

**P371 TWO MISSED DIAGNOSED PATIENTS WITH STING-ASSOCIATED VASCULOPATHY WITH ONSET IN INFANCY IN CHINA**

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**Background and aims** Stimulator of interferon genes (STING)-associated vasculopathy with onset in infancy (SAVI) is firstly described in 2014 as a type I interferonopathy that resulting from heterozygous mutations of *TMEM173*. This gene encodes the STING adaptor protein, and mutations lead to a gain of function on STING and overproduction of interferon beta. SAVI is characterized by neonatal-onset systemic inflammation, a severe cutaneous vasculopathy and interstitial lung disease. JAK inhibitors are considered as an effective therapeutic strategy. We sought to describe two missed diagnosed patients with SAVI (P1 and P2) in order to draw attention to this illness.

**Methods** The clinical data were collected and Sanger sequencing of the gene *TMEM173* was performed. An introspection of missed diagnosis and differential diagnosis was discussed.

**Results** The two boys shared similar manifestations including recurrent skin abscess in winter with skin lesions and recurrent respiratory tract infections since their births. It occurred to us neutrophil defects like CGD but NBT test and DHR123 were normal for both of them in 2011. Peroxidase staining was positive and no mutations in *MYD88* (P1). Computed tomography of the chest revealed pulmonary fibrosis yet no relevant genes (including *ABCA3*, *SFTPC*) mutations were found. Joint pain was significant for P2 but Naproxen was ineffective. Treatments with antibiotics turned out little improvement and wouldn't prevent the progression. Finally, both of them died of respiratory and circulatory failure in 2012 and 2016 respectively. Recently, genetic mutations (c.463 G>A and c.461 A>G) in exon5 of *TMEM173* were discovered, confirming the diagnosis of SAVI.

**Conclusions** Owing to unfamiliarity with this disease, some cases would have been misdiagnosed or missed diagnosed. Thus, we propose that SAVI should be taken into consideration for children with chilblain skin lesions and pulmonary fibrosis after ruling out other related genes mutations.

**P372 CLINICAL CHARACTERISTICS OF A CHILD WITH MENDELIAN SUSCEPTIBILITY TO MYCOBACTERIAL DISEASE DUE TO MUTATIONS OF IL12RB1**

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**Background and aims** Autosomal recessive interleukin-12 receptor  $\beta$ 1 (IL-12R $\beta$ 1) deficiency is the most common cause of Mendelian susceptibility to mycobacterial disease (MSMD). Here we report a case with multi-drug resistant tuberculosis (MDR-TB) due to mutations of *IL12RB1* to investigate the clinical characteristics of MSMD.

**Methods** The clinical features of a child with mutations of *IL12RB1* were summarized and the mutations were analyzed by Sanger sequencing.

**Results** The 10-year-old boy was vaccinated with Bacille Calmette-Guérin (BCG) at birth, and suffered BCG disease within 3 months of age. A progressive left side axillary adenopathy was developed, then infections disseminated to the lung, thoracic cavity, peritoneal cavity, intestine, brain, skin, and ear. The patient was diagnosed with MSMD due to *IL12RB1* deficiency at five years old. There were no significant abnormalities in routine immunological examinations including lymphocyte subsets, immunoglobulins, complement and neutrophil respiratory burst test. The child didn't receive early and standard anti-tuberculosis therapies after onset, and the disease progressed to MDR-TB. During the whole disease course, the anti-tuberculosis treatments were adjusted several times owing to poor responses. rIFN- $\gamma$  was added twice a week but stopped because of the adverse reaction of fever. The infections were uncontrolled. Genetic testing revealed the compound heterozygous *IL12RB1* mutations c.632G>C in exon 7 and c.1106T>C in exon 10 inherited from father and mother respectively. Those variations lead to R211P and I369T amino acid changes reported previously.

**Conclusions** Mutations of *IL12RB1* can lead to severe MSMD. When there were no obvious abnormalities in routine immunization assessments of patients with BCG disease, the possibility of MSMD needs to be considered. *IL12RB1* protein detection and gene analysis are helpful for diagnosis. Standard anti-tuberculosis treatments are conducive to tuberculosis or BCG infection control. The efficacy should be evaluated to determine whether to stop it when adverse reactions of rIFN- $\gamma$  happen.

**P373 YOU GIVE ME FEVER! – THE AUTOINFLAMMATORY CLINIC IN AN IRISH TERTIARY PAEDIATRIC HOSPITAL**

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**Introduction** Auto-inflammatory syndromes (AIS) are inherited disorders of the innate immune system that lead to pathogenic inflammation. Multiple organ systems can be involved and disease can lead to significant end organ damage. Early diagnosis and treatment may improve outcome;<sup>1</sup> Diagnosis is often delayed due to the low incidence of these disorders. An auto-inflammatory clinic was established in O'Connell's Crumlin, Dublin, a tertiary level Paediatric Hospital, in November 2015 to assess patients with known or suspected auto-inflammatory disease within a multi-disciplinary setting. This clinic is attended by immunology, rheumatology and dermatology specialists with multi-disciplinary team input as required.

**Aim** The aim of this study was to survey the diagnosis and management of patients attending this clinic over a 2 and a half year period.

**Methods** A retrospective observational chart review of all patients attending the Autoinflammatory clinic from November 2015 to June 2018. Age of onset, demographic details, diagnosis (if known) and management were documented. Details of any genetic analysis if undertaken were also included.

**Results** A total of 47 patients attended the auto-inflammatory clinic over the identified period. Age of onset at first