ABSTRACT BOOK ABSTRACTS



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GENETICS AND GENODERMATOSES

PAPA SYNDROME: STERILE PYOGENIC ARTHRITIS, PYODERMA GANGRENOSUM AND ACNE

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Man of 27 years admitted in the Dermatology Department of the General Hospital of Mexico in January 2017. He presented 4 vegetative ulcers, 2 in the abdomen and two in thigh and right leg, all ulcers with irregular shape, defined, undercut and violaceous borders, of 8 x 5 and 2 x 1.5 cm with hemato-purulent exudate. He also had inflammatory acne. In 2007 he suffered "joint inflammation" without providing more data and that since 2015 he is dependent on crutches due to joint pain. Our clinical suspicion was PAPA syndrome. Skin biopsy reported compatible with pyoderma gangrenosum. Pelvic radiography showed IV degenerative osteoarthritis. We started prednisona 1 mg / kg / day with total improvement in 4 months. Analysis of the coding sequence of exon 10 of the PSTPIP1 gene using PCR amplification and Sanger type nucleotide sequencing showed the homozygous c.688G >A. p.Ala230Thr variant, previously reported as pathogenic of the PAPA syndrome with dominant heritage. The immunophenotype reported a decrease in absolute numbers and percentage of helper and NK lymphocytes and the functionality of NK cells was reduced. This syndrome is a rare autosomal dominant disorder caused by mutations in the PSTPIP1/CD2BP1 gene on 15q chromosome. It is an autoinflammatory syndrome characterized by systemic inflammation in the absence of typical autoimmuity features. It combines the triad of aseptic pyogenic arthritis, pyoderma gangrenosum and acne. Mutations in PSTPIP1 results in abnormal function of the inflammasome complex with increased signaling through the proinflammatory cytosine IL-1B. The first line treatment options are systemic corticosteroids. Targeted therapies include blockade of the IL-1 receptor by anakinra. Canakimumab is a monoclonal antibody against IL-1ß with sustained clinical response. Anti TNF agents have shown promising results, mainly in cutaneous manifestations. For the treatment of acne the use of topical and oral retinoids is recommended.





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