Abstract AB0972 Figure 2

Abstract AB0972 Figure 3

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AB0973 ATYPICAL KAWASAKI DISEASE SHOCK SYNDROME CASE REPORT: NOT ALL SHOCKS ARE SEPTIC OR TOXIC

Sara Ganhão¹, Mariana Rodrigues², Francisca Aguiar², Margarida Tavares³, Iva Brito^{2,4}. ¹Centro Hospitalar e Universitário de São João, Department of Rheumatology, Oporto, Portugal; ²Centro Hospitalar e Universitário de São João, Pediatric Rheumatology Unit, Oporto, Portugal; ³Centro Hospitalar e Universitário de São João, Pediatric Infectious Diseases Unit, Oporto, Portugal; ⁴Faculty of Medicine of Oporto University, Oporto, Portugal

Background: 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.

Objectives: Present and discuss a rare case of KDSS in an adolescent patient.

Methods: Review of patient's clinical records and scientific literature.

Results: A 12 year-old boy was admitted in the pediatric intensive care unit (ICU) with a 5-day history of high persistent fever, abdominal pain, vomiting and headaches. On examination, he looked ill, was tachycardic and hypotensive with delayed capillary refill, had non-exsudative oropharyngeal erythema, cracked lips, terminal nuchal rigidity, jaundice and small palpable supraclavicular lymph nodes. On day 3 a generalized morbilliform rash and diffuse swelling of the hands became apparent. Blood gas showed metabolic acidosis. He had decreased platelet count of 72,000/mm³, AST 122 U/L, ALT 192 U/L, LDH 366 U/L, triglycerides 207 mg/dl, total bilirubin 85,5 µmol/l (direct 68,4). Renal function, urinalysis and fibrinogen were normal. ESR was 42 mm/h, CRP was markedly high (268 mg/l) and ferritin 544 ng/ml. Lumbar puncture revealed mild sterile pleocytosis. Due to progressive hemodynamic instability he required fluid resuscitation, inotropic drugs and mechanical ventilation. Broad-spectrum antibiotherapy was started due to suspected septic shock/streptococcal toxic shock syndrome. IVIG was empirically started 24h later due to non-

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. Thoracoabdominal 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, thoracoabdominal angioMRI and serial ecocardiograms were normal. He was weaned off corticosteroids and aspirin without recurrence of symptoms.

Conclusion: In this case, despite the atypical age and lack of some classical signs/symptoms, broad-spectrum antibiotic refractoriness and the described clinical presentation raised the hypothesis of KDSS. Diagnosis can be difficult, especially if shock occurs in incomplete forms of KD, but must be suspected early and treatment promptly started in order to ensure a good prognosis. Clinicians should be aware that thrombocytopenia and hepatitis are risk factors for refractory severe KD. MAS must always be excluded in cases of hemodynamic instability.

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AB0974 A CASE OF ADENOSINE DEAMINASE 2 DEFICIENCY (DADA2) WITH AN UNCOMMON CLINICAL PRESENTATION AND RESPONSE TO IV IG

Francesca Garbarino¹, Roberta Caorsi², Stefano Volpi², Alice Grossi³, Isabella Ceccherini³, Marco Gattomo². ¹Università degli studi di Genova, Genova, Italy; ²IRCCS Istituto Giannina Gaslini, Clinica Pediatrica e Reumatologia, UOSD Malattie Autoinfiammatorie-Immunodeficienze, Genova, Italy; ³IRCCS Istituto Giannina Gaslini, UOC Genetica Medica e UOSD Genetica e Genomica delle Malattie Rare, Genova, Italy

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 in 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 HLH (Hemophagocytic Lymphohistocytosis), 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 Immunoglobulins (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