

response to protein and polysaccharide antigen with normal floctometric study. **Results:** In this patients whole exome sequencing revealed a homozygous mutation in DNMT3B with a c.2397-11G>A (intronic) (rs547940069) variation. Mutation in DNMT3B cause Immunodeficiency-centromeric instability-facial anomalies syndrome. In order to confirm the diagnosis of ICF syndrome a conventional karyotype was performed to evaluate the presence centromeric instability at chromosomes 1,9 and 16. The karyotype confirm the presence of centromeric instability and the diagnosis of ICF syndrome. **Conclusion:** ICF syndrome should be considered in young children with growth retardation, facial anomaly like as saddle nose, epicantal fold, hypertelorism and recurrent infection.

**Keywords:** Growth retardation, recurrent infection, centromeric instability

**Abstract ID: 096**

**Novel Interferon Alfa Receptor1 Mutation: Death after MMR Vaccination in 2 Yreas Old Baby and Meningoencephalitis in His Brother**

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**Background:** Interferon alfa and beta receptor subunit1 (IFNAR1:cytogenic location; 21q22.11) encodes a type I membrain protein that associate with IFNAR2 to form the type I interferon receptor, receptor for interferons alfa and beta.deasease associated with IFNAR1 include septicemic plague, pnemonic tolaremia, hepatic c infection, papillomatosis and multiple sclerosis. **Methodes:** The patients were 2 brother. First brother died after MMR vaccination at 2 years old and other developed sever meningoencephalitis at 6 years old.this patient had normal laboratory finding.he had normal lymphocyte count, normal antiboby response to protein and polysaccharid antigen and normal floctometric study. **Results:** Whole exome sequencing showed novel mutation in IFNAR1 at spelling site of exon 6.Our sanger analysis indicated that the parents , as it was expected, are heterozygous for such mutation. **Conclusion:** As far as we know non pathogenic mutation has been reported befor. However, given the function of IFNAR1, the high effect of splicing

mutation and heterozygosity of parants, causative effect of this mutation on the disease in is highly suggested.

**Keywords:** MMR vaccination, Death, Interferon alfa receptor gene mutation

**Abstract ID: 141**

**Hyper IgE Syndrome in Cipto Mangunkusumo Hospital: A Case Series**

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**Background:** Hyper IgE syndrome (HIES) is a primary immunodeficiency characterized with eczema, recurrent infection of skin and lungs, and increase of IgE level. Diagnosis was based on clinical manifestation, IgE level, and genetic examination. A scoring system by Grimbacher was accepted by the National Institue of Health (NIH) to identify patients with HIES. **Methods:** We describe 4 cases with HIES during 2013-2015 in Cipto Mangunkusumo Hospital, Jakarta. **Results:** The 4 cases with feature of Hyper IgE syndrome consist of 2 boys and 2 girls. Range of age at the time of diagnosis was 1 to 7 years old. All of the patients had skin problem (abscess, rash), recurrent respiratory tract, candida infections, 2 of them had bronchiectasis and multiple bullae lungs. Time from onset to diagnosis was 1 to 6 years. The highest IgE level for each patient was ranging from 10, 442 to 130, 420ng/mL. NIH scores for HIES were ranging from 21 to 43. Two patients had NIH score more than 40. Three patients get itraconazole and cotrimoxazole prophylaxis. Genetic evaluation was not done in these patients. **Conclusions:** It is a great challenge to diagnose HIES for clinician. Scoring system by NIH can be used to identify patients with HIES, however the genetic examination should be done to confirm the disease.

**Abstract ID: 143**

**Approach to Pediatric Patients with Phagocyte Defects**

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Defects of neutrophil function and/or differentiation, defects of motility, and defects of respiratory burst are the main sub-classification of phagocytes defects, while Mendelian susceptibility to mycobacterial diseases (MSMD) should also be considered as a progressing area. Severe congenital neutropenia (*ELANE*, *GFI1*, *HAX1*, *G6PC3*, *VPS45*, etc), cyclic neutropenia, glycogen storage disease type 1b, p14 deficiency, Barth syndrome, Cohen syndrome, and Clericuzio syndrome poikiloderma with neutropenia are primary immunodeficiency diseases (PIDs) with neutrophil function/differentiation defects, while leukocyte adhesion deficiency (LAD types I-III), Rac2 deficiency,  $\beta$ -actin deficiency, localized juvenile periodontitis, Papillon-Lefèvre syndrome, specific granule deficiency, and Shwachman-Diamond syndrome are classified in group of motility defects. Chronic granulomatous disease (*CYBB*, *CYBA*, *NCF1*, *NCF2*, *NCF4*) is the prototype of defects of respiratory burst. MSMD is another group of PIDs that predisposes individuals to mycobacterium. Mutations in several gene loci have been detected for MSMD, including *IL12RB1*, *IFNGR1*, *IFNGR2*, *IL12B*, *STAT1*, *CYBB*, *IRF8*, and *ISG15*. GATA2 deficiency (Mono MAC Syndrome), pulmonary alveolar proteinosis along with AR form of *IRF8* deficiency are other diseases that have been classified as phagocytes defects. Long-term follow-up of patients with known gene mutation(s) could give us more insight into the pathophysiology of diseases, while the clinical description of patients could also help us to have better understanding on nature of these diseases.

#### **Abstract ID: 149**

#### **Case Report: Fatal Case of Chromobacterium Violaceum Septicaemia in a Patient with Chronic Granulomatous Disease**

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*Chromobacterium violaceum* infection is rare but causes a high mortality rate particularly in immunosuppressed persons. A localized infection

followed by an overwhelming septicaemia and metastatic lesions is the usual pattern of this illness. We report a 5-year-old boy presented with right foot infection and rapidly progressed to gangrenous of toes and fulminant septicaemia with brain abscesses and end organ failures. Multiple sets of blood cultures, pus and tissues swab yielded *C. violaceum*. Primary immunodeficiency studies was suggestive of Chronic Granulomatous Disease which showed profoundly low respiratory burst response to PMA and p47-phox deficiency with p47-phox band was absent in the RT-PCR and deltaGT scan showed homozygous  $\Delta$ GT at beginning of exon 2 of *NCF1* gene. Since the first case of *C. violaceum* infection in human from Malaysia in 1927, more than 100 cases has been reported worldwide. To our best knowledge, seven other cases have been reported previously from Malaysia including two CGD patients.

#### **Abstract ID: 152**

#### **Classic Wiskott Aldrich Syndrome with Mild Symptoms in Two Cousins: A Case Report**

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**Background:** Wiskott-Aldrich syndrome (WAS) is characterized by microthrombocytopenia, eczema, recurrent infections, and an increased incidence of autoimmunity. Classic WAS presents with severe clinical symptoms, commonly. **Material:** We report a new phenotype of classic Wiskott-Aldrich syndrome (WAS) with mild symptoms in two cousin who were 7 years of age. They did not present severe infections or hemorrhage despite their genetic assessment revealed mutation in WAS gene. The symptoms and infections of the patients were removed after treatment with IVIG and prophylaxis antibiotic.

**Results:** Exom sequencing for WAS gene performed. Hemizygous missense mutation, NG\_007877.1: g. 1974G>A, c. 397G>A, p. E133K, a known disease-causing mutation variant for the classical WAS was reported. **Conclusion:** It seems that our patients presented a novel clinical phenotype of classic WAS with milder symptoms than usual.

**Keyword:** Wiskott-Aldrich syndrome, Symptom, Phenotype