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Diversity in clinical manifestations of autoinflammatory syndromes

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Background

The autoinflammatory syndromes (AIS) include monogenic and polygenic disorders characterized by primary dysfunction of the innate immune system.

Aim

To describe the spectrum of AIS in a Pediatric Rheumatology reference center.

Methods

Medical records of AIS patients followed until November 2010 were studied.

Results

Fifty six patients were included: 17 CAPS, 4 TRAPS, 5 HIDS, 18 FMF, 6 CRMO, 2 SAPHO and 4 Behcet. The median follow-up period was 2 years (0-14 years). The male/female ratio was 20/36. The median age was 2.5 years at disease onset and 4 years at diagnosis. Family history was positive in 34% of patients. Clinical manifestations included fever (79%), mucocutaneous (61%), musculoskeletal (77%), ocular (34%), cardiorespiratory (12%), gastrointestinal (62%), neurological (41%) and genitourinary (2%) findings, lymphadenopathy with/or hepatosplenomegaly (16%) and growth impairment (27%). Seven patients presented severe manifestations: neonatal peritonitis (1 CAPS), pancreatitis (1 TRAPS), acute glomerulonephritis (1 FMF), complicated Henoch-Schönlein purpura (1 FMF), peritoneal adhesions with intestinal occlusion (1 FMF), periorbital pain (1 CRMO), and cerebral thrombosis (1 Behcet). One mutation was found in 93% of CAPS, and in all TRAPS patients. Two mutations were present in 11% of FMF, and in all HIDS patients. 43% of patients received colchicine, 23% steroids, and 54% biologics (Anakinra, Canakinumab, Etanercept). 57% of patients were in complete and 41% in partial remission.

Conclusion

AIS in children are associated with a broad spectrum of symptoms. A detailed history, thorough review of clinical symptoms and molecular testing of specific causative genes can provide early and accurate diagnosis.

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