Scholars Journal of Medical Case Reports
Sch J Med Case Rep 2015; 3(1):25-26
ISSN 2347-6559 (Online)
ISSN 2347-9507 (Print)

(An International Publisher for Academic and Scientific Resources)
www.saspublishers.com
DOI: 10.36347/sjmcr.2015.v03i01.009

Coexistence of Ankylosing spondyloarthritis (AS) and Familial Mediteraneal Fever (FMF) in First Degree Relatives of an Iranian family

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Abstract: Review of literature shows increased frequency of AS among FMF patients. We present the first Iranian family with coexistence of AS and FMF in the first degree relatives.
Keywords: FMF, AS, First degree, Association.

INTRODUCTION

FMF is a genetic disorder caused by mutations in MEFV gene encoding pyrin. Recurrent self-limited polyserositis is the most common presentation. Pattern of articular involvement in FMF may be similar to spondyloarthritis. Literature review shows increased frequency rate of AS among FMF families [1-4].

CASE REPORT

A 26 year of old single man was admitted because of microytic hypochromic anemia. He was a known case of spondyloarthropathy from 15 years ago and had psychological problems from childhood without a precise diagnosis. He had been admitted to a psychiatric disease ward two months ago because of agitation, auditory and visual hallucinations and suicidal thoughts. At presentation he complained of mouth dryness, sore throat and tinnitus in his right ear. His sister was a known case of FMF, homozygote for M694V mutation and was under treatment with colchicine. He had not any history of smoking, alcohol consumption and drug abuse.

He consumed methotrexate, indomethacin, prednisolon and sulfasalazine due to spondyloarthropathy, and used amitriptiline, chlorpromazine, haloperidol, biperidin and omeprazole since 2 months ago.

Physical examination revealed: blood pressure of 120/70 mmHg, pulse rate of 80/min, respiratory rate of 16/min, and body temperature of 37ºC, pale conjunctiva and oral candidiasis. Perforated right tymphanic membrane were noticed in authoscopy. Auscultation of heart and lungs were normal. He had kyphosis in the thoracic and lumbar vertebrae. Schober and occiput to wall distance tests were positive. Neurologic examination was normal except for personality changes and mental retardation in mental status exam.

Laboratory tests are included: Hgb: 7.5 gr/dl; MCV: 56 fl, WBC: 5000/µl; Plt: 442000/µl. Serum Iron and ferritin levels were low. TIBC, liver function tests, renal function tests, and thyroid function tests were normal. B12 and folate levels in serum were normal; stool exam was negative for occult blood. ANA, HBS Ag and HCV Ab were negative. Anti Endomesial Ab (IgA) was in normal range.

Abdominopelvic sonography revealed splenomegaly (151 mm). Color Doppler sonography of portal vein was normal. Audiometry was done and showed mild conductive defect in his right ear. Echocardiography revealed normal findings. Endoscopy showed Antral erythema and Colonoscopy revealed few aphtus lesions in sigmoid colon and an aphtus lesion in ileum. Biopsy of the lesions revealed dense infiltration of PMNs and plasma cells in lamina propria with marked cryptitis and gland distortion compatible with inflammatory bowel disease. On further evaluation genetic test for FMF was performed and like his sister, he was homozygote for MEFV gene while negative for HLA B27 antigen. His sister was tested for HLA B27 antigen and the result was negative as well.

DISCUSSION

FMF is an autosomal recessively inherited disorder that is most prevalent among the hereditary autoinflammatory syndromes. It occurs primarily in

people originating from the Mediterranean basin, including Armenians, Sephardic Jews, Arabs, and Turks [5].

Ankylosing spondylitis (AS) is a chronic inflammatory disease of unknown cause associated with human leukocyte antigen (HLA)-B27. It usually affects the sacroiliac joints at early stages and may involve the axial skeleton at later stages of the disease. Peripheral joint involvement may also be an important feature [6]. AS occurs in all parts of the world, but there are race-related differences in prevalence [7].

FMF arthritis is most frequent among individuals homozygous for the M694V mutation. Chronic sacroiliitis can occur in FMF irrespective of the HLA-B27 antigen, which is the characteristic feature of seronegative spondyloarthropathy (SNSA). A clinical association between FMF and AS has been reported previously. The association of FMF and AS is frequently seen with negative HLA-B27 antigen. However, occurrence of FMF and AS in first relatives has been described by a case report in which both inherited MEFV gene and HLA-B27 persisted [8, 9].

Simultaneous occurrence of FMF and AS in one individual or in first-degree relatives appears to be due to several circumstances. First is the same genetic distribution of FMF and AS in specific regions including Turkey, Saudi Arabia, Iran, Armenia and Italy. Second, there is the phenomena of linkage-disequilibrium that occurs between the responsible genes of both diseases. IL1- gene is one of the responsible genes. Finally, there are similarities in cytokine pathways of FMF and AS.

In our case the patient was a known case of ankylosing spondylitis from childhood. He was homozygote for MEFV gene while negative for HLA B27 antigen. According to aphtous lesions in colonoscopy, biopsy result implying inflammation in the mentioned lesions, abdominal pain, iron deficiency anemia and no response to colchicine ,it seems that the patients is a case of AS with inflammatory involvement of bowels and the simultaneous carrier of FMF gene as well. The patient's sister was a case of FMF homozygote for MEFV gene that was treated with colchicine. She had unilateral sacroilitis and was negative for HLA B27 antigen. It is presumed that unilateral sacroilitis in this case is due to articular features of FMF disease.

In this case report the simultaneous incidence of FMF and AS in one family seems to bethe result of incidental occurrence of two prevalent diseases in one region.

REFERENCES

1. Akkoc N, Gul A; Familial Mediterranean fever and seronegative arthritis. Curr Rheumatol Rep., 2011; 13(5): 388-394.
2. Langevitz P, Livneh A, Zemer D, Shemer J, Pras M; Seronegativespondyloarthropathy in familial Mediterranean fever. Semin Arthritis Rheum., 1997; 27(2): 67-72.
3. Akkoc N, Sari I, Akar S, Binicier O, Thomas MG, Weale ME *et al*.; Increased prevalence of M694V in patients with ankylosing spondylitis; Additional evidence for a link with familial Mediteranean Fever. Arthritis Rheum., 2010; 62(10): 3059-3063
4. Borman P, Gökoğlu F, Taşbaş O, Yilmaz M, Yorgancioğlu ZR; Familial Mediterranean fever-related spondyloarthropathy. Singapore Med J., 2009; 50(3): e116-119.
5. Yuval Y, Hemo-Zisser M, Zemer D, Sohar E, Pras M; Dominant inheritance in two families with familial Mediterranean fever (FMF). Am J Med Genet., 1995; 57(3): 455-457.
6. Dougados M, van der Linden S, Juhlin R, Huitfeldt B, Amor B, Calin A *et al*.; The European Spondylarthropathy Study Group: Preliminary criteria for the classification of spondylarthropathy. Arthritis Rheum., 1991; 34(10):1218-1227.
7. D'Amato M, Fiorillo MT, Carcassi C, Mathieu A, Zuccarelli A, Bitti PP *et al*.; Relevance of residue 116 of HLA-B27 in determining susceptibility to ankylosing spondylitis. Eur J Immunol., 1995; 25(11): 3199-3201.
8. Arthritis & Rheumatism, Vol. 62, No. 11, November 2010, pp 3232–3236DOI 10.1002/art.27683
9. Cosan F, Ustek D, Oku B, Duymaz-Tozkir J, Cakiris A, Abaci N *et al*.; Association of familial mediterranean fever–related MEFV variations with ankylosing spondylitis. Arthritis Rheum., 2010; 62(11): 3232-3236.