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Patient with FMF presented by isolated myositis

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Introduction: Familial Mediterranean fever (FMF) is a genetic disease characterized by recurrent febrile episodes and mostly by the inflammation of serous membranes. We presented our case whom presented with acute myositis and was diagnosed as FMF.
Results: A six year and 5 mounth old girl complained of severe pain in her right leg and a gait while walking for a period of one week. It was learned from the history that the right ankle swelled and hiperemic macular rash were determined on lateral malleol area three years ago and the acute phase reactants were elevated at that time.There was no consanguinity and family history of autoinflammatory diseases. Physical examination of the patient revealed swelling, redness, thenderness, heat increase of on the calf muscles and limitation of extension of the knee joint. White blood cells, C-reactive protein (CRP), and eritrocyte sedimantation rate (ESR) were eleveated, muscle enzymes were normal levels.In the USG, the right popliteal fossa fat tissue was prominent and inflamed, and the neural structures were thick and edematous.Magnetic Resonance imaging of the thighs and right leg posterolateral muscles was consistent with myositis. After NSAID treatment, the clinic improved. However, the high CRP and ESR persisted for six mounths. There was no mutation on TRAPS gene. When the M694V homozygous mutation was detected in the FMF genetics of the patient, Familial Mediterranean Fever was diagnosed and colchicine treatment was started. Other autoinflammatory diseases genes could not studied. Acute phase reactans of the patient regressed after the treatment. The patient experienced arthritis attack at once for two years follow up.
Conclusion: The patients who carried M694V homozygous mutations may presented nonclassical findings of FMF.

Consent for publication has been obtained from patient
Yes

Disclosure of Interest
None Declared

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Interstitial lung disease in a newborn affected by mevalonic aciduria

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Introduction: Mevalonic aciduria (MA) is the most severe phenotype of mevalonate-kinase deficiency (MKD), with a onset in early infancy and poor prognosis. MA diagnosis may be challenging in the neonatal period given its rarity and its unspecific symptoms that frequently recall those of other neonatal diseases. To our knowledge, interstial lung involvement has never been described as onset feature in a newborn with MKD.
Objectives: We report the case of a newborn affected by MKD characterized by interstitial lung disease.
Methods: The patient underwent laboratory and radiology evaluation as clinically indicated. Direct Sanger sequencing was used to screen the 10 exons of the MVK gene.
Results: A female neonate born at term from consanguineous parents was referred to our hospital at 16 days of life (DOL) for mild hypotonia and persistent raised inflammatory markers despite antibiotic therapy. Infectious work-up was negative for both viral and bacterial infections. Chest x-ray revealed bilateral perihilar peribronchial thickening. Electroencephalography (EEG) reported moderate diffuse anomalies of background activity without major abnormalities. On DOL 20 the first episode of fever was recorded. Due to worsening tachypnea and persistent abnormal chest x-ray, a pulmonary CT scan was performed and showed diffuse ground-glass bilateral infiltrates consistent with alveolar-interstitial lung disease. On DOL 22 a palpable maculo-papular skin rash appeared on feet and hands, vanishing spontaneously 24 hours later. Bone marrow examination and levels of perforins, neuron-specific enolase and urinary catabolites of catecholamines were normal. A total body MRI was normal except for a mild cerebellar hypoplasia and the known interstitial lung disease. The patient kept presenting hypotonia, relapsing episodes of fever and skin rashes, developed anemia requiring blood transfusions and failure to thrive became evident. Type-I IFN signature was negative. A genetic test was requested, as well as quantification of urinary levels of mevalonic acid, which were markedly above the normal range. Direct Sanger sequencing allowed to detect a homozygous c.709A>T missense mutation in the exon 8 of the MVK gene, coding for a protein substitution p.T237S already classified as pathogenic in the INFEVERS database (http://fmf.igh.cnrs.fr/ISSAID/infevers/) and therefore consistent with the diagnosis of MKD. Both parents and her sister were found to be heterozygous carriers of the same mutation. On DOL 38 treatment with anakinra was started, with prompt regression of fever and skin rash, decrease in inflammatory markers, increase in reticulocytes count and weight gain. Hypotonia improved but persisted. The patient was discharged from hospital on DOL 56 in good clinical conditions, with acute phase reactants within the normal range and mild hypotonia. She is now 4 months old, still on anakinra treatment without adverse events.
Conclusion: Autoinflammatory diseases in the neonatal period are a diagnostic challenge. Clinical suspicion is crucial in order to perform specific laboratory and genetic testing and start appropriate treatment. Interstitial lung involvement may be present in MKD and, together with increased inflammatory markers, could be the first manifestation of the disease.

Consent for publication has been obtained from patient
Yes

Disclosure of Interest
None Declared

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Familial Mediterranean fever related damage assessed by auto-inflammatory disease damage index (ADDI) and associated factors with damage

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Introduction: Familial Mediterranean Fever (FMF) is the most frequent auto-inflammatory disease caused by MEFV gene mutations. Although FMF is characterized by intermittent inflammatory attacks some patients exert chronic persistent inflammation that can result