heterozygotes, 1 homozygous patient); I591T (3 heterozygotes). Some of them had 2 or more mutations in association. All the patients were treated with colchicine with a complete (95%) or partial response to the treatment. None of the patients developed amyloidosis. Three patients from unrelated families, had a vasculitis (2 Kawasaki Disease; 1 Schoenlein-Hoenoch Purpura) and all presented the same association: P369S and R408Q.
Conclusion: This finding verifies the importance of molecular diagnosis and detailed sequencing which is recommended to perform in particular for the countries with a high risk of FMF.
In several instances, family studies provided the prevalence of a single mutation in patients experiencing a pathogenic effect, with molecular evidence for pseudodominant transmission. We evidenced a variable clinical and serological pattern between patients in the same family; the genetic study was in fact extended to parents and brothers of the index case, with the recommendation to dose Serum amyloid A (SAA), blood pressure and evaluate an urine analysis to exclude proteinuria.
The M694V homozygous and heterozygous genotype was found to be associated with a higher prevalence of amyloidosis and arthritis and higher levels of SAA. The parents of our patients with M694V mutation have more severe clinical manifestations and a lower response to colchicine.
Further studies are needed to highlight generational differences of clinical spectrum and SAA levels in FMF patients of the same family, carrying the same mutations.
Disclosure of Interest
None Declared.

P188

Anti-IL1 in patients with low penetrance mutations for autoinflammatory diseases: tuscany and sicilian case series from paediatric to adult age

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Introduction: Patients with low penetrance mutations for Autoinflammatory syndromes (AID) can have severe clinical manifestations, which require to be treated with biological drugs anti-IL-1.
Objectives: To evaluate the response of AID to treatment with the recombinant human IL-1 receptor antagonist anakinra or with the anti-IL-1b.
Methods: We enrolled from 3 centers (U.O. of Rheumatology, Universitary Hospital "S. Maria Le Scotte", Siena; S.O.D.C. of Paediatric Rheumatology, "A. Meyer" Hospital, Florence and U.O. of Paediatric Clinic, Children Hospital "G. Di Cristina", Palermo) 26 patients with SAI and low penetrance mutations, with age: 0.8-58 years (11 M, 15 F; age:4-62 years; medium age of paediatric patients: 11.2 years). The symptoms started in paediatric age in all the patients; however adult patients received the diagnosis in adult age.
Results: All the patients (9 CAPS, 10 TRAPS, 2 HIDS, 2 FMF, 3 sJIA) received anti-IL-1b drugs (anakinra or canakinumab). The subjects kept a diary of symptoms at the diagnosis and at the outset, and underwent clinical and laboratory assessments, including measurement of the SAA, ESR, CRP, blood count, urinalysis.
At the outset, the 84.6% showed recurrent episodes of fever, variously associated with: rash (61.5%), abdominal pain (50%), arthralgia and/or myalgia (88%), arthritis (46%). All the patients, before starting anti-IL-1b drugs, were treated with NSAIDs, steroids, DMARDs, colchicine with a poor control of the disease. The 30.7% associate the anti-IL-1b to one or more of other drugs. The 57.7% (38.5% between children, 19.2% between adults) showed a complete remission; the 19.2% incomplete, the 23.1% did not respond. SAA was increased in 88.5% (M: 155,86; n.v. < 6,4 mg/l), reduced in 58%. CRP was increased before anti-IL-1b drug in the 50%, with a normal value in the 92% after the drug was started. ESR was increased in the 69.2%, with a normal value in the 42.3%.
Proteinuria was detected in the 8% before the anti-IL-1b drug was started and was in the normal range after they was treated with the biological drug.
In children, prevalent clinical manifestations were abdominal pain and arthritis; in adults thorax pain and pericarditis were more frequent.
Conclusion: The clinical features of the AID were correlated with age, also in patients with low penetrance mutations: some manifestations were more frequent in adults, others in childhood. The remarkable response on clinical and haematological parameters (76.9% of all the patients) to anakinra or canakinumab suggests that IL-1β has a fundamental role in the pathogenesis of inflammation associated with low penetrance mutations as well. In paediatric age, IL-1 blockade higher efficacy was probably linked to a more severe clinical phenotype.
Disclosure of Interest
None Declared.

P189

Genetic and clinical profile of a sicilian population with R92Q mutation

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Introduction: Gene TNFRSF1A mutation is linked to TRAPS, autosomal dominant Autoinflammatory Disease (AID) with recurrent attacks of fever (2-3 weeks long), abdominal pain, vomiting, serositis, arthralgia and/or arthritis, myalgia, fasciitis, rash. The disease starts precociously and amyloidosis is reported in the 25% of the patients. Patients carrying the mutation R92Q usually show a mild clinical phenotype, with an extreme interindividual variability. Arthralgia and serositis are frequently less severe, however oral ulcers and pharyngitis are recurrent.
Objectives: We studied the clinical and biochemical impact of the mutation R92Q in our population and the treatment outcome in all the patients with clinical relevant symptoms.
Methods: We followed 15 patients (6 M and 9 F), 11 children and 4 adults, carrying the R92Q heterozygous mutation of the gene TNFRSF1A. The diagnosis of children were done at the age of 4-14 years, o the adults was performed following the sons diagnosis. SAA levels were significantly high in 8/11 children and in 2/4 adults.
Results: All the symptomatic patients were treated with NSAIDs, steroids, colchicine with a variable control of the disease. The colchicine was not sufficient in 4/5 patients and 2 of them were treated with the anti-IL-1 β biological drug canakinumab.
Conclusion: Functional studies performed on R92Q evidenced a changed conformational structure vs. the wild type.
Our patients showed polymorphic clinical features: some of them are asymptomatic, other record different symptoms with an intrafamilial variability. In paediatric age, the clinical phenotype is more severe also in correlation with the symptoms of parents carrying the same mutation. We stress the data that all our patients underwent the genetic study because they recorded symptoms linked to AID.
Disclosure of Interest
None Declared.

P190

A paediatric FMF patient with recurrent priapism during attacks

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Introduction: Priapism, a prolonged penile erection lasting >4 hours, is a rare condition in childhood. The commonest causes of priapism in children are sickle cell disease, leukaemia, trauma, idiopathic, and pharmacologically induced. To date, the association of familial Mediterranean fever (FMF) and priapism has not been reported.
Objectives: To report an FMF patient experiencing priapism during attacks.
Methods: To report an FMF patient experiencing priapism during attacks.
Results: A 20-months-old boy was referred to our pediatric rheumatology department with the episodes of fever, monoarthritis, and pustules on the face recurring every month since 10-months of age. Because MEFV analysis revealed M694V homozygote mutation, he was started 0.25 mg/day colchicine by his pediatrician when he was 14-months-old. Despite the adjustment of colchicine to 0.5 mg/day, he continued to have recurrent fever and pustules. In his immunologic evaluation, white blood cell number, immunogobuline G,M,A levels, lymphocyte subgroup disturbation, phagoburst test was normal.He was given a short-course of corticsteroid because of fever and a painful erythema nodosum-like lesion when he was 26-months old. PSTPIP1 mutation was negative for PAPA. When he was 2.5 years-old, he had penile erection during an attack. Colchicine dose was increased to 1 mg/day. He experienced two more attacks of priapism with fever and pustules of the face. He was started an anti-interleukine 1 treatment, anakinra 1 mg/kg/day. He never had priapism, pustules, arthritis and fever attacks after the initiation of anakinra. After one year, anakinra was stopped. He is currently taking only colchicine since one year and doing well.
Conclusion: To the best of our knowledge, this is the first case showing an association of priapism with FMF attacks.
Disclosure of Interest
None Declared.

P191

Severe arthritis as clinical presentation in a a novel case of COPA syndrome

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Introduction: Heterozygous mutations of *COPA* gene have been recently linked to an autoimmune syndrome characterised by polyarthritis and lung disease with interstitial fibrosis or alveolar haemorrhages as complication (1). The gene encodes for a regulator of vesicular retrograde transport between Golgi and the endoplasmic reticulum (ER). In most cases the presenting symptoms are cough and tachypnea, that appear in the early childhood with articular manifestations appearing lately before di age of 10. At laboratory tests, autoimmunity is characterised by the presence of autoantibodies, with the majority of patients showing a frankly positive ANA titer (80%). Rheumatoid factor is reported to be raised in 43% of cases; a few patients showed also positivity for cANCA and pANCA (2).
Objectives: We report one case referring to our centre of a young girl that presented at 3 year of age with polyarticular arthritis.
Methods: The patient was clinically evaluated by the pediatric rheumatology unit of Istituto Giannina Gaslini. The family gave permission for publication of clinical and laboratory data and photographic images as well as molecular analysis.
Results: The patient firstly referred to our centre at 3 year of age for a polyarticular arthritis involving metacarpophalangeal joints, hip and cervical spine. Lab results were performed showing raised ESR and CRP, high titer rheumatoid factor and antinuclear antibodies positivity. For a newly-onset of a persistent cough with no evidence of infectious process a chest X-ray and then a lung CT scan was performed showing an interstitial lung disease with tree-in-a-bud appearance and air-filled cysts. In order to treat the arthritic process oral and intra-articular steroids were administered with good response. Methotrexate and abatacept used as steroid-sparing drugs failed to control disease progression. The molecular analysis of *COPA* gene showed the reported c.698G > A mutation. Patient intentionally discontinued therapy for 3 years as well as clinical follow-up. After that time with patient referring again at our centre, a very severe osteoarthritis with joint damage and deformities of multiple joints was found.
Conclusion: COPA syndrome could be more common than thought due the autosomal dominant inheritance. When molecular diagnosis is confirmed a therapeutic aggressive approach is required to prevent permanent joint deformities and reduce the risk of life threatening complications such as pulmonary haemorrhages.
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Disclosure of Interest
None Declared.

P192

Paediatric autoinflammatory diseases in cyprus: insights from the national registry

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Introduction: Autoinflammatory diseases (AID) are rare in theory; however with raising awareness their incidence seems to be increasing. Early recognition and appropriate management prevents long-term sequelae and establishes an improved quality of life.
Objectives: To describe the epidemiology, demographic, clinical and disease characteristics as well as treatment options in patients with AID followed up in a national referral center for Pediatric Rheumatology.
Methods: Cross sectional observational study including thirty-five children with an AID regularly followed up at "Archbishop Makarios III" Paediatric Hospital of Nicosia, Cyprus from January 2012 to date. Children with PFAPA, SJIA and Behcet's disease were excluded from this cohort.
Results: Of those 35 children (55% boys), 62% were diagnosed with mutation proven FMF, 3.5% with TRAPS, 3.5% with HIDS, while the remaining were classified as a multisystem autoinflammatory disease. 69% of patients were Cypriots, 8% were of Eastern Mediterranean origin, 8% were Eastern European while the remaining were of mixed ethnic origin. Mean age at disease onset was 8.4 years (range 0.4 to 17 years). Mean time from disease onset to diagnosis was 1.5 years (range 0.6 to 3.5 years). Positive family history was evident in 25% of cases. All patients presented with fever of unknown origin. Other major symptoms included abdominal pain 65%, diarrhoea 18%, thoracic pain 7%, asthenia 27%, weight loss 14%, headache 42%, arthralgia and myalgia 23%. Skin manifestations were evident at 21% of patients; arthritis was present in 27% of patients, 25% of patients developed serositis (primarily pericarditis) at some point throughout the course of their disease. Lymph node enlargement was noted in 11%, parotid enlargement in 7%, while 14% had splenomegaly. Uveitis was present in 3.4%. Renal involvement was observed in 12% of our cohort (proteinuria). Renal biopsy was performed on two occasions proving secondary amyloidosis. Oral apthous ulcers were noticed in 15% of patients while hidradenitis suppurativa in 3.5%. The majority of our patients had received NSAIDS prior to referral; 22% reported temporal alleviation of their constitutional symptoms. Colchicine remained the mainstay of treatment for the majority of these patients (79%). 67% of these children had received steroids during the course of their disease. Eight 23% of the patients received