diagnostic work-up are essential. We report the case of a 3 year old previously well girl, who was referred to A&E with shortness of breath. On admission, she was found to be in acute heart failure.

Echocardiography showed a restrictive cardiomyopathy and a very large pericardial effusion which was subsequently drained. A full cardiomyopathy screen was performed but all her metabolic indices were normal. Genetic investigations identified a novel MYH7 variant. The *MYH7* gene is usually associated with hypertrophic cardiomyopathy. This patient is the second in the world in whom RCM has resulted from a variant invariant in *MYH7*, the first with a phenotypic effect and provides further evidence that this gene is linked with different cardiac phenotypes.

511 ATYPICAL WHEEZING CASES DURING INFANCY DUE TO GASTRIC VOLVULUS

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Background Gastric volvulus is rarely seen abnormality during childhood. Nonbilious vomiting, abdominal distension and dyspepsia are the most common presenting symptoms. Yet, it may cause reactive airway disease symptoms such as wheezing attacks, because of gastroesophageal reflux, and chronic cough in infants. Surgery after early diagnosis in gastric volvulus resolves symptoms completely and its prognosis is excellent.
Aim Here, two infants having chronic cough and recurrent wheezing attacks, unresponsive to bronchodilators, are presented.
Patients/Methods Physical examination of both cases revealed about 6-month-old male patients having recurrent wheezing, tachypnea, dyspnea and subcostal retractions. Widespread ronchi and sometimes rales were heard on both lungs of the patients. Rest of the examination was normal. In their laboratory evaluations, acute phase reactants, renal and liver function tests were within normal. Immunoglobulin G, its subgroups and other immunoglobulins were found to be normal. Sweat tests were normal. High resolution computerized tomography (HRCT) demonstrated minimal mosaic pattern on both lung parenchyma. Bronchoscopy was normal in the first infant. Cranial, abdominal ultrasonography and fundus (eye) examinations showed normal findings in both patients. Cardiac examination and echocardiography were normal. Oesophago-gastro-duodenography showed gastric volvulus, organo-axial and mesentero-axial, in both infants; respectively.
Results After the surgery for gastric volvulus and gastroesophageal reflux in the first case, the symptoms entirely improved. But, the symptoms of second case resolved without surgery.
Conclusion Our cases are being reported to emphasize the necessity of thinking gastric volvulus in the differential diagnosis of atypical wheezing infant, even which it is very nadir cause.

512 NEONATAL TESTES TORSION: CASE REPORT

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Introduction Torsion of the testes results from twisting of the spermatic cord, which leads to a compromised testicular blood suply and subsequant testicular infarction. The consequant ischemic damage affects long-term testicular morphology and sperm formation.
Case We describe a case of 2 day-old boy who presented with bilateral visible swelling in the scrotum. Physical examination findings showed enlarged, hard, nontender right scrotal mass, and left hydrocele. Doppler ultrasonography demonstrated torsion of right testes and bilateral hydrocele. Exploration was performed using a median raphe incision in the scrotum. Right testes was delivered, detorsed, and placed in warm, moist sponges but it appeared nonviable and necrotic. Right orchiectomy was done. Left testes was normal, it was fixed to the scrotum. Postoperative recovery was uneventful.
Conclusion Perinatal testicular torsion is a rare condition. During newborn examination it must kept in mind if a scrotal mass or swelling occurs.

513 HEERFORDT´S SYNDROM IN AN ADOLESCENT BOY

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Introduction Heerfordt's Syndrome is characterized by bilateral uveitis, facial paralysis, fever and parotitis which is a rarely seen condition in sarcoidosis with neurological manifestations.
Case Report A fifteen year old boy admitted to our pediatric emergency service with fever, vomitting and swellings on his cheeks. Bilateral conjunctivitis, bilateral preauricular swellings, maculopapular rash on his anterior and posterior body areas and uncertain neck stiffness were detected on his physical examination. The rest of the examination was considered as in normal range. His medical history revealed an upper respiratory tract infection one week ago.

Lomber puncture was performed because of patient's neck stiffness. No cell and culture growth were detected on puncture sample. During his observation, left peripheral facial paralysis was developed and bilateral uveitis was detected while his high fever was still persisting.

Whole body gallium 67 scan was performed. Focal accumulations of gallium 67 in both lacrimal glands, parotid and submandibular glands(panda sign) were seen. Because of existence of uveitis, parotitis, left peripheral facial paralysis and fever, the patient was diagnosed as Heerfordt's Syndrome. Steroid (Prednisolone) treatment was started. The regressions of facial paralysis and parotid gland size were noticed after two weeks of the steroid therapy.
Result This case report was chosen to take attention to a rare cause of parotitis.

514 THE AUTOIMMUNE LYMPHOPROLIFERATIVE SYNDROME: A CASE REPORT

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Background The Autoimmune Lymphoproliferative Syndrome (ALPS) is an impairment of lymphocyte apoptosis expressed by generalized non-malignant lymphoproliferation, lymphadenopathy and/or splenomegaly. Majority of patients with ALPS harbor heterozygous germline mutations in the gene for the TNF receptor-family member Fas (CD 95, Apo-1) which are inherited in an autosomal dominant fashion. Somatic Fas mutations are the second most common genetic etiology of ALPS.
Case report We describe a two year old boy who was admitted with hepatosplenomegaly, generalized lymphadenopathy and anemia. Histopathological and immunohistochemical analysis of lymph nodes suggested a lymphoproliferative disorder in large granular lymphocytes. The lymphocyte phenotyping performed in the patient showed an increased population of T cells αβ double

negative (LTCD3+ TCR αβ+ CD4- CD8-) about 27% (control < 2.5%). The study of FAS gene allowed the identification of a mutation in exon 9.

Conclusion ALPS is an underestimated entity that must be considered in non malign lymphoproliferation, autoimmunity and expansion of an unusual population of a/ß CD3+CD4-CD8- (double-negative T cells>1%).

515 A RARE CASE OF LANGDON DOWN SYNDROME WITH COMPLETE ENDOCARDIAL CUSHION DEFECT, TETRALOGY OF FALLOT, DEFICIENCY OF FACTOR VII

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Aims We sought to summarize a very rare association between multiple rare incidence diseases in a patient with Langdon-Down syndrome and also to correctly document each pathology and use the best course of treatment.

Background Factor VII deficiency has an incidence of 1 in 500.000 reported cases. Complete endocardial cushion defect [ECD] occurs in 2% percent of all congenital heart defects. Additional cardiac abnormalities (persistent ductus arteriosus and tetralogy of Fallot [ToF]) may occur in 10% of all ECD's. Associated defects are rare in children with Down syndrome.

Methods A 5 weeks old infant with a Down phenotype was admitted in the Intermediate Care Unit for severe tonic-clonic seizures and an unexplored heart murmur. A computed tomography scan revealed a massive hemorrhaging in the fronto-parieto-occipital left cerebral region. Trauma was excluded and the prothrombin time was prolonged with the activated partial trhomboplastin time normal so we sent a blood sample for the factor VII activity.

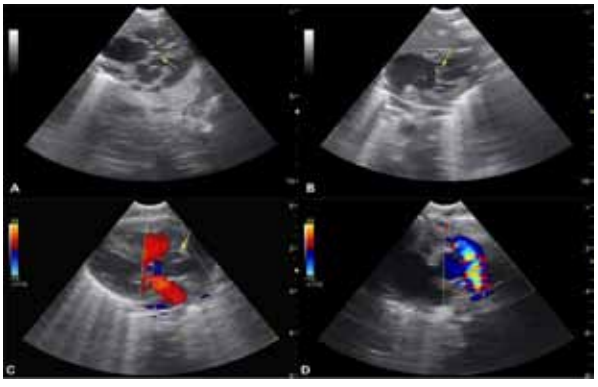
We performed an echocardiography.

A karyotype study was carried out.

Results

- Complete ECD with the common atrio-ventricular valve in dextroisomerism, left ventricle hypoplasia, associated with ToF.
- The factor VII activity showed a 2% activity level
- classical 21 trisomy

Conclusion We provided a good documentation of a very rare association between separate severe pathologies and we showed that when faced with a congenital malformative syndrome one should never stop looking for other abnormalities.



Abstract 515 Figure 1

A- subcostal view- the arrow points to the insertion of anterior left leaflet on a papillary muscle, the X's show a ventricula

B- subcostal view- the common AV valve and the atrial septal defect;

C- parasternal long axis- the over-riding aorta;

D- parasternal short axis- turbulent flow through the pulmonary valve (pulmonary artery stenosis)

516 A VERY RARE CASE REPORT OF HERMANSKY-PUDLAK SYNDROME TYPE II

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Introduction Hermansky-Pudlak syndrome type 2 (HPS-2) is a very rare multi-system disorder characterized by oculocutaneous albinism, reduced visual acuity, horizontal nystagmus, bleeding diathesis and recurrent infections due to neutropenia and impaired cytotoxic activity. HPS-2 is caused by mutations in the AP3B1 gene (5q14.1) and is transmitted in an autosomal recessive manner. The gene product is the Beta 3A subunit of adaptor protein 3 (AP3), involved in vesicle formation and protein sorting. Here we report a very rare case of HPS-2 who admitted because of fever.

Case Report A 5 months-old female patient admitted to our hospital because of fever. She is the first child of a consanguinous parents. She had mild facial dysplasia, whitish-yelllow hair and horizontal nystagmus. Opthalmological evaluation showed oculocutaneous albinism. Moderate hepatosplenomegaly was revealed. Anemia (Hb; 8.3 gr/dl) and neutropenia (0.4×109/μL) with normal platelet count were documented. Bone marrow aspiration yielded hemophagocytosis. Triglyceride, ferritin and fibrinogen levels were in normal limits. She was treated with proper antibiotic treatment and discharged to follow-up in outpatient clinic. Neutropenia was subsequently fluctuated. She had been hospitalized six more times due to febrile neutropenia and at each admission cytopenia including thrombocytopenia (15×109/μL) in addition to hepatosplenomegaly were revealed. Time to time increased triglyceride levels were documented. All episodes were resolved with proper antibiotic and r-HuG-CSF treatment,. without requiring HLH treatment. Genetic analysis revealed homozygous nonsense mutation in exon 18 of the AP3B1 gene.

Conclusion Patients with albinism and opthalmological complaints should be evaluated for Hermansky-Pudlak syndrome.

517 THALASSEMIA PREVENTATION AND ACTIVITY OF PEDIATRIC HEMATOLOGY AND ONCOLOGY DEPARTMENT AT BANGABANDU SHEIKH MUJIB MEDICAL UNIVERSITY IN BANGLADESH

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Background and Aims Thalassemia is a genetic and crucial disease. Approximately 240 million peoples are suffering from this disease. Every year 10 million children are suffering from this disease. Hb-E dieases is available in south east Asia, north east India and Bangladesh. Originalally Hb-E disease is 5 times more than Beta Thalassemia in Bangladesh.

Method The outbreak of this disease is not calculated at this moment but carrier is 15 million. We are collected experimental data from 3 hundred voluntiers in our center. They have not family history of Thalassemia. In that experimental data Beta Thalassemia carrier 2.33% and Hb-E Carrier 10%. If we are experiment among the people who have history of Thalassemia, this disease is increased no doubt. This disease have actually no curable treatment except BoneMarrow tranplantation. Treatment cost is excessive and unbearable. Only time to time Blood transfusion and costly drug is given for the increasing of life span. Treatment cost of every 30kg child need 4 lac taka every year. If 2 bag blood need every patient in every month, 1 lac 20 thousand bag blood will be need every month in Bangladesh.