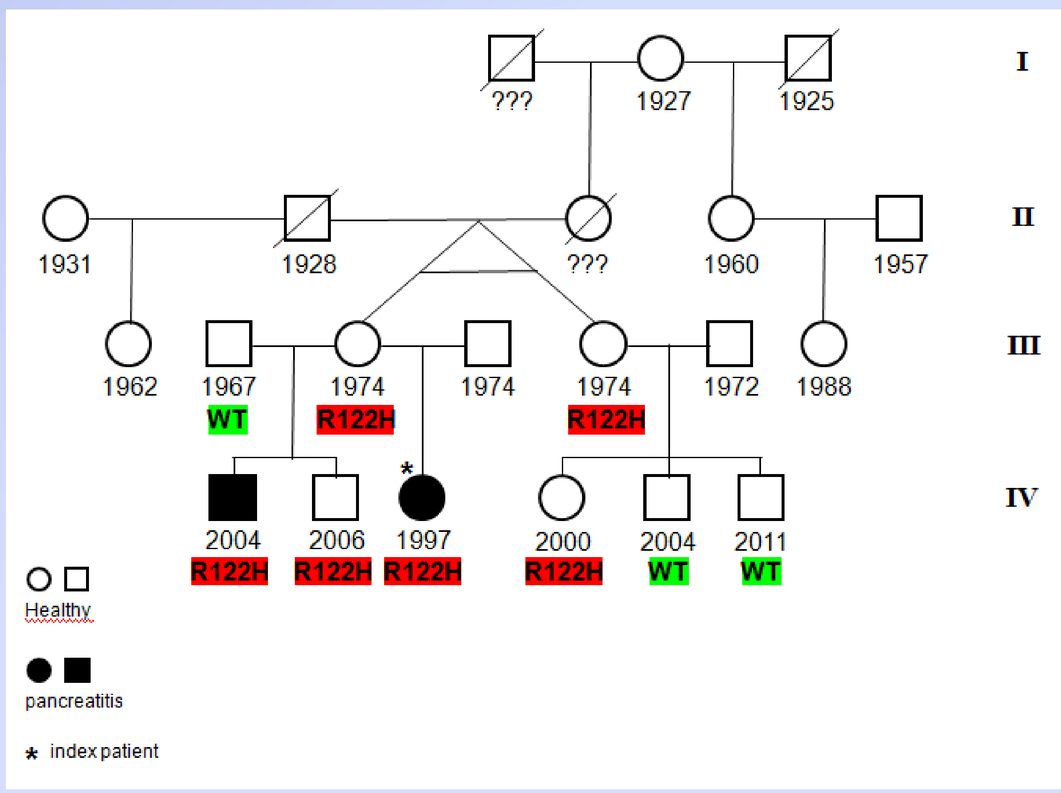
CASE DESCRIPTION

A fourteen-year-old girl had had acute pancreatitis on ten occasions, the first episode occurring at the age of four years, with subsequent recurrences at six to nine-month intervals apart from a four-year-long silent period at the age of six years.

The pancreatitis was edematous in all cases, but on two occasions intensive care was needed. A viral infection was suspected in the etiology, but the laboratory and morphological work-up did not identify any etiological factor (e.g. bile stone, dietary abuse, anatomical malformations, hyperlipidemia, cystic fibrosis or autoimmune pancreatitis) either then or later.

After the third recurrence, the abdominal MRI indicated chronic pancreatitis, while ERCP following the fifth attack demonstrated changes in the caliber of the main pancreatic duct.





Genetic examination revealed heterozygous mutation R122H in the cationic trypsinogen gene (PRSS1 gene) and genetic screening in the family showed the same mutation in her two stepbrothers, her mother, her mother's twin sister and the twin sister's daughter, though acute pancreatitis had occurred in only one of the stepbrothers.

Genetic examination of N34S mutation in SPINK1 (trypsin inhibitor) gene and of CTRC (chymotrypsin C) gene did not show any mutation either in the patient or in her two stepbrothers.

DISCUSSION

Hereditary pancreatitis is an autosomal dominant condition where recurrent attacks of acute pancreatitis lead to chronic pancreatitis. In most of the cases, the causative factor is a point mutation in the cationic trypsinogen gene (R122H: Arg117 → His or N29I: Asn21 → Ile), which results in the enhanced autoactivation of trypsinogen and decreased autocatalytic degradation of trypsin.
Acute and particularly recurrent pancreatitis at a young age should suggest the possible role of genetic factors, even if the family history is not typical.

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