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PReS-FINAL-2241: Cases of cryopyrin-associated periodic syndromes (CAPS) in one single rheumatologic center of Russia

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Introduction

Cryopyrin-associated periodic syndromes (CAPS) are the rare hereditary autoinflammatory diseases. CAPS include three similar conditions are distinguished which lie along a phenotypical continuum with increasing levels of severity: familial cold autoinflammatory syndrome (FCAS), Muckle-Wells syndrome (MWS) and CINCA/NOMID. Distinguishing features include cutaneous, neurological, ophthalmologic and rheumatologic manifestations. CAPS caused by the mutation of the NLRP3 (CIAS1) gene coding for cryopyrin.

Objectives

to investigate clinical and genetic characteristics of the pts with CAPS in single Federal rheumatologic center of Russia.

Methods

7 patients with CAPS were followed at our centre for 5 years since 2007. There are 3 boys, 4 girls, aged 2-16 years (mean 7,9 ± 5,2 years) with disease's duration from 2 to 15 years. In 2 boys was diagnosed CINCA/NOMID, 5 - MWS. All patients were submitted to routine rheumatology examination and laboratory parameters, genetic testing in all pts was performed in research centre for medical genetics in Moscow.

Results

Age of the first manifestation was from the birth to 9 years. All pts have had persistant or recurrent episodes fever, cutaneus rash (urticaria-like rash or maculo papular). 5 of 7 pts have had ocular manifestations: 3- conjunctivitis, 4-uveitis. 2 - no ocular abnormalities. Sensoryneural hearing loss was diagnosed in 2 boys with CINCA/NOMID and in 2 girls with MWS. Joint involvement were observed in 6 of 7 (artralgias/non-destructive arthritis - 5, bone deformities - 1 and growth retardation in 2 boys with CINCA/NOMID). Central nervous system damage (hydrocephaly, atrophy on brain, mental retardation) was in 3 pts (2 - CINCA/NOMID, 1- MWS). The laboratory findings included an elevation of acute phase reactants: ESR, neutrophil leukocytosis, C-reactive proteine rise, anemia. ANA and RF were negative. A genetic analysis on all pts showed a new mutations (heterozygous) in the NLRP3 gene: G569R, Met406Thr, Thr436Ile, Y441H, P350L, Thr438Ile, Gly455Term. No pts with AA amyloidosis and renal failure.

Conclusion

CAPS (CINCA/NOMID and MWS) are rare hereditary auto-inflammatory diseases but it can occur in Russia. CINCA/NOMID pts have the most serious clinical manifestations and prognosis. MWS pts more often have recurrent episodes fever, cutaneous rash, conjunctivitis. Pts with CAPS need early anti IL-1 treatment for the successful in suppressing inflammation, reduction in the number and duration attacks, for prevention of amyloidosis and improvement of life prognosis.

Disclosure of interest

None declared.

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