Rare Cause of Pancreatitis in a 20-year-old Female

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Eighty percent of pancreatitis cases are caused by alcohol or gallstones. Some medications such as HIV drugs, diuretics, anticonvulsants and estrogen may cause pancreatitis. When no obvious cause can be identified, a genetic one should be considered.

Case Report
A 20-year-old woman presented to urgent care with dull, non-radiating abdominal pain in the right upper quadrant that lasted for one day. She was treated symptomatically with ibuprofen and a gluten-free diet. Three days later, she presented to the emergency department with the same symptoms. Her past history was notable for “stomach problems” throughout childhood. She had been diagnosed with irritable bowel syndrome and had undergone a colonoscopy, which was normal. Her lipase was elevated at 237 (normal to 120), and she was diagnosed with acute pancreatitis and admitted to the hospital for management. She reported having no more than eight to 10 alcoholic drinks in her lifetime. Her medications included a combination oral contraceptive and ibuprofen as needed. Her lipid panel on admission was normal.

During her hospital stay, abdominal ultrasound, esophagogastroduodenoscopy (EGD) and magnetic resonance cholangiopancreatography (MRCP) studies were done and results were normal. After 24 hours of IV fluids and NPO status, her lipase improved and the pain resolved. She presented to our family medicine clinic two weeks after hospital discharge.

We elected to pursue genetic testing in this patient. A genetic panel looking at the three different inheritance patterns of pancreatitis (CFTR, SPINK1, PRSS1) was done. Our patient was found to be heterozygous for mutations in the CFTR gene, which predisposed her to pancreatitis. Because of the complexities involved, the patient was referred to a pancreatitis center for ongoing management.

Discussion
This young patient had no classic risk factors for pancreatitis, and her case illustrates the value of considering genetic causes of pancreatitis. It is important to identify patients with hereditary pancreatitis because nearly 50% of them will develop type 1 diabetes by mid-adulthood. Their risk for pancreatic cancer is also elevated, especially among those who also smoke, use alcohol, have type 1 diabetes, or have a family history of cancer.

REFERENCES