P024

a pediatric case of familial chilblain lupus with R152H homozygous mutation in TREX-1 gene

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Introduction: Familial chilblain lupus (FCL) is a rare form of monogenic systemic SLE that typically develops in early childhood with chilblain-like skin involvement. It presents with autosomal dominant inheritance (1). Typical clinical findings are painful and ulcerated erythematous plaques that occur after cold contact on the acral faces (2).
Objectives: Here, we present a case of FCL who was followed with the diagnosis of lupus pernio since infancy and diagnosed with FCL after show up homozygous mutation in the TREX1 gene.
Methods: Written consent form was taken from our patient and his family.
Results: A 13 year-old male patient presented with the complaint of painful wounds on his ears, cheeks and toes. It was reported that similar complaints developed in the ears and cheeks, since nine-year-old. There was no additional rheumatologic complaint in the history. Skin biopsy of erythematous lesions on the soles of the feet performed in infancy was found consistent with lupus pernio. Physical examination of the patient shown painful and in places ulcerated erythematous plaque lesions on the both ears helix, on both cheeks and dorsal faces of bilateral toes. All laboratory parameters were found in normal range. Brain MRI and echocardiography were performed and found normal. FCL was considered as a preliminary diagnosis in the patient with chilblain-like lesions onset at an early age and triggered by cold. Genetic panel analysis was performed. Homozygous R152H mutation was shown in TREX-1 gene. The patient was diagnosed as FCL due to the presence of just chilblain lesions. Hydroxychloroquine, prednisolone and tofacitinib treatment were started, respectively. All chilblain lesions healed, leaving hyperpigmentation. During the follow-up, prednisolone was tapered and discontinued. The 23-year-old sister of the index patient, who had arthritis and chilblain lesions, had also the same TREX-1 mutation and was diagnosed with FCL.
Conclusion: Familial chilblain lupus should consider in differential diagnosis in patients with chilblain lesions beginning at early age and with a similar family history.

References

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Trial registration identifying number: According to the legal status in our country, ethics committee approval is not required for case reports. It is adequate to obtain informed patient consent.

Disclosure of Interest

None declared

P025

From smart working to smart co-working in the covid-19 era: a pilot program of cooperation around autoinflammatory diseases

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Introduction: The last time was signed by the pandemic diffusion of COVID-19, with an emergency area COVID-19 dedicated and the need to minimize the inflow of children and adolescents affected by chronic diseases into the hospitals. Otherwise, paediatricians had to limit visits and to consider a new setting for febrile children.
Objectives: Patients affected by autoinflammatory diseases were assisted by telephonic consultations guaranteed by the paediatricians of free choice and by the paediatric rheumatologists.
However, the patients frequently needed a direct clinical approach and a specialistic evaluation in the case of flares and/or abnormal laboratory parameters and adverse reactions to drugs.
Another frequent question was the differential diagnosis of febrile episodes, to distinguish a recurrent fever, linked to autoinflammation, from an infectious disease.
Methods: we proposed to paediatricians of free choice in west-Sicily a questionnaire about difficulties met in the follow-up of children with autoinflammatory syndromes; needs of scientific or bibliographic support, number of patients with these diseases and treated with biological drugs.
Results: 55 questionnaires were collected: the most frequent recorded conditions were PFAPA and Familial Mediterranean Fever (FMF); a lower percentage followed CAPS; MVK, TRAPS.
The most frequent treatment in PFAPA was steroids on demand; in FMF was colchicine. A low percentage (10%) was treated with anti-IL-1 drugs, needing the access to the hospital to receive the therapy.
All the paediatricians needed specialistic support to adequately control flares, especially in FMF, CAPS, TRAPS and MVK. PFAPA patients were almost individually controlled by paediatricians.
Conclusion: Patients and paediatricians needed a specialistic help to organize the follow-up of these patients and to guarantee a good compliance to treatment.
This period characterized by smart working, telemedicine, strategies to monitor remotely the patients, can find the winning strategy in the approach of the "Co-working", a new cooperation between hospital and paediatricians of free choice, in the global follow-up of autoinflammatory diseases.

Disclosure of Interest

None declared

P026

Juvenile Idiopathic Arthritis with NOD2/CARD5 gene mutation or blau syndrome with arthritis and uveitis: lessons from familial case report

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Introduction: Blau syndrome (BS) is systemic autoinflammatory disease characterized by an early onset granulomatous arthritis, uveitis and skin rash, caused by a mutation in the NOD2/CARD5 gene. In real practice an extremely rare monogenic disease like BS is difficult to recognize and it's initially diagnosed as Juvenile idiopathic arthritis (JIA) due to phenotypically similar symptoms.
Objectives: To analyze the diagnosis pathway and results of biologics therapy in a family case of two siblings with BS.
Methods: Case report of 2 brothers with BS genetically confirmed by NOD2/CARD5 gene mutation.
Results: Two brothers of 15 and 3 years old were examined in our clinic. The elder brother presented arthritis of both wrist joints at the age of 2 years. The appearance of a scaly erythematous maculopapular rash on the trunk and extremities preceded the development of onset of arthritis. 3 years later he developed polyarthritis involved knees, ankles and three PIF joints of the left hand. There was no significant improvement after treatment with NSAID, methotrexate (MTX) and cyclosporine A in regional hospital, so etanercept was added since 2012 with variable result. Despite of