SUMMARY: Familial Mediterranean fever (FMF) is an inherited disorder characterized by fever, abdominal and/or chest pain. The etiology and pathogenesis of the disease remain unknown. A large amount of amorphous urate excretion in urine was found in five FMF patients during acute episodes. Increased excretion of uric acid in the patients with FMF during acute episodes can lead to the thesis that FMF is an inherited disorder of purine metabolism.

Key Words: FMF, purine metabolism, inherited disease.
trifugation of the urine, amorphous urate was also seen in the sediment. The characteristic of the urine returned to normal after resolution of the attack. This change of urine has been uncovered in numerous attacks observed in these 5 patients. Serum uric acid level however remained normal during acute attacks in these patients.

In humans, uric acid is the ultimate catabolite (end product) of purines. Reasoning from observations made in humans with inherited enzyme deficiencies, it appears that over 99% of the uric acid derived from substrates of purine nucleosid phosphorylase, a component of the purine salvage pathway. Sodium urate is a monosodium salt of uric acid (6,7).

The best known purine metabolism disorder is gout. This disease is mainly manifested by an increase in the uric acid level and recurrent attacks of acute arthritis. Colchicine is used effectively during the acute gout attack (6,7,14). It is well known that colchicine strikingly reduces the incidence of symptomatic febrile episodes, the mechanism of amelioration however remains unclear (3, 4, 11, 14).

Demonstration of large amounts of urates in urine present in FMF patients during acute attacks has led us to hypothesize that it may be a variant of inherited disorder of purine metabolism, which under other conditions may manifest itself as gout disease. While excessive production of urate was observed in FMF patients during acute attacks, questions about the nature and extent of the metabolic defect in purine metabolism responsible for FMF and its spontaneous resolution remain to be answered. Nevertheless it should be recalled that spontaneous resolution of the acute attacks are also seen in the gouty subjects (14).

REFERENCES


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