

Poster presentation

MEFV mutations in systemic JIA

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Background

Systemic form of juvenile idiopathic arthritis (JIA) is regarded as an autoinflammatory disease. Certain genetic polymorphisms in genes coding inflammatory proteins have been associated with the disease. On the other hand mutations of the MEFV gene cause a monogenic autoinflammatory disease, Familial Mediterranean Fever (FMF).

In a previous study in adult rheumatoid arthritis 3 out of the 25 British patients who developed secondary amyloidosis had a mutation/polymorphism in the MEFV gene.

Aim

To analyse whether mutations in the MEFV gene had an association with systemic JIA.

Patients and methods

MEFV mutations were screened in a total of 32 systemic JIA patients. All had been classified as systemic JIA according to the Durban JIA criteria. None had disease characteristics that met the Tel Hashomer criteria for the diagnosis of FMF.

Results

2 carrier for M694V and two patients who were homozygote for MEFV mutations. Both of these patients were among the most severe patients in the group. One had an excellent response to etanercept whereas the other was resistant to anti-TNF and other conventional treatments and had only a partial response to thalidomide. Although the number of severe mutations were increased in this

small group of patients with systemic JIA the difference with the Turkish population did not reach statistical significance, but the disease causing mutation (M694V) was significantly high in the patients with systemic JIA ($p = 0.02$).

Conclusion

However, the severe disease course in the aforementioned patients suggest that MEFV mutations may be a modifying genetic factor in systemic JIA.