ATYPICAL KAWASAKI DISEASE SHOCK SYNDROME: SHOCK SYNDROME

Background: Kawasaki Disease Shock Syndrome (KDSS) was introduced in 2009 after an unknown etiology with peak incidence at 9-12 months of age. The term KDSS describes clinical presentation raised the hypothesis of KSSD. 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 hemodinamic instability.

REFERENCES

Disclosure of Interests: None declared

Abstract AB0973

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

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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 120 beats per minute, tachypneic and 34 breaths per minute, with a temperature of 39°C. He had an ideal weight for age of 10th percentile. He had palpable supraclavicular lymph nodes. On day 3 a generalized morbiliform 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 fibri-
nogen were normal. ESR was 42 mm/h, CRP was markedly high (268 mg/l) and ferritin 544 ng/ml. Lumbar puncture revealed mild sterile pleocy-
tosis. Due to progressive hemodynamic instability he required fluid resus-
citation, inotropic drugs and mechanical ventilation. Broad-spectrum antibiotic therapy 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, neo-
plastic 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 effu-
sion, mild hepatosplenomegaly, retroperitoneal/mesenteric lymphadenop-
athy. Brain MRI and echocardiogram were normal as well as ophthal-mologic evaluation. In the presence of prolonged fevers, mucosal changes, extremity edema and non-specific rash in a severely-ill adoles-
cent with no response to broad spectrum antibiotics and negative cul-
tures, 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, normaliza-
tion of liver function and CRP. In this phase, skin desquamation (but-
tocks/perineal region) was noted. At follow-up, thoracoabdominal angioMRI and serial ecocardiograms were normal. He was weaned off corticoste-
roids 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 KSSD. 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 hemodinamic instability.

Disclosure of Interests: None declared

Abstract AB0974

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, cere-
bral 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, mea-
asles after inoculation) were reported. After few months the patient developed clinical and laboratory findings of HLH (Hemophagocytic Lympho-
histocytosis), confirmed on bone marrow samples; treatment with intrave-
nous (IV) high dose (HD) steroids was started, with prompt response. During steroids tapering fever and systemic inflammation reappeared; anti-
nIL1 treatment (anakinra) was not effective. Immunologic assessment dem-
strated 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 fre-
quency of administration was progressively reduced to every 4 months due to persistent wellbeing. At the age of 9, after switching IG to the...