Abstracts

508

REGIONAL CARDIAC NETWORKS: CAN THEY WORK?

doi:10.1136/archdischild-2012-302724.0508

1NK Puppala, R Phatak, K Atasanov, K Parkins, S Ali. North West & North Wales Paediatric Transport Service, Central Manchester University Hospitals NHS Foundation Trust, Warrington; North West & North UK Paediatric Transport Service, Alder Hey Children's Hospital, Liverpool, Wales

Background Only eleven paediatric intensive care units (PICUs) in UK have cardiac surgical services on-site. Small proportion of patients admitted to non-cardiac surgical PICUs require transport to surgical centres. North West and North Wales Paediatric Transport Service (NWTS) provides PIC transport service, including ability to conference call specialists. Alder Hey Children’s hospital (AHCH) is the cardiac surgical centre and Royal Manchester Children’s hospital (RMACH) only has cardiology on site.

Methods Retrospective review suspected cardiac cases transferred in 12 months- assessing if time critical surgical patients underwent single transfer to surgical centre- interventions by referring hospital or NWTS.

Results Total 29 patients of suspected cardiac diagnosis.

70

510

A NOVEL MYH7 VARIANT IDENTIFIED IN A CHILD WITH RESTRICTIVE CARDIOMYOPATHY

doi:10.1136/archdischild-2012-302724.0510

C Peers. Paediatric Cardiology, Bristol Children’s Hospital, Bristol, UK

Restrictive cardiomyopathy (RCM) is very rare in children and usually associated with a poor prognosis. Identification and thorough understanding of genetic causes is important for appropriate counseling and treatment. At Bristol Children’s Hospital, a one year old boy presented with progressive heart failure. Genetic testing revealed a novel c.510+1G>A mutation in the MYH7 gene. This variant has not been previously reported and is not known to cause disease. Further research is needed to determine the clinical significance of this variant.
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 and 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

doi:10.1136/archdischild-2012-302724.0511

Ö Özdemir, G Direk, A Altunkara. Pediatrics, İstanbul Medeniyet University, Gaziştepe Research and Training Hospital, Kadıköy, Turkey

**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, dyspepsia 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.

### 513 HEERFORDT’S SYNDROM IN AN ADOLESCENT BOY

doi:10.1136/archdischild-2012-302724.0513

1ê Ankan, 1ê Özkan, 1ê İşık Ağırs, 1ê Zengin, 1ê Çağatay, 1ê Dallar Bilge. ‘Pediatrics; 2Pediatric Nephrology, Ministry of Health, Ankara Training and Education Hospital, Ankara, Turkey

**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, vomiting 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 larcimal 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

doi:10.1136/archdischild-2012-302724.0514

1ê Sfaihi, 1ê S Kmiha, 1ê Maaaloul, 1ê Ben Mustapha, 2ê Barbouch, 1ê Hachicha. ‘CHU Hedi Chaker, Sfax; 2Institut Pasteur, Tunis, Tunisia

**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 hepatitisplenomegaly, 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...