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The case study of undifferentiated autoinflammatory disease treated with anakinra

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Introduction: Patients with symptoms commonly found in autoinflammatory disorders may not fit a specific diagnosis, either because their clinical phenotype is nondiagnostic or genetic tests are negative. The term undifferentiated systemic autoinflammatory disorder (uSAID) has been used to describe such cases (*Harrison et al*., 2016). Recognizing uSAID requires a high degree of clinical suspicion and remains a diagnosis of exclusion with suboptimal treatment.
Objectives: To report an unsolved case of uSAID with successful anti-IL-1 treatment.
Methods: We report a case of a 10-year-old boy, presented in Children's Clinical University Hospital in Riga, Latvia in December 2013 with fever lasting for 3 weeks and knee joint pain.
The boy was born full term from physiological pregnancy and had no family history of autoinflammatory disease or consanguinity. In medical history he had surgical treatment of cryptorchidism, frequent pneumonia, bronchitis episodes and asthma diagnosis in later childhood. At the age of 7 he experienced an episode of bloody diarrhoea. Since the age of 8 he had recurrent acute otitis media.
In December 2013, while in hospital, ultrasound showed bilateral suprapatellar bursitis. Temperature elevations were not observed. The patient was treated as reactive arthritis with NSAIDs. In January 2014 he developed recurrent episodes with fever up to 40°C every 3-4 days. Laboratory tests showed CRP 10 mg/L, ESR 73 mm/h, negative autoimmune profile and normal results of bone marrow biopsy (BMBx). The examinations and clinical findings suggested systemic JIA (> 1 joint involved, fever > 2 weeks, hepatosplenomegaly).
Fever and knee joint pain disappeared at first with pulse corticosteroid (CS) treatment. Until July 2016, he received treatment with 6 CS pulses, low-dose oral CS daily and methotrexate (MTX).However, with CS dose reduction subfebrility and erythema nodosum-like (EN-like) lesions appeared periodically. Skin biopsy showed non-specific changes.
During July and August 2017, the boy had febrile temperature every other day when CS were not given and experienced aphthous stomatitis (possibly MTX-related). The patient became CS dependent. He had recurrent elevated temperature, fatigue, headache, abdominal pain, EN-like lesions.Secondary osteoporosis with vertebral compression fractures was diagnosed. Repeated skin biopsy showed undifferentiated panniculitis. Weber-Christian disease was excluded. Laboratory tests showed Serum Amyloid A 138 mg/L (N <6.4).The diagnosis of Still's disease was reconsidered. Genetic tests for 50 autoinflammatory genes and NOD2 gene were negative. Urinalysis showed normal mevalonic acid level. Treatment with Azathioprine 2 mg/kg and Plaquenil 200 mg once daily was started without significant response. In December he had a fever with CRP 99 mg/L, without signs of infection and CRP normalization without a/b therapy, with an episode of EN-like eruptions with non-specific panniculitis in histologic examination. BMBx remained normal. Cyclosporin A 3 mg/kg was initiated and condition improved slightly. In May 2018 Anakinra 100 mg/day was started, with good response: no recurrent episodes of fever, inflammatory marker values normalized, EN-like lesions appeared periodically.
Results: The boy presented with uSAID, with a time of 4 years to diagnosis, with the initial diagnosis of systemic JIA. The results of genetic testing were negative. The disease was only partially controlled by or unresponsive to DMARDs and steroid treatment. Good response was achieved with IL-1 blocker.
Conclusion: 1. USAID should be considered in the differential diagnostics of systemic JIA.
2. Genetic testing must be performed in patients presented with a range of nonspecific autoinflammatory symptoms.
3. We conclude that Anakinra seems a feasible treatment option for uSAID.

Consent for publication has been obtained from patient
Yes

Disclosure of Interest
None Declared

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Etanercept in the treatment of familial Mediterranean fever: a case report

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Introduction: Familial Mediterranean Fever (FMF) is an autoinflammatory disease characterized by recurrent fever,peritonitis,pleuritis or arthritis attacks.The main treatment of FMF is daily oral colchicine treatment. There is strong evidence that colchicine inhibits both acute inflammatory attacks of FMF and systemic amyloidosis.We aimed to report the response of a 2-year-old patient with colchicine-resistant FMF to the treatment of Etanercept.
Objectives: A two-year-old girl complained of recurrent fever one year ago. R202Q heterozygote positivity was determined in the gene analysis and colchicine 0.5mg / day was started with the diagnosis of FMF. Although the patient received this treatment regularly, she had fever and abdominal pain attacks for the last 4 months and her attacks started to increase day by day. She lost appetite and weight loss. The older brother had FMF in his family history. Physical Examination: height: 89,5cm, weight: 11,5kg, Fever: 38 degrees, abdominal sensitivity was present, there was no arthritis and rash. In laboratory tests; Sedimentation: 31mm/ h, C-reactive protein: 65mg / L, Fibrinogen: 4.27g / L. The patient was treated with colchicine 1mg / day, ibuprofen 200mg / day and prednisolone 5mg / day. Four weeks later, the patient complained of abdominal pain, fever, loss of appetite and weight loss. The frequency of attacks was not decreased.
Methods: The patient who was treated with subcutaneous Etanercept at 0.8mg / kg / week with the diagnosis of colchicine-resistant FMF, had no abdominal attacks and fever episodes.
Results: After 6 weeks there was no attack and sedimentation, CRP and fibrinogen levels decreased to normal limits.
Conclusion: There are no more alternatives for patients who do not respond to colchicine or cannot tolerate the drug.Considering cytokine patterns; Anti -TNF agents may be an alternative in the treatment of FMF.

Consent for publication has been obtained from patient
Yes

Disclosure of Interest
None Declared