✪ A new genomic mutation in the mediterranian familiar fever

D. Siclari, M. Allò, M.C. Calindro, R. Costa, E. Corigliano, A. Galdieri, M. Falbo, M. Nocera, G. Perrone, G.F. Mauro

Unit of Internal Medicine, "San Giovanni di Dio" Hospital, Crotone, Italy

Familial Mediterranean Fever is an inherited disease characterized by recurrent fever, peritonitis and, sometimes other sierosites or skin lesions. Transmission is due to an autosomal recessive mechanism. The gene responsible is allocated on chromosome 16 and encodes for a protein of 781 aminoacids, called Marenostrina, expressed only by the circulating neutrophils. The disease is more often present in subject of Mediterranean origin and, about 50%, of patients have a family history. The onset of symptoms is usually present before the age of 15, but, frequentely, it does not have a regular occurrence, varying in the same patient for frequency from a few weeks to years. The possible complication, related to the genotype, is the development of amyloidosis. We present the case of a 23 years old woman came to our hospital for fever and abdominal pain. One of the two brothers was suffering from similar symptoms. In anamnesis there were recurrent episodes, sine causa, of fever from the age of three years accompanied by abdominal and joint pain. The molecular analysis for Familial Mediterranean Fever has identified two different mutations in heterozygous M694 and R761H in the gene encoding the Marenostrina. This genetic pattern confirms the diagnosis of Familial Mediterranean Fever and it is the first described mutation with this genetic pattern for such disease. At the moment it is not possible to know whether this genetic pattern can evolve in the development of amyloidosis.