

# MANAGEMENT OF YOUNG PATIENTS WITH RECURRENT EPISODES OF ACUTE PANCREATITIS

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## ABSTRACT

### Hereditary pancreatitis (HP)

- Recurrent episodes of acute pancreatitis
- Chronic pancreatitis
- Multidisciplinary evaluation
- Personalized management
- High risk of pancreatic cancer

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## INTRODUCTION

**Hereditary pancreatitis (HP)** is a rare genetic condition, the evolution of which is marked by **recurrent episodes of acute pancreatitis** that begin in childhood or adolescence and lead to the early inception of **chronic pancreatitis** and a significant increase in the **risk of pancreatic cancer** in young adults.

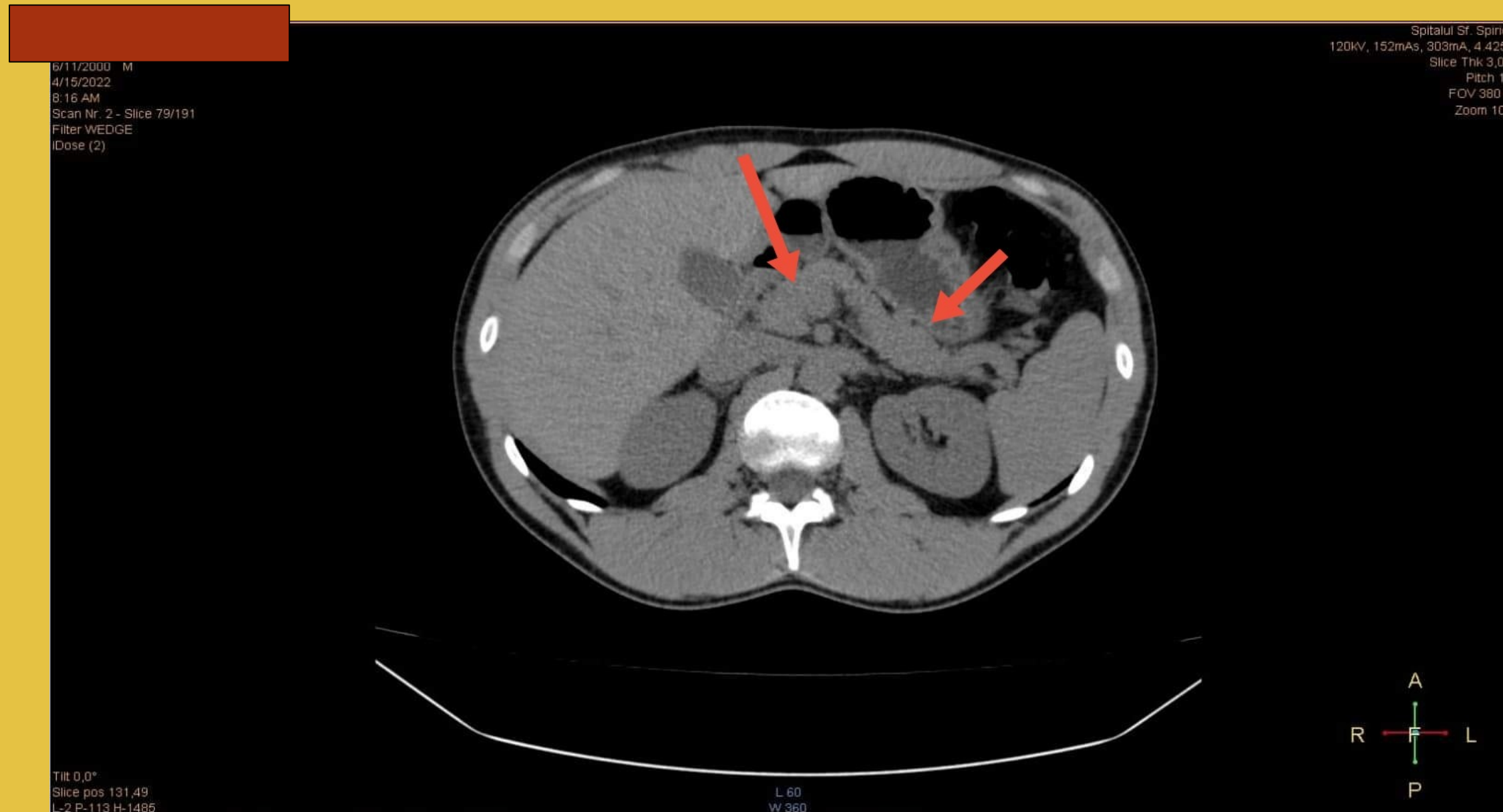


Figure 1. Sclerolipomatous pancreas

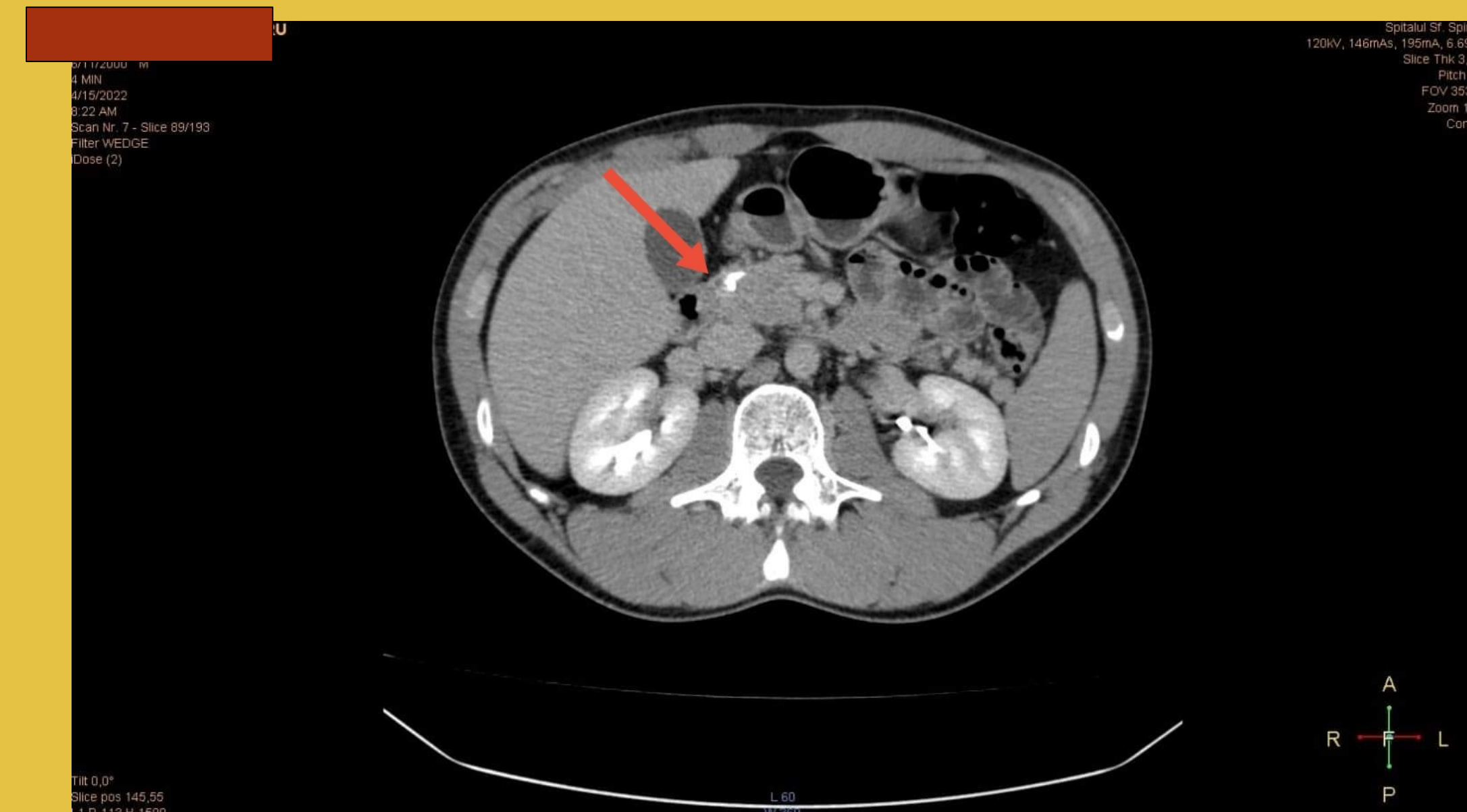


Figure 2. Pancreatic calcification

## CASE DESCRIPTION

We present the case of a 22-year-old patient, with repeated appearances in the gastroenterology department for **episodes of acute pancreatitis** (11 episodes), started at the age of 15, for which recent computed tomography scan (2022) showed **changes suggestive for chronic pancreatitis** (Figure 1,2).

➤ **Anamnestic and laboratory tests** have ruled out *alcohol use, medications, dysmetabolic syndrome*, as the most common causative factors involved in triggering episodes of acute pancreatitis.

➤ **The echo-endoscopic investigation** with biopsy was negative for specific markers of autoimmune pancreatitis.

Given the positive history of acute pancreatitis (multiple episodes) of relatives (father and uncle), the presence of inherited **genetic mutations** was taken into account.

➤ **The result of the genetic panel** revealed the presence of a pathogenic variant probably heterozygous in the PRSS1 gene associated with hereditary pancreatitis (Figure 3).

**Clinical Information:** Patient with recurrent episodes of acute pancreatitis. The etiological autoimmune cause was excluded, metabolic or toxic. Relevant family history: father and uncle have history of recurrent episodes of acute pancreatitis.

### RESULT AND INTERPRETATION

The presence of a heterozygous likely pathogenic variant in the PRSS1 gene associated with hereditary pancreatitis has been identified. (See Recommendations)

Gene	Variant*	Zygoty	Inheritance pattern	Classification^
PRSS1	NM_002769.5:c.311T>C p.(Leu104Pro)	Heterozygosis	Autosomal dominant	Likely Pathogenic

Figure 3. Hereditary pancreatitis – genetic tests

## CASE MANAGEMENT

The case was included in an individualized management program with **multidisciplinary involvement**, with a focus on pain management, medical therapy for endocrine and exocrine insufficiency, and surveillance of the sequelae of chronic pancreatitis and pancreatic adenocarcinoma.

## CONCLUSIONS

➤ The recurrence of acute pancreatitis episodes at a young age, with negative results for the causes frequently involved in the onset of acute episodes (toxic, metabolic, autoimmune), but with a positive family history of acute pancreatitis, justifies the extension of genetic testing investigations to establish an **early diagnosis** on hereditary pancreatitis.

➤ Given that there are no clearly established methods for preventing the development or progression of the disease in the context of the presence of a genetic mutation associated with HP, the emphasis in case management will focus on avoiding **the triggers** that can exacerbate and aggravate pancreatitis and monitoring the progression toward **adenocarcinoma pancreatic**.