Kawasaki disease (KD) is an acute, self-limited vasculitis of unknown etiology with peak incidence at 9-12 months of age. The term Kawasaki Disease Shock Syndrome (KDSS) was introduced in 2009 after reports of hemodynamic instability during the acute phase of the illness. Antibiotherapy was started due to suspected septic shock/streptococcal citation, inotropic drugs and mechanical ventilation. Broad-spectrum antibiotics were prescribed due to fever and inflammation, and aspirin was added without response. Viral screening, all blood and urine cultures and ASO titers were negative. Myelogram revealed no signs of hemophagocytosis, neoplastic changes or growth of microorganisms in culture. Fine needle biopsy of a supraclavicular lymph node showed nonspecific inflammatory changes. Thoracocardioabdominal scan revealed moderate bilateral pleural effusion, mild hepatosplenomegaly, retroperitoneal/mesenteric lymphadenopathy. Brain MRI and echocardiogram were normal as well as ophthalmologic evaluation. In the presence of prolonged fevers, mucosal changes, extremity edema and non-specific rash in a severely-ill adolescent with no response to broad spectrum antibiotics and negative cultures, the possibility of KD was raised.

Due to ongoing fevers and inflammation after 36h of first IVIG, he received a second IVIG dose 2g/Kg, methylprednisolone IV pulses for 3 days and aspirin, with rapid clinical and laboratorial improvement. Within 24h the patient was off inotropic support, 48h off mechanical ventilation and 3 days later he was discharged from the ICU. His general condition continued to improve gradually, with increased platelet counts, normalization of liver function and CRP. In this phase, skin desquamation (buttocks/perineal region) was noted. At follow-up, thoracocardioabdominal angioMRI and serial eecardiograms were normal. He was weaned off corticosteroids and aspirin without recurrence of symptoms.

Results:

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Disclosure of Interests: None declared


A CASE OF ADENOSINE DEAMINASE 2 DEFICIENCY (DADA2) WITH AN UNCOMMON CLINICAL PRESENTATION AND RESPONSE TO IV IG

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Background: DADA2 is an autoinflammatory disease with autosomal recessive inheritance characterized by a heterogeneous clinical phenotype ranging from multisystemic inflammation (fever, polyarteritis nodosa, cerebral stroke, livedo reticularis etc.) to immune-dysregulation and immunodeficiency.

Objectives: To extend the clinical spectrum of DADA2 reporting a case of isolated nonspecific systemic inflammatory syndrome associated with signs of immune-dysregulation in a patient with a novel ADA2 mutation.

Methods: In a patient with nonspecific inflammatory phenotype associated to susceptibility to viral infections, Next Generation Sequencing (NGS) panel was performed; mutations detected were confirmed by Sanger analysis. ADA2 enzymatic activity was analyzed on a monocyte isolated from the patient and incubated with adenosine and an ADA1 inhibitor.

Results: The girl, adopted and of Asian origin, began to suffer from non-specific systemic inflammatory symptoms (persistent fever and arthralgias) at the age of 6. In past history recurrent respiratory infections and impaired immunological response to viruses (CMV related hepatitis, measles, after vaccination) were reported. After few months the patient developed clinical and laboratory findings of HHL (Hemophagocytic Lymphohistiocytosis), confirmed on bone marrow samples; treatment with intravenous (IV) high dose (HD) steroids was started, with prompt response. During steroids tapering fever and systemic inflammation reappeared; anti-IL1 treatment (anakinra) was not effective. Immunologic assessment demonstrated mild hypogammaglobulinemia and moderate NK deficiency on lymphocyte subsets. HD IV Immunglobulins (IG) (2 g/kg every month) allowed to achieve a complete control of the clinical picture; the frequency of administration was progressively reduced to every 4 months due to persistent wellbeing. At the age of 9, after switching IG to the

Disclosure of Interests: None declared


AB0972 Figure 2

AB0972 Figure 3

Disclosure of Interests: Lisa Gamalero: None declared, Ilaria Paglini: None declared, Gabriele Simonini Grant/research support from: Abbvie, Speakers bureau: Abbvie, Rolando Cimaz: None declared, Teresa Giani: None declared


AB0974

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Scientific Abstracts

AB0975 INITIAL BIOLOGICAL THERAPY RESPONSE IN PATIENTS WITH SUSPECTED AUTOINFLAMMATORY DISEASE

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Background: The choice of the initial biological therapy for patients with suspected autoinflammatory diseases and not conclusive genetic test remains challenging.

Objectives: To assess the clinical response to the initial biological therapy in pediatric patients with suspected autoinflammatory disease without any genetic diagnosis.

Methods: We retrospectively reviewed the clinical charts of patients followed in our clinic who started empirical biological therapy after being diagnosed with suspected autoinflammatory disease(sAID) due to the intensity of their symptoms and no response to colchicine or FAMEs.

Results: We identified 9 patients: 6/9 were male, median age at fever onset 1.33 years old IQR(0.46-4), age at diagnosis of sAID 3.8 years old IQR(1.75-7). Clinical presentation included fever(9/9), abdominal pain and arthromyalgia(7/9), aphthous(6/9), headache, rash and adenopathy(9/9), delayed growth(4/9), tonsillitis and pericarditis(3/9) as well as diarrhea and pleuritis(2/9). One patient presented with stroke, cutaneous lesions, vasculopathy and haemolytic uraemic syndrome and 1 patient with amyloidosis and secondary hepatosplenomegaly. None of the children suffered from uveitis or meningitis. The flares lasted a median of 14 days(IQR 8-20). Two patients had persistent symptoms. Their mean/median lab values are shown at table. 4/9 patients had homozygous mutations with uncertain significance, heterozygous mutations or polymorphism but their symptoms or familiar study was not suggestive of the corresponding AID. One patient had an heterozygous mutation in MEFv(p.P3696p.R408q) and also a CECR1 heterozygous mutation with uncertain significance, one patient had pR92Q heterozygous mutation in TNFRSF1A, one patient had MEFv pR202Q homozygous mutation, other patient had a NOD-2 heterozygous mutation and the patient with amyloidosis had NOB0 deficiency and a NOD2 mutation (p.A918D). All patients responded to steroid therapy; subsequently 8/9 received anti IL-1Receptor kineret as first biological therapy and 1/9 with suspected vasculopathy received anti-TNF. Response to IL-1R antagonist was complete in 3/8 and partial in 4/8 children:1/8 showed no response, 2/8 patients were switched to anti-TNF. One each to etanercept and Infliximab with good response. The patient with amyloidosis was changed to anti IL-6R with incomplete response but clear improvement compared to anti IL-1R response. The patient with suspected vasculopathy and initial anti-TNF treatment had partial response with no recurrent stroke but persistent severe symptoms.

Conclusion: An important group of patients with sAID lack genetic confirmation. Empirical use of IL-1R antagonist is promising but not effective in all patients as it was observed in our case series, where 5/8 children showed partial or no response.3/5 needing a second biologic treatment in form of anti-TNF due to persistent moderate-severe symptoms. A model to predict the response to different therapeutic strategies, based on clinical features and immune profile (including inflammatory cytokines) might help to choose the most appropriate immunomodulatory treatment.

Disclosure of Interests: None declared


AB0976 PSYCHIATRIC DISORDERSDURING TRANSITION CARE IN ADOLESCENTS WITH RHEUMATIC DISEASES


Background: Chronic rheumatic diseases (CRD) have a strong impact on psychosocial development of pediatric patients. There are several factors associated with psychiatric disorders (PD) in these children; physical disability, complex treatments, long-term follow-up, and flares, are the most cited in literature. Juvenile Idiopathic Arthritis (JIA) is the first cause of disability in children with CRD, on the other hand, Major Depressive Disorder (MDD) and Dysthymia are the third cause. Some PD, mostly MDD, appear as a consequence of disability caused by CRD, but immune pathways might be implicated in pathogenesis as well. Adolescents with CRD need to transition to an adult-centered care while deal with emotional and physical changes. This implies a difficult situation in which patients could be in higher risk for develop PD. There are a lack of information how this process affects emotional health in this population.

Objectives: The aim of the study is to calculate the prevalence of PD in adolescents with CRD during transitional care and their relationship with clinical and social factors.

Methods: Patients older than 16 years with an established CRD, who were in transitional care during the period between July 2017 and January 2019 were included in this transversal study.

We used MINI KID assessment tool to characterize PD in our patients. Each patient performs an interview with both a clinical psychologist and a

References:


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Disclosure of Interests: None declared


Mean SD Median IQR Min Max

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<th>PC75 18%</th>
<th>LysicaseU/ml</th>
<th>Neutrophils/μl</th>
<th>Lymphocytes/μl</th>
<th>Haemoglobin g/dl</th>
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Disclosure of Interests: None declared