challenging in the newborn. To our knowledge, interstitial lung involvement has never been described as onset feature in 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. Chest x-ray revealed bilateral perihilar peribronchial thickening. Electroencephalography 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 groundglass bilateral infiltrates consistent with alveolar-interstitial lung disease. On DOL 22 a 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 severe anemia and failure to thrive. Type-I IFN signature was negative. A genetic test was requested, as well as quantification of urinary levels of mevalonic acid, which were markedly elevated. 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, classified as pathogenic in the INFEVERS database and therefore consistent with the diagnosis of MKD. Both parents and her sister were 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.

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AB1064

LONG-TERM SAFETY AND EFFICACY OF ADALIMUMAB IN GREEK ADULTS WITH JIA OR NON-INFECTIOUS UVEITIS AT THE TRANSITION PERIOD

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Background: Our previous publication on Juvenile Idiopathic Arthritis (JIA) revealed that half of the pts had flares in adulthood, despite previous administration of several anti-TNFs.

Objectives: a. To describe the long-term outcome of adults with JIA or non-infectious uveitis (NIU), exposed to Adalimumab (ADA) as the single

or last biologic during the transition period. b. To evaluate our model of transition in a subgroup of refractory to conventional DMARDs JIA or NIU pts, in the 1st Greek Transition Clinic for Pediatric Rheumatic Diseases.

Methods: Retrospective cohort analysis of Greek adults with JIA or NIU between 2004 and 2018. The JIA activity state was assessed by 2 quantitative tools: the Juvenile Arthritis Disease Activity Score-10 (JADAS-10, Clinical Remission [CR], Low, Moderate and High Disease Activity [LDA, MDA, HDA], respectively) and Wallace criteria (CR on/off ADA). Baseline was defined as the 1st ADA dose

Results: 28 pts were enrolled: 24 JIA pts (female 18), aged 20.6±3.1 yrs, for a f/u of 6.5±3 years (Group-A) and 4 female pts with active uveitis (2 with JIA-associated and 2 with NIU, mean aged: 18.9 yrs, f/u: 2.3 yrs) (Group-B). The age at baseline was 14.1±3.2 yrs in Group-A and 16.7 yrs in Group-B. The lag time from JIA onset to baseline was 6.3±3.8 yrs and the JIA subtypes were: 37.5% polyarthritis RF neg, 33.3% extended oligoarthritis, 20.8% enthesitis-related arthritis, 4.2% persistent oligoarthritis, 4.2% psoriatic arthritis. 11/24 (45.8%) pts (Group-A) and 3/4 pts (Group-B) were ANA positive, 5/24 JIA-pts had a history of uveitis (20.8%). Naïve to anti-TNF drugs were 18 pts (75%) of Group-A and 4/4 from Group-B. The baseline JADAS-10 was 13.9±5.9. The ADA yr-administration was 5.3±2.3 in Group-A and 2 in Group-B. Compliance to ADA had 17/24 (70.8%) pts in Group-A and all from Group-B. Regarding safety, 4/28 patients (14.3%) experienced Events of Special Interest (1/33.6 patient-years), herpes zoster (n=1), anorexia nervosa (n=1), breast fibroadenoma (n=1) and uveitis (n=1). Among the Group-A pts: 4/24 (16.7%) discontinued ADA due to inefficacy and 1/24 due to pregnancy planning. 6/24 pts (25%) achieved CR and discontinued ADA after 5.3±1.6 yrs. CR off ADA lasted 15.5±9.8 mo. The rest 13/24 continued ADA at the last f/u. In Group-B, 3/4 were still on ADA and 1 pt discontinued ADA due to NIU remission after 4.2 yrs and sustained CR 16 mo off ADA. The median% of CR remission on ADA total administration period was 75% in Group-A and 100% in Group-B. Among the ongoing receivers: a) the JIA activity state at the last assessment was: CR 84.6%, LDA 7.7% MDA 7.7% (median JADAS: 0, Group-A), b) 100% were in remission in Group-B.

Conclusion: 86% of adult pts with JIA or NIU tolerate ADA well. Pts exposed to ADA achieved CR on/off medication in 75% and 25%, respectively.

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AB1065

THE UTILISATION OF S100 PROTEINS TESTING IN PEDIATRIC RHEUMATOLOGY PATIENTS IN A TERTIARY CARE INSTITUTION AND IMPLICATIONS FOR CARE

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Background: S100 proteins are calcium-binding proteins of increasing value as biomarkers in various inflammatory conditions. The two highly studied members in this family, S100A8/9 and S100A12 have been considered sensitive biomarkers of disease activity in rheumatologic disorders as rheumatoid arthritis (1) and juvenile idiopathic arthritis (JIA) (2). They are considered more accurate than conventional inflammatory indices such as the erythrocyte sedimentation rate (ESR) and C-reactive protein (CRP) (3).

Objectives: To evaluate the utilization of S100 proteins in the clinical setting as supportive diagnostic tools as well as their performance in evaluating disease activity.

Methods: Patients seen at the hospital's specialty clinics who had S100 proteins tested during their care from April 2017 to August 2018. A retrospective chart review was performed to collect data on serum S100 protein levels, other inflammatory markers as CRP and disease activity measured as active joint count (AJC) in JIA patients and the presence of systemic symptoms in systemic JIA (sJIA) patients.

Graphpad Prism Version 7.05 was used for data analyses. Descriptive statistics were calculated for all variables. Comparisons were done by the Mann-Whitney U test and the Kruskal-Wallis test. Spearman correlation were calculated to assess association between S100 proteins and other numerical variables.

Results: A total of 164 patients were included. All 164 patients had S100A8/9 collected but only 151 had S100A12 samples. Patients with sJIA had higher levels of S100A8/9 and S100A12 compared to patients with non-systemic JIA (nsJIA) and periodic fever syndrome (PFS). CRP levels were not statistically different among diagnosis while S100 proteins were. Both S100A8/9 and S100A12 showed a good ability to differentiate sJIA from PFS (AUC=0.76 for both) compared to AUC=0.6 for CRP. At a cut-off value of 164 ng/ml, S100A12 was 71% sensitive and 73% specific in differentiating between sJIA and PFS (Figure 1). In JIA there was no correlation between S100 protein levels and AJC, either sJIA or nsJIA patients. S100A8/9 and S100A12 were significantly elevated in active sJIA patients compared to inactive ones while CRP levels were not different. S100 proteins levels were significantly higher during a flare of PFS compared to inactive disease, as well as CRP. There was no difference in S100 proteins and CRP in patients with or without tocilizumab treatment (small number on tocilizumab).

Conclusion: S100A8/9 and S100A12 are particularly elevated in sJIA compared to nsJIA and PFS. They are useful in distinguishing sJIA from other diagnoses with similar presentations, such as PFS. S100 proteins were significantly higher in active disease status of patients with sJIA and PFS compared to inactive status and their levels were closer associated with sJIA disease activity than CRP. The S100 proteins could provide valuable data in the work-up of patients with autoinflammatory conditions and monitoring disease activity in sJIA and PFS patients.

Figure 1. Panels (a) and (b) show serum levels of S100A8/9 (a) and S100A12 (b) across diagnoses. Values represent median and IQR (error bars). Comparison between diagnoses are done by Kruskal-Wallis test. Panels (c) and (d) show receiver operating characteristic (ROC) curves for S100A8/9 (c) and S100A12 (d) in differentiating systemic juvenile idiopathic arthritis (sJIA) vs. periodic fever syndrome (PFS). The area under the curve (AUC) for both S100A8/9 and S100A12 is 0.76. Sensitivities and specificities for the cut-off values are outlined.

* $p \leq 0.05$, ** $p \leq 0.01$, *** $p \leq 0.001$, **** $p \leq 0.0001$

Figure 1

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AB1066

DEFICIENCY OF ADENOSINE DEAMINASE 2: THE IMPORTANCE OF A DIAGNOSIS BEFORE IRREVERSIBLE DAMAGE

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Background: In 2014, two independent groups, Zhou et al.(1) and Navon-Elkan et al.(2), described the first 34 patients with disease-associated mutations in adenosine deaminase 2 (ADA2).

Objectives: To describe a new case of ADA2 deficiency (DADA2) in a 15 years old girl presenting with acute lower limb flaccid paralysis.

Methods:

Results: A 15 years old girl presented with acute lower limb flaccid paralysis. Neurologic exam showed peripheral deficit of the VII cranial nerve, absent motor activity at lower limbs with sensitive deficit. At physical examination she had marked and diffuse livedo reticularis. She underwent a brain and spinal magnetic resonance imaging (MRI) revealing multiple areas of signal abnormalities (T2 hyperintensity) in both thalamus and basal ganglia, small area in the spinal cord at C2-C3, at D10-D11 and L1 levels; these images were related to vascular damage. Blood exams showed high blood counts and inflammatory markers (C-reactive protein 16.76 mg/dl). Coagulation was normal, antiphospholipid and antinuclear antibodies were negative, immunological work-up showed normal immunoglobulins level, T and B phenotype. She received intravenous steroid and high-dose intravenous immunoglobulin. The constellation of findings led to clinical suspicion for DADA2, which was confirmed by low plasma ADA2 activity. For the presence of microhematuria, worsening proteinuria and hypertension, vascular ultrasound was performed with no evidence of renal artery stenosis. She underwent renal biopsy negative for glomerulonephritis, renal vasculitis and amyloidosis. Following the diagnosis of DADA2, she was initiated on etanercept 50 mg once weekly and she was able to taper steroid with normalization of inflammatory markers and improvement of skin rash.

The girl is the second of 3 children, born from consanguineous parents of Italian origin. By the age of 1 she started suffering from fever, arthritis, pericarditis with elevated inflammatory markers and was diagnosed with systemic juvenile idiopathic arthritis (sJIA) and chronic disease anemia. At that time, she already presented livedo reticularis. She was treated with short term oral steroid and methotrexate with remission of fever and articular symptoms. At the age of 2 she had a convulsive crisis, polysomnography and brain MRI were negative. One year later she presented a convulsive status epilepticus, brain MRI was repeated and resulted negative. She was started on topiramate with no further crisis, treatment was stopped 2 years later. When she was 9, she was admitted for pancytopenia and she was diagnosed of Leishmania infection. The two siblings of the girl were tested for plasma ADA2 activity and both resulted deficient. The boy is 12 years old with mild development delay and a previous diagnosis of sJIA when he was 5. He was treated with steroid and Methotrexate with remission. The older sister is now 19 years old and she had an episode of erythema nodosum. Genetic test of all family members is still pending.

Conclusion: Given the important morbidity, especially in consanguineous families, a high index of suspicion is needed for early diagnosis and precocious intervention, before persistent neurologic damage has been established. Our report describes for the first time a new neurological presentation as acute transverse myelitis in a patient with DADA2.

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