Hereditary Pancreatitis

A Practical Guide For Patients



GastroDoxs

defenders of the digestive system

A Patient Guide to Understanding Cardiac Cirrhosis

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Introduction: Hereditary Pancreatitis

Hereditary pancreatitis is an inherited condition that causes repeated episodes of inflammation in the pancreas. The term generally refers to pancreatitis caused by a disease-causing change in the PRSS1 gene. Symptoms often begin during childhood, adolescence, or early adulthood, although the age and severity can vary considerably.
Patients may experience recurrent acute pancreatitis and later develop chronic pancreatitis with permanent pancreatic damage. Over time, complications can include persistent abdominal pain, difficulty digesting food, greasy stools, vitamin deficiencies, diabetes, pancreatic duct abnormalities, and an increased risk of pancreatic cancer. Diagnosis involves documenting pancreatitis, reviewing the family history, excluding other causes, and completing genetic testing. Treatment focuses on controlling attacks, preserving nutrition, managing complications, and reducing preventable risks.

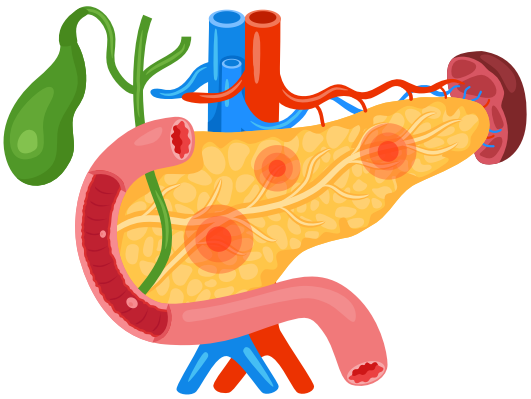
Key Takeaways

- Hereditary pancreatitis is an inherited tendency to develop recurrent pancreatic inflammation.
- The condition most commonly refers to pancreatitis caused by a pathogenic PRSS1 variant.
- Symptoms often begin during childhood or early adulthood.
- Patients may experience repeated episodes of acute pancreatitis.
- Recurrent attacks frequently progress to chronic pancreatitis.
- Upper abdominal pain may spread into the back.

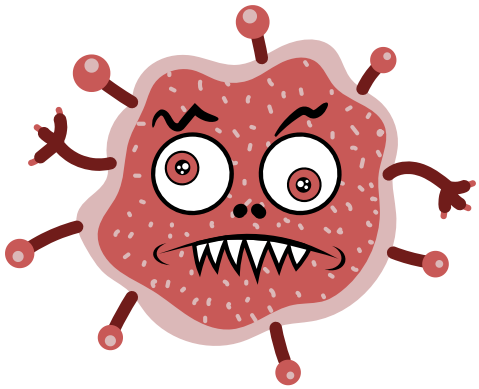
What Causes Hereditary Pancreatitis?

Known Causes:

Hereditary pancreatitis usually results from an inherited change in the PRSS1 gene, which provides instructions for producing cationic trypsinogen. Certain variants allow trypsin to become active too early or remain active too long inside the pancreas. This can cause the pancreas to begin digesting and injuring its own tissue, leading to repeated inflammation.



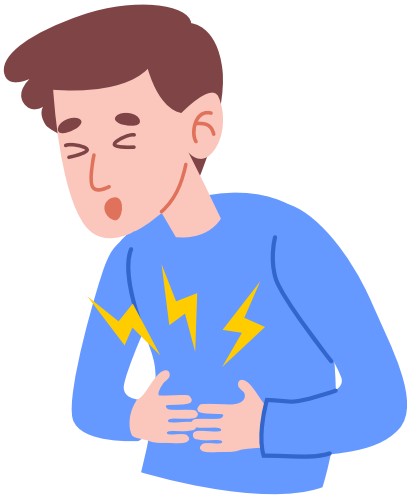
- **PRSS1 pathogenic variants:** Disease-causing changes in this gene are the primary cause of classic hereditary pancreatitis.
- **Premature trypsin activation:** Digestive enzymes may become active inside the pancreas instead of after reaching the small intestine.
- **Autosomal dominant inheritance:** One altered copy of the PRSS1 gene can be sufficient to increase disease risk.



Common Signs & Symptoms

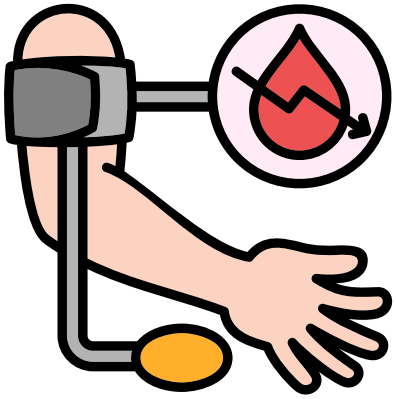
Symptoms may occur as separate attacks with symptom-free periods in between. Acute episodes commonly cause intense upper abdominal pain, nausea, and vomiting. As the pancreas becomes permanently damaged, patients may develop chronic pain, digestive problems, weight loss, or diabetes.

Core Symptoms Include:



- Upper abdominal pain
- Pain that spreads into the back
- Pain that becomes worse after eating
- Nausea with or without vomiting
- Abdominal tenderness or swelling
- Fever during an acute episode
- Rapid heartbeat

Severe cases can also cause:



- Severe or persistent abdominal pain
- Repeated vomiting and dehydration
- Low blood pressure or shock
- Pancreatic tissue damage or necrosis
- Infected pancreatic collections
- Pancreatic pseudocysts
- Blockage of the pancreatic or bile duct

Diagnosis & Medical Testing

Diagnosis combines evidence of acute, recurrent, or chronic pancreatitis with family history and molecular genetic testing. PRSS1-related hereditary pancreatitis is confirmed when a person with an appropriate pancreatitis history has a pathogenic PRSS1 variant. A positive genetic result in a person without symptoms does not, by itself, establish that active pancreatitis is present.

Diagnostic Tools:

- **Medical history evaluation:** Reviews the age symptoms began, attack frequency, pain pattern, hospitalizations, and previous pancreatic complications.
- **Family history:** Identifies relatives with pancreatitis, unexplained abdominal attacks, diabetes, pancreatic insufficiency, or pancreatic cancer.
- **Physical examination:** Checks abdominal tenderness, swelling, dehydration, jaundice, fever, and nutritional status.
- **Lipase and amylase testing:** Detects pancreatic enzyme elevation during an acute attack.
- **Complete blood count:** Checks for infection, inflammation, anemia, or other abnormalities.
- **Metabolic panel:** Evaluates electrolytes, kidney function, liver function, dehydration, and blood glucose.
- **Triglyceride and calcium testing:** Excludes other causes of recurrent pancreatitis.

Diet

Diet cannot change the inherited PRSS1 variant, but nutrition can support pancreatic function and reduce complications. Recommendations depend on whether the patient is recovering from an acute attack, has chronic pain, or has pancreatic enzyme insufficiency. Patients with weight loss or malabsorption should receive individualized guidance rather than following an excessively restrictive diet.

Key Insights:

- Avoid all alcohol because it can trigger attacks and accelerate pancreatic damage.
- Drink adequate fluids throughout the day.
- Eat smaller, more frequent meals if large meals increase pain or fullness.
- Follow a healthy eating plan that is moderate or low in fat when medically recommended.
- Avoid very high-fat or greasy meals if they worsen symptoms.
- Include adequate protein to maintain muscle and support recovery.
- Maintain sufficient calories to prevent unintended weight loss.





Medications & Treatment Options

There is no medication that corrects the inherited genetic change. Treatment is directed at acute attacks, chronic pain, impaired digestion, diabetes, and other complications. Care is often coordinated among gastroenterology, genetics, nutrition, endocrinology, pain management, endoscopy, and pancreatic surgery specialists.

Treatment Options

- **Intravenous fluids:** Treat dehydration and support circulation during acute pancreatitis.
- **Oral or enteral nutrition:** Provides early nutritional support when eating is safe.
- **Pain medication:** Controls acute or chronic pancreatic pain using an individualized plan.
- **Anti-nausea medication:** Relieves nausea and vomiting.
- **Antibiotics:** Treat a confirmed infection but are not routinely used for uncomplicated pancreatic inflammation.

Advanced Therapies & Procedures

Advanced treatment may be required when chronic pancreatitis causes pancreatic duct blockage, stones, pseudocysts, persistent pain, malnutrition, or other complications. The most appropriate procedure depends on pancreatic anatomy, symptom severity, previous treatments, diabetes status, and overall health.

Therapy Options:

- **Endoscopic ultrasound:** Evaluates chronic pancreatitis, pancreatic lesions, cysts, and duct abnormalities.
- **ERCP therapy:** Treats selected pancreatic or bile-duct narrowing and obstruction.
- **Pancreatic duct stenting:** Helps improve drainage through a narrowed duct.
- **Stone extraction:** Removes pancreatic duct stones causing obstruction or pain.
- **Shock-wave lithotripsy:** Breaks larger pancreatic duct stones into pieces that can be removed.
- **Pseudocyst drainage:** Treats symptomatic, infected, enlarging, or complicated pancreatic fluid collections.
- **Celiac plexus block:** Temporarily reduces pain signals from the pancreas in selected patients.
- **Surgical drainage:** Relieves pressure within a dilated or obstructed pancreatic duct.

Coping With Hereditary Pancreatitis

Living with hereditary pancreatitis may involve unpredictable attacks, chronic pain, dietary challenges, hospital visits, and concern about relatives. A long-term care plan can help patients reduce avoidable triggers, maintain nutrition, recognize complications, and understand their inherited risk.

Genetic counseling is important because the condition may affect children, siblings, and other relatives. Family members should not assume that they are affected based only on symptoms or family history; testing should be guided by a genetics professional.

Practical Strategies



- Attend regular gastroenterology and pancreatic specialist appointments.
- Keep copies of genetic testing, imaging, and hospitalization records.
- Avoid tobacco products and secondhand smoke.
- Avoid alcohol completely.
- Maintain adequate hydration, particularly during exercise or hot weather.
- Take pancreatic enzymes with every prescribed meal and snack.
- Monitor stool changes, weight, appetite, and pain patterns.





FAQS

Q1: What is hereditary pancreatitis?

A: Hereditary pancreatitis is an inherited condition that causes recurrent pancreatic inflammation, often beginning during childhood or early adulthood. Classic hereditary pancreatitis is generally associated with a pathogenic gain-of-function variant in the PRSS1 gene.

Q2: What causes hereditary pancreatitis?

A: It is usually caused by a disease-causing PRSS1 gene variant that interferes with the normal regulation of pancreatic digestive enzymes. Classic PRSS1-related hereditary pancreatitis is commonly inherited in an autosomal dominant pattern, meaning each child of an affected person has a 50% chance of inheriting the variant.

Q3: How is hereditary pancreatitis diagnosed?

A: Diagnosis involves confirming acute, recurrent, or chronic pancreatitis; reviewing the family history; excluding other causes; and completing molecular genetic testing. PRSS1-related hereditary pancreatitis is established when an affected patient has a pathogenic PRSS1 variant.

Q4: How is hereditary pancreatitis treated?

A: Treatment may include hospital care during acute attacks, pancreatic enzyme replacement, nutritional support, pain management, diabetes treatment, endoscopic procedures, or surgery. Patients should avoid tobacco and alcohol and may require specialist pancreatic cancer surveillance.

Take Action

If you or several relatives have experienced recurrent pancreatitis, unexplained pancreatic disease at a young age, chronic pancreatic pain, pancreatic insufficiency, or pancreatic cancer, do not ignore the family pattern. Genetic evaluation may clarify the cause and guide treatment, family counseling, and long-term surveillance.

Schedule a consultation with GastroDoxs for expert guidance on evaluating and managing hereditary pancreatitis.

Your health starts with knowledge.
Take the first step today!

SCHEDULE AN APPOINTMENT



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