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ABSTRACTS

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Diagnosis of familial mediterranean fever masked by symptoms of chronic pancreatitis

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Familial mediterranean fever (FMF) is the most common hereditary periodic fever syndrome in populations of Mediterranean origin (Arabs, Turks, Jews, Lebanese, Greeks, etc.). The frequency of heterozygous carriers of MEFV, responsible for the development of FMF, is more than 1/5 of the total population. Case report: 22-year-old Lebanese student of Kharkiv National Medical University complained of acute pain in mesogastrium, nausea and fatigue. Similar episodes had occurred two years earlier, and the diagnosis of chronic pancreatitis was made. Given the cyclical nature of pain and the Mediterranean origin of the patient, molecular genetic testing was performed. by a combination of restriction fragment length analysis and allele-specific hybridization methods (Tibnin Governmental Hospital). The patient is compound heterozygous for the mutations M694I and M694V in MEFV exon 10. The mutations V726A, E1480Q, M680I, P369S, R408Q, A744S, M680Ib, R761H, R65, F479L, E167D, K695R, and I692del were excluded. FMF was diagnosed in this patient with symptoms of chronic pancreatitis. It is known that the severity of clinical signs may be decreased in heterozygous carriers of mutant alleles. However, given the risk of FMF-associated amyloidosis, rheumatologist clinical supervision and control of serum amyloid were recommended.