Pediatric Rheumatology

Poster presentation

A strange fever in two brothers

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from 15th Paediatric Rheumatology European Society (PreS) Congress
London, UK. 14–17 September 2008

Published: 15 September 2008

Pediatric Rheumatology 2008, **6**(Suppl 1):P202 doi:10.1186/1546-0096-6-S1-P202

This abstract is available from: http://www.ped-rheum.com/content/6/S1/P202

Background

The Hyper Immunoglobulin D and period fever syndrome (HIGD), one of hereditary auto-inflammatory syndromes, is caused by mutations in the gene coding for Mevalonate kinase. This autosomal recessive disease is never reported in siblings.

Case report

A 5 year-old boy was admitted to the hospital for limping and periodic fever. His family medical history was remarkable for the same symptoms in his brother (3 years old). Their medical history started at the age of three months in both cases, after the first vaccination, with monthly periodic fever associated with laterocervical and submandibular lymphadenopathy, severe abdominal pains with vomit and diarrhea and maculo-papular rash. The duration of fever was 3–6 days (40°C).

On physical examination

laterocervical lymphadenopathy, maculo-papular rash and arthritis with swollen on right knee.

Blood tests

neutrophil leukocytosis (WC 15000 mmc, N 82%). ESR, CRP, IgA and Serum Amyloid A levels were elevated. Additional investigations included IgD and autoantibody pattern and were unremarkable.

Based on clinical history and symptoms of hereditary autoinflammatory syndromes, we looked for the mutations of mevalonate kinase (MKD) in both siblings.

Conclusion

We found a deletion, on chromosome 12: compound heterozygous for *I268T* e *V377I* mutations. Thus a diagnosis of "Hyper IgD Syndrome (HIGD)" was made.

We controlled the disease in both with colchicine (1 mg/day). The peculiarity of our case report is the presence of a rarely periodic fever in two brothers, never reported.

References

1. D'Osualdo A, *et al.*: **MVK mutations and associated clinical features in Italian patients affected with autoinflammatory disorders and recurrent fever.** *European Journal of Human Genetics* 2005, **13(3):**314-20.