PEDIATRIC RHEUMATOLOGY

MEETING ABSTRACT Open Access

P03-018 - Diversity in presenting manifestations of AUTOINFL

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Introduction

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

Objectives

To describe the clinical spectrum, genetic background and therapy in a cohort of AID patients followed in a reference Pediatric Rheumatology center.

Methods

Medical records of AID patients followed between May 2007 and November 2010 and entered in the Eurofever Registry were studied.

Results

Fifty six patients were included: 17 Cryopyrin-Associated Periodic Syndromes (CAPS), 4 TNF-Receptor-Associated Periodic fever Syndrome (TRAPS), 5 Hyperimmunoglobulinaemia D with periodic fever Syndrome (HIDS), 18 Familial Mediterranean Fever (FMF), 6 Chronic Recurrent Multifocal Osteomyelitis (CRMO), 2 Synovitis, Acne, Pustulosis, Hyperostosis, Osteitis (SAPHO) syndrome and 4 Behçet's Disease (BD). The median follow-up was 2 years (0-14). The male/female ratio was 20/36. The median age was 2.5 years at disease onset and 4 at diagnosis. Family history was positive in 34% of patients. Clinical manifestations included fever (79%), musculoskeletal (77%), gastrointestinal (63%), mucocutaneous (61%), neurological (41%), ocular (34%), cardiorespiratory (13%), and genitourinary (2%) findings, lymphadenopathy with/or hepatosplenomegaly (16%) and growth impairment (25%). Complications/sequelae developed in 45% of patients. Six patients presented with unusual 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 BD). AID was associated with other diseases in 2 patients (FMF/Henoch-Schönlein purpura and CRMO/enthesitis-related arthritis). One mutant allele was found in 16/17 CAPS, 4/4 TRAPS and 4/18 FMF patients. Two mutant alleles were present in 5/5 HIDS and 11/18 FMF patients. The most used therapeutic agents were biologics (54%) (Anakinra, Canakinumab, Etanercept, Adalimumab), NSAIDs (48%), colchicine (45%) and corticosteroids (29%). Anti-interleukin-1 therapy and colchicine proved efficacy in CAPS and FMF patients, respectively. In addition, favorable responses demonstrated anti-interleukin-1 therapy in TRAPS, HIDS and colchicine-resistant FMF patients, as well as Etanercept in TRAPS, HIDS and CRMO patients non-responsive to NSAIDs. 57% and 41% of patients were in complete and partial remission, respectively, at last visit.

Conclusion

AID in children are associated with a broad spectrum of manifestations. Early diagnosis and referral are essential as efficient therapy can be proposed in most cases.

Competing interests
None Declared.

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