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Poster abstract: 01

Singleton-Merten Syndrome: A rare syndromic type I interferonopathies with an evolving autoimmune domain (Case Report)

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Background: Type I interferonopathies represent discrete examples of a defective homeostatic regulation of this system with a striking phenotypic overlap. These disorders can present with a classic autoinflammatory phenotype with or without an adding autoimmune spill over. We describe here a 9-year-old male patient,1st sibling of 1st degree cousin marriage, presented with a lifelong history of recurrent episodes of fever, multiorgan involvement, developmental delay & pathological short stature. The patient was referred at the age of 4 years with history of mucoid diarrhea & failure to thrive since birth misdiagnosed as cow milk allergy. Throughout the first two years he suffered recurrent attacks of infections with an irrelevant assay for an immunodeficient status. At the age of three left ankle monoarthritis with periodic attacks of high-grade fever, abdominal pain was reported, ESR (85mm/1st hr.). Familial Mediterranean Fever genetic studies showed no abnormality. On examination, the patient was<5th centile for weight and height with peculiar facial features (high anterior hairline, broad forehead, smooth philtrum, thin upper vermillion, destroying caries), joint laxity, dry skin and multiple lentigines. Skeletal radiological survey revealed marked generalized demineralization. Echo study and ophthalmological examination were free. Immunological survey for an autoimmune status was negative. The unique features, prominent autoinflammatory state and the lack of autoantibody production raised the clinical suspicion of a pathology related to enhanced type I IFN signaling. Singleton-Merten syndrome was confirmed by genetic Studies. The patient showed marked clinical improvement on oral colchicine and full dose oral prednisone. While on therapy, the patient developed pancreatitis (resolved on Pulse methylprednisolone for 5 days). Six months ago, patient was diagnosed pathologically as Crohn's disease. Infliximab was added with a non-significant response. An interferon score and a trial of a targeted therapy are planned.
Conclusions: Autoinflammatory disorders are enigmatic diagnostic challenges. Singleton-Merten is an uncommon autosomal dominant genetic disorder with variable expression and diverse symptoms. It constitutes part of type I interferonopathy disease spectrum. Autoinflammation was prominent but with recent evolution of multisystem autoimmune involvement. Steroid dependence and limited efficacy of anti-TNF drove to a complicated situation.

Poster abstract: 02

Erythrodermic Atopic Dermatitis: A window to Systemic Vasculitis

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Background: Erythroderma is a potentially fatal dermatological emergency; it is characterized by generalized erythema and exfoliation involving > 90% of the body surface area. The most frequent cutaneous conditions linked to erythroderma are psoriasis, drug eruption, systemic contact dermatitis and atopic dermatitis. It is challenging to diagnose the underlying causes of erythroderma. Case presentation: A 13.5-year-old boy of consanguineous parents presented at the ER with fever, productive cough, bloody diarrhea and generalized pigmented lichenified skin lesions. No family history of similar condition or atopy. The patient's medical history includes a hospital admission for pneumonia at the age of two months, followed by repeated hospital admissions for severe episodes of wheezy chest. The patient has been receiving asthma controller therapy since the age of twelve. He had widespread erythematous eczematous skin lesions at the age of six. Skin biopsy revealed hyperkeratosis and elongated rete ridges, together with intra-epidermal vacuolar degeneration associated with

large number of neutrophils. The underlying dermis shows inflammatory cells consistent with pustular dermatosis. The patient also reported allergies to several foods, including bananas, fish, milk, and strawberries. Physical examination showed fever 38.5°C, respiratory rate 35 breaths/min. Oxygen saturation was 98% on room air. His weight and height were below the 3rd percentile. Local Chest examination showed bilateral crepitation. Abdominal examination revealed tender rigid distended abdomen (acute abdomen). Abdominal ultrasound that showed target sign of intussusception that improved with conservative management. CT scan of the chest confirmed the left lower bronchiectasis alterations seen on the chest x-ray. Pan sinusitis with an anterior nasal septum defect measuring approximately 1.2 cm was revealed by a Sino-Nasal CT scan. A contrast-enhanced CT scan of the abdomen revealed extra- and intrahepatic biliary radicle dilatation but no definite signal void/ stones which could indicate sclerosing cholangitis. Additionally performed, MRCP which verified the outcomes. Echocardiography results were normal. The patient also experienced numbness and was unable to elevate his right leg while climbing stairs, so NCV was done and showed mild axonal neuropathy affecting right common peroneal and posterior tibial nerves. Flexible bronchoscope was done that revealed left lower sequestrated lobe with severe inflammation and yellowish thick secretions. BAL cytology showed large number of neutrophils in a background of proteinaceous eosinophilic material, no fungal colonies and the microbiological analysis showed Pseudomonas spp. Presence of asthma, peripheral eosinophilia, pulmonary infiltrates, paranasal sinusitis, and multi-organ system involvement, our patient met the American College of Rheumatology diagnostic criteria for Eosinophilic granulomatosis with polyangiitis (Churg-Strauss syndrome). The patient went into remission on oral steroids with Mycophenolate mofetil and elimination diet together with antimicrobials. To the best of our knowledge, this is the first pediatric case of EGPA presenting with erythrodermic atopic dermatitis, bronchiectasis, and sclerosing cholangitis which are infrequent with a worse presentation to present separately. This case highlights a rare condition that causes erythroderma and stresses the need to reevaluate such patients to diagnose the underlying disease.

Poster abstract: 03

MIS-C associated Nephrotic Syndrome and Kawasaki like Syndrome with recent FMF: Case Report

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Introduction: Since the establishment of COVID-19 infection and MIS-C among the pediatric age group, we are faced with different uncommonly associated spectra of illness. Hereby, we experienced one of these uncommon presentations in the following case report. Case presentation: A three and half year-old-male patient, of non-consanguineous parents, presented one year ago, with acute onset, progressive course of post viral infection generalized pitting edema of several days' duration. Provisionally diagnosed as nephrotic syndrome and confirmed through laboratory investigations including nephrotic range proteinuria (albumin/creatinine ratio 24979.17 mg/g), serum albumin 1.76 g/dl, fasting triglycerides 372mg/dl, total cholesterol 453 mg/dl. Although afebrile, the boy was toxic, tachycardic, hypertensive with chest asthma and hepatosplenomegaly and the boy was admitted in the pediatric ICU. Laboratory investigations revealed normal white blood cell count 8,283/cm with relative lymphopenia (2,893/cm), normocytic normochromic anemia (Hb 8.1 g/dl), normal platelet count 338,000/cm and reticulocyte count 2.8% without fragmented red blood cells. ESR was 84 in the first hour. Kidney functions and liver enzymes were normal and C-reactive protein (CRP) was negative. Further investigations denoted elevated levels of D-dimer (2.1 mg/dl), interleukin (IL)-6 (5.5%), creatinine kinase (CK)-MB (63 mg/dl, N< 25) without parallel elevation of the ferritin serum level (33.6 mg/dl). Covid-19 IgG was positive. Radiologically, the patient had mild bilateral pleural effusion, minimal perihepatic ascites and increased renal sizes with preserved echogenicity. Diagnosis of multisystem inflammatory syndrome (MIS-C) was settled with echocardiographic findings of mild mitral regurgitation, concentric left ventricle hypertrophy, mild pericardial effusion and significant uniform dilatation in the left coronary artery (z score 5.2). Treatment included IVIG 2 gm/kg, pulse methylprednisolone followed by prednisolone 2mg/kg/day and prophylactic dose of low molecular weight heparin with marvelous systemic and cardiac response in the 1st week followed by renal response within 3 weeks. After tapering steroids completely, partial renal relapse occurred and renal biopsy was done after 6 months of the primary disease, showing focal mesangial proliferative glomerulonephritis

suggestive of resolving postinfectious glomerulonephritis. Oral mycophenolate mofetil was initiated with gradual withdrawal of steroids with incomplete remission in attacks. During the last three months, the patient experienced two attacks of unexplained acute high-grade persistent fever associated with severe abdominal pain and non-nephrotic proteinuria lasting for 3-4 days, with symptom-free interval of 6 weeks. Considering the recent maternal and maternal pedigree diagnosis of familial Mediterranean fever (FMF), inflammatory markers were done during the febrile illness. Highly elevated serum amyloid A level (>146 mg/dl), mild proteinuria (+1), normal ESR (20 in the first hour), CRP (1.9 mg/l) and absolute neutrophil count (5.33/cm) were found and normalized amyloid A four days later. Genetic testing for FMF revealed heterozygous mutation at the codon M694I. Currently the patient is on therapeutic trial of oral colchicine therapy for follow up. Conclusion: Patients with MIS-C should be cautiously dealt with during their initial presentation and follow up, particularly, those with unresolved chronic illness and on prolonged treatment with immunosuppressant agents.