instructed to take vitamin D supplementation (600 UI/day). The parents of the children were then provided with a standardized diary to register the clinical course of PFAPA and were asked to come back 6 months later for evaluation

Results: At baseline 13/18 PFAPA children had a vitamin D deficiency. In two children the parents refused to test the vitamin D levels. Quantitative ultrasound showed a reduced bone mineral density in 10 children, with 6 children showing ADSOS and BTT z-score lower than 1SD and 4 children showing values lower than 2 SD. 3 patients had reduced bone mineral density with normal levels of vitamin D.

All the patients with vitamin D deficiency were put on vitamin D, no matter of the results of bone mineral density evaluation. 8/20 patients completed the 6 months f/up, 8 are still in the follow/up period, 4 were lost at follow/up for clinical remission after tonsillectomy (3 were on vitamin D supplementation). Of the 8 patients completing the 6 months f/up, 5 patients had vitamin D deficiency at baseline and 3 patients had normal vitamin D levels. The 5 children who received supplementation had normalization of the 25-hydroxyvitamin D serum level. Sonographic parameters reached normal values only in one patient, while one patient with normal bone mineal density at baseline showed worsening of the sonographic parameters. In 2 children on vitamin D supplementation we noticed the remission of disease, in 2 children there was a reduction in the frequency of PFAPA episodes, 2 didn't show any significant clinical modification. The three patients without vitamin D deficiency at baseline, showed normal levels of vitamin D at follow/up and no changes in disease characteristics. There was no apparent correlation between bone mineral density parameters and disease outcome.

Conclusion: Although very preliminary and limited to a small number of patients, our data seem to confirm a role for vitamin D deficiency on the occurrence of PFAPA syndrome. Vitamin D supplementation may be a possibility in children with PFAPA, at least in those with concurrent vitamin D deficiency. Larger studies are needed to further confirm this role and to better understand a possible role of bone mineral density determination on such children.

Disclosure of Interest

None Declared.

P247

Ethosuximide-induced lupus-like syndrome: a case report

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Introduction: Drug-induced lupus erythematosus is an autoimmune disorder caused by a drug overreaction. To our knowledge only one case of possible correlation between the use of ethosuximide and SLE has been described so far

Objectives: To describe a case of drug induced lupus and the use of interferon signature as a marker of disease activity in clinical practice

Methods: we describe a case of drug induce lupus followed clinically, with routine immunological work-up and with serial determinations of interferon signature, by means of quantitative reverse transcription PCR, measuring 6 different interferon I stimulated genes (IFI27, IFI44L, IFIT1, ISG15,RSAD2,SIGLEC1), as previously described[1]

Results: M.L. is a 16-year-old girl who was suffering since birth from a very mild cognitive delay, focal epilepsy and ESES (continuous spike wave epilepsy during sleep) since 9 y/o. Extensive neurological work-up failed to revealed any underlying disease. The girl was put on Ethosuximide with good seizures control. Two years later she started to complain from generalized arthralgia and myalgia, transient hands and feet swelling, and recurrent, evanescent rash on cheeks. The girl was then referred to our rheumatology unit. Physical exam was normal but lab-works showed ANA 1:640 homogeneous, positive anti ds-DNA antibodies, reduced complement levels and mild proteinuria. We also run an in-house interferon signature assay, revealing a very high expression of IFN-stimulated genes, similarly to children with clear-cut SLE. For this reason a drug-induced lupus was hypothesized and the collegial decision was to stop ethosuximide. In the following 6 months the girl remained free of seizures, the myalgia and rash disappeared, all the laboratory abnormalities normalized. We also found a decrease in the interferon signature, testifying a lower expression level of the 6 interferon-stimulated genes analyzed

Conclusion: We herein present a case of what we believed was a drug-induced lupus. Interferon signature may be very useful, together with classic immunological work-up, to follow the disease course in these patients.

[1]Rice GI et al. "Assessment of interferon-related biomarkers in Aicardi-Goutières syndrome associated with mutations in TREX1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, and ADAR: a case-control study", Lancet Neurol. 2013 Dec;12(12):1159-69

Disclosure of Interest

None Declared.

P248

Platelet count and MPV as predictive markers of atherosclerosis in familial Mediterranean fever

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Introduction: Familial Mediterranean Fever (FMF) is an auto inflammatory syndrome, characterized by recurrent febrile episodes, arthritis, oral aphthous stomatitis, rash, serositis, abdominal and thoracic pain. Long-term outcome is conventionally linked to the severity of the recurrent attacks and to the risk of systemic amyloidosis. However recent studies highlighted the role of chronic inflammatory diseases in the insurance of atherosclerosis. Risk factors for atherosclerosis are also recently identified in a higher medium platelet volume (MPV).

Objectives: We evaluated platelet parameters (MPV; platelet distribution width: PDW) in children affected by FMF and considered them as indexes or atherosclerotic risk. We evaluated the efficacy of colchicine treatment on clinical and biochemical parameters.

Methods: We enrolled 33 children (8,6 ± 4,6 years; 12 F and 21 M), followed since 2012, with the diagnosis of FMF, confirmed by genetic study. We evaluated platelet parameters (count, MPV, PDW), CRP, ESR, leukocyte count, neutrophils percentage, serum amyloid-A (SAA) during the acute phase, during the remission and far from febrile events., All the patients, with the exception of 5, receive colchicine treatment. Biochemical parameters were matched with those of 23 healthy controls, matched for sex and age.

Results: Platelet count was significantly higher in acute phase (379,800 ± 111,600) than in free-disease periods (328,400 ± 88,200) and in controls (314,200 ± 72,800). MPV was lower in acute phase (8,18 ± 0,85) than in free-disease periods (8,22 ± 0,67); these parameter was significantly lower than in the control group (8,5 ± 0,82). PDV was lower in acute phase (37,97 ± 15,45) than in free-disease periods (45,1 ± 9,2) as in control group (45,96 ± 6,07). In acute phase, MPV was directly correlated with CRP, ESR, SAA; showed a statistically significant correlation with MCV ($p < 0.05$). During the free-disease periods, MPV was directly correlated with: CRP, ESR; inversely with SAA and MCV ($p < 0.05$).

SAA was maintained in the normal range in free-disease periods in all the patients, with the exception of 5 who had an incomplete response to colchicine.
Conclusion: Long-term complications of FMF include kidney amyloidosis and atherosclerosis, however we do not have a valid predictive marker of thrombosis in these children. An increase in MPV and PDW, expression of platelet activation, can predict an increased risk of thrombosis. In our children these parameters are adequately controlled by colchicine, preventing the endothelial dysfunction.
Disclosure of Interest
None Declared.

P249

PAPA syndrome with latent tuberculosis successfully treated with canakinumab

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Introduction: Pyogenic sterile arthritis pyoderma gangrenosum and acne (PAPA) syndrome is a rare autosomal dominant autoinflammatory disease caused by mutations in PSTPIP1 gene.
Steroids, anti-TNFa and anti-IL-1 agents have been proposed as treatment options, with variability in response being observed.
There is evidence that IL-1B secretion in PAPA is increased and correlates with disease activity.
Objectives: To present the case of a child with PAPA syndrome and latent tuberculosis successfully treated with canakinumab.
Methods: An 8-year-old male arrived to our Pediatric Rheumatology Unit diagnosed with juvenile idiopathic arhritis (JIA). The patient presented recurrent arthritis of his knees from the age of 3 years. The first episode was treated as septic arthritis because of elevated acute phase reactants and purulent synovial fluid. Cultures were negative. Subsequently he was treated with intraarticular steroid injections, systemic steroids and methotrexate, persisting activity of the disease. At the age of 4 years the patient developed ulcerated cutaneous lesions in right underarm region and groin that lasted for several months. He also had history of frequent intense local vaccine reactions. His mother was diagnosed with a systemic JIA and his grandmother with a rheumatoid arthritis. Father with psoriasis.
When the patient was assessed in our unit, he presented arthritis of his left knee. The synovial fluid was purulent with 131090 leucocytes (94% neutrophils). The blood tests showed elevated acute phase reactants. The Mantoux test was positive with 17 mm of induration. The possibility of a pathergy phenomenon was thought, but IGRA tests were also positive. Chest X-ray and high resolution thoracic tomography did not show pathological findings. Synovial fluid cultures were negative, including mycobacteria, and PCR of *Mycobacterium tuberculosis* was not detected. In the gastric fluid there was no presence of acid-fast bacilli and cultures were negative. He was treated with isoniazid because of latent tuberculosis infection.
Results: The study of mutations in the PSTPIP1 gene revealed the mutation E250Q in heterozygosity, that was also present in the mother and in the grandmother.
An intraarticular steroid injection was performed in the knee and treatment with canakinumab 2 mg/Kg/4 weeks was started after 4 weeks of treatment with isoniazid, with good response.
The patient has received treatment with canakinumab now for 34 months with good control of the articular disease. One year after treatment onset he had eczematous recurrent lesions in antecubital flexure at site of puncture for blood test, that were thought to be a clorhexidine contact dermatitis. No other cutaneous lesions apart from dry scaly lesions in the scalp and eczematous psoriasiform lesions in the pinna have appeared during the follow-up. Normal blood tests with no elevation of inflammatory parameters. Good physical growth. The treatment has been well tolerated without significant infections. The patient is currently on canakinumab 2 mg/Kg/8 weeks.
Conclusion: We present a family with PAPA syndrome with one member treated with canakinumab and good control of the disease during 34-month follow-up on treatment. Long-term efficacy and control of cutaneous disease, that is described to appear later on the course of the disease, will be established during follow-up.
A particular feature in this case is the difficulty related to the latent tuberculosis infection because of the possible initial interpretation of Mantoux test as pathergy phenomenon and the treatment with isoniazide before starting biological treatment.
Disclosure of Interest
None Declared.

P250

Successful use to tocilizumab to treat pigmentary hypertrichosis and non-autoimmune insulin-dependent diabetes mellitus (PHID) syndrome

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Introduction: Autoinflammation is increasingly recognised as a feature of Pigmentary hypertrichosis and non-autoimmune insulin-dependent diabetes mellitus (PHID) syndrome. We previously reported a 16-year-old girl of Pakistani origin with PHID syndrome, associated with severe autoinflammation that was recalcitrant to treatment with blockade of interleukin-1 (IL-1) and tumor necrosis-α (anti TNF)[1]. Herein we report the therapeutic response to IL-6 blockade with Tocilizumab.
Objectives: To report the first case of successful treatment of PHID syndrome using IL-6 blockade with Tocilizumab.
Methods: Retrospective medical records review of a single case of PHID, caused by *SLC29A3* mutation, documenting clinical and serological response to Tocilizumab over 23 months. We noted improvement in clinical features such as change in sclerotic skin, reported levels of fatigue, and acute phase response (C-reactive protein [CRP], erythrocyte sedimentation rate [ESR] and serum Amyloid A [SAA]).
Results: We have previously published on the lack of response to Anakinra, Adalimumab and Methotrexate[1]. Immediately prior to commencing Tocilizumab, there was evidence of significant systemic inflammation: SAA 178 mg/L; CRP 54 mg/L; and ESR 86 mm/hr. We therefore decided to trial treatment with intravenous Tocilizumab 8 mg/kg, every 2 weeks. Within 12 weeks of treatment her inflammatory markers drastically improved: CRP <5 mg/L, ESR 22 mm/hr and SAA 8.4 mg/L. Her dose was increased to 12 mg/kg after three initial doses, due to ongoing fatigue and sclerotic skin changes. She subsequently reported marked clinical improvement in her energy levels, improved appetite, reduced fevers, less skin tightness and night sweats. Methotrexate was discontinued 9 months after commencing Tocilizumab due to continued excellent clinical and serological response, which has been sustained over 23 months. The cutaneous lesions remain significantly improved (decreased sclerosis and hypertrichosis); her weight and height have improved but remain below the 0.4th centile. Tocilizumab had no impact on her diabetes.
Conclusion: PHID syndrome is associated with recessive mutations in *SLC29A3* which encodes for the equilibrative nucleoside transporter hENT3 expressed in mitochondria, causing PHID and H syndromes, familial Rosai-Dorfman disease, and histiocytosis-lymphadenopathy-plus syndrome.
Tocilizumab is a humanised, monoclonal, antihuman IL-6 receptor (IL-6R) antibody that binds to membrane and soluble IL-6R, inhibiting IL-6–mediated signaling. It has been used to successfully treat rheumatoid arthritis, systemic JIA, and polyarticular JIA. There are no published reports of the treatment of PHID syndrome using IL-6 blockade.