Pediatric Rheumatology

Poster presentation

Mevalonate kinase deficiency (MKD): long-term follow-up of clinical and biological features in 40 patients

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MKD causes hyper-IgD syndrome and mevalonic aciduria. Disease severity and clinical phenotype has not yet been linked to specific mutations. In this study, we intended to review clinical presentation and outcome of pediatric and adult patients.

40 patients were identified through MVK mutation database in one central reference laboratory and physicians' networking.

Medical charts were reviewed (21/40) or questionnaires were filled in by treating physician (19/40).

Follow-up ranged from 2 years to 60 years. First symptoms occurred before age 2 in 90% of patients. Main clinical features were fever (100%), abdominal pain (92%), diarrhea (88%), enlarged lymph nodes (88%), skin rash (86%), arthralgias (85%), arthritis (43%), growth delay (33%), CNS white matter involvement (7,5%) and macrophage activation syndrome (5%). We recorded 3 disease-related deaths. IgD level ranged from normal (<100 IU/ml) to 2614 IU/ml and were persistently normal in 6 patients. IgA level was above upper limit in all but 2 patients. Mevalonate kinase activity was deeply impaired (0,7–7% of normal controls). The most prevalent mutation was V377I. In 3 independent kindreds, only one mutated allele was found in the coding sequence of the gene. Most patients received NSAIDs, steroids and colchicin. Etanercept and anakinra at high doses showed some efficacy. Some patients were refractory to all treatments. One patient succesfully received allogenic stem cell transplantation.

MKD is a challenging condition, with a wide array of severity, potentially severe complications and a high burden on quality of life.