G19(P) GENETIC ANOMALIES IN INFANTS WITH CONGENITAL HEART DEFECTS: A RETROSPECTIVE OBSERVATIONAL ANALYSIS

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Background and Aims Congenital heart defects (CHD) are the most common group of congenital malformation. With advanced testing - such as microarray and Next Generation Sequencing (NGS) - applied into routine clinical practice, more genetic anomalies may be detected in infants with CHDs.

Aims and Objectives The study was aimed to investigate the association between the congenital heart defects and genetic anomalies.

Methods A retrospective observational study included all the infants with a recoded new diagnosis of born CHD between 01/01/2017 and 31/12/2018, from a single tertiary neonatal intensive care unit. The data were recorded from the electronic patient records. For analytic purposes, CHDs cases were divided into 4 categories: critical, serious, significant or non-significant types.

Results 229 infants were identified, of which 8 were excluded due either to a lack of follow up or death from non-cardiac causes, which made classification impossible.

93% of infants with critical CHD, and 85% of those with serious CHD, had relevant genetic testing, of which 31% and 41% had positive diagnoses, respectively. Significantly more infants with critical or serious CHD underwent relevant genetic testing than the infants with significant (50%) or non-significant (12%) CHD (*p-value* <0.01).

Abstract G19(P) Table 1 The distribution of severity of CHD within the patient cohort and number of associated genetic tests

Classification	Number of Infants	Number of Infants who had Genetic Testing	Positive Genetic Test (%)
Critical	14	13	31
Serious	26	22	41
Significant	38	19	58
Non-Significant	143	17	35
Total	221	71	42

Of the 24 infants with critical, serious or significant CHD diagnosed with a CHD related genetic anomaly: 8 were diagnosed with Trisomy 21; 3 with Noonan's syndrome; 1 with DiGeorge and the remainder with various microdeletions, microdeletions and single nucleotide variants.

Conclusions In our cohort, around one-third of the infants with critical and serious heart conditions had underlying genetic defects. The infants with critical and serious heart condition needing surgery or intervention within one year after birth are often tested for the underlying genetic defect. The uptake of genetic testing was far less in infants with significant or minor CHDs.

G20(P) CARDIAC SCREENING IN CHILDREN PLANNED FOR STIMULANT MEDICATIONS: IS IT NECESSARY?

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Introduction ADHD (Attention Deficit Hyperactivity Disorder) affects 5–8% of children and adolescents. Stimulant medications have been proven to be an effective management strategy in many of these children. Children being considered for commencement of these stimulants are sometimes referred for cardiac screening to predict and prevent sudden death. NICE (National Institute of Clinical Excellence) has a set of criteria when cardiac screening is advised prior to commencement of treatment. However there is not much scientific data to suggest an increased risk in these children with use of stimulant medications.

Aim To look at the appropriateness of referral, investigations and outcomes of children referred to the PEC (Paediatrician with cardiac expertise) clinic for cardiac screening prior to commencement of ADHD medications.

Methods Retrospective analysis of referrals and consultation outcomes from Jan 2018 to Jan 2019.

Results 17 cases were referred by the community paediatricians to the PEC clinic. Of these 17 cases, 16 were boys and 1 was a girl. The age range was between 7–15 years with the average age being 11 years.

6 of the 17 cases were inappropriate and were not complying to NICE criteria, with the main reason being second degree relative having history of sudden deaths or pacemaker. The rest of the 11 cases were appropriate with the reasons being note of murmur, chest pains, palpitations & history of sudden death in 1st degree relative.

All 17 patients had ECGs (Electrocardiogram) and echocardiography which were all reported as normal. 5 patients also had ambulatory ECG monitoring which were also normal. All 17 patients were discharged from the clinic with no cardiac contraindications found for start of stimulant medications.

Discussion Our study found that many of these referrals are inappropriate. Cardiac screening tests were normal in all patients. Many of these patients waited between 3 to 6 months to be screened in these clinics. The conclusion from this study is that delaying ADHD medications in these children is not warranted as there is substantial evidence regarding the safety and efficacy of these medications.

G21(P) EBSTEIN'S ANOMALY IN THE NEONATAL PERIOD IS ASSOCIATED WITH VERY POOR PROGNOSIS – A 10 YEAR EXPERIENCE

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Introduction Ebstein's anomaly is a rare form of congenital heart disease (CHD), occurring in less than 1% of patients with CHD. With advances in Fetal Medicine and Cardiology, Ebstein's anomaly is being diagnosed antenatally. The literature highlights that it can be difficult to predict the prognosis particularly during the neonatal period.

Aims Our aim was to review the neonatal outcomes of babies with Ebstein's anomaly admitted to our tertiary neonatal intensive care unit (NICU).

Method We retrospectively identified all babies admitted to the NICU with a documented diagnosis of Ebstein's anomaly between 01/04/2009 and 31/04/2019. Their outcome data was collected using electronic patient records (BadgerNet).

Results In the last 10 years, 11 babies had a documented diagnosis of Ebstein's anomaly. Ten of these were antenatally diagnosed. Four were born prematurely (<37 weeks). Eight babies required resuscitation at birth and nine babies required intubation within the first 24 hours of life. Many of the babies had other significant co-morbidities including: hydrops fetalis, seizure activity, ventriculomegaly, trisomy 21 and persistent pulmonary hypertension (PPHN). Two babies also underwent therapeutic hypothermia. All of the babies born prematurely died within the neonatal period. Of the seven term babies with Ebstein's anomaly, three were discharged home, two were transferred to the Regional Paediatric Cardiology unit and two died on the neonatal unit. The two babies that were transferred the Regional Paediatric Cardiology unit subsequently died. The three surviving infants continue to be followed up. One of these patients is now seven years old and has not required any surgical intervention.

Conclusion In our experience, Ebstein's anomaly was uniformly fatal in babies born prematurely. Furthermore, in term babies the survival was 43%. The overall survival of Ebstein's anomaly presenting in the neonatal period in our cohort was 27%. Although this is an outcome of a small cohort of neonates with a rare congenital heart disease, it is important that parents are counselled appropriately antenatally so that they are adequately prepared for the potentially poor outlook of babies born with Ebstein's anomaly in the neonatal period.

G23(P) CHILD WITH HYPERTROPHIC CARDIOMYOPATHY DUE TO PRIMARY CARNITINE DEFICIENCY (PCD)

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Introduction Primary carnitine deficiency (PCD) is a rare autosomal recessive disorder of fatty acid oxidation, in which the transport of carnitine into cells is impaired. Carnitine deficiency blocks the oxidation of long-chain fatty acids in the mitochondria that leads to heart and hepatic disease, myopathy, nonketotic hypoglycaemia, and neurological complications. **Case Report** 7 month old child presented with 3 day history of vomiting.

He was born at term to non-consanguineous parents. He had returned to the UK recently, after a 4 month stay in India. Whilst in India he had had 2 hospital admissions with vomiting; one of which required admission to PICU with hypoglycaemia and metabolic acidosis, diagnosed as gastroenteritis and the next was for UTI.

O/E: Baby was drowsy, with evidence of subcostal recession and hepatomegaly.

Started on antibiotics for possible sepsis, given saline bolus, and started on maintenance fluids. He later developed hypoglycaemia, metabolic acidosis, and fluctuating consciousness. After a discussion with the metabolic team, he was intubated and transferred to PICU.

Investigations Ammonia levels increased from 164 to 340 micromol/L in 6 hours, high ALT and deranged clotting. Free carnitine <1 umol/l.

Initial ECHO showed mild left ventricular hypertrophy. He had cardiopulmonary arrest on day 3 of PICU admission and consequently needed ECMO for 4 days. He was noted to have hypertrophic cardiomyopathy with left ventricular dysfunction. He required multiple inotropes and nitric oxide

and also developed intracranial bleeding and subsequent seizures, requiring Levetiracetam. Other complications included AKI (required CVVH), liver failure, MRSA Bacteremia and E-Coli septicemia, which were treated.

Significant improvement was noted following levocarnitine replacement- he made a good recovery and was discharged home.

Discussion Primary carnitine deficiency (PCD) usually presents with cardiomyopathy and muscle weakness and is one of the treatable causes of cardiomyopathy.

Treatment of this disorder with L-carnitine is highly effective in correcting cardiomyopathy and muscle weakness.

Child may be asymptomatic until they develop signs of cardiomyopathy and congestive heart failure. Cardiac function does not respond to inotropes or diuretics. If no carnitine is supplemented, progressive heart failure eventually leads to death.

G23(P) A 10 YEAR EXPERIENCE OF NEONATES WITH TRUNCUS ARTERIOSUS

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Background Congenital heart defect affects around 10 in a 1000 live born babies. Truncus arteriosus is a rare conotruncal defect in which a common arterial trunk supplies systemic, pulmonary and coronary circulation. The incidence of truncus arteriosus is quoted between 1–2% of all congenital heart malformations.

Aim To evaluate the initial neonatal management and outcomes of newborns with truncus arteriosus.

Method We conducted a retrospective review of all newborns with truncus arteriosus admitted to our neonatal unit between 01/04/2009 and 31/03/2019. Data were collected from Badger and Cardiology databases to evaluate the initial neonatal management and long-term outcomes of this cohort of patients.

Results Sixteen newborns with truncus arteriosus were admitted to our neonatal unit in the last 10 years. Their mean gestational age was 36 weeks with a mean birth weight of 2407 grams. Cardiac defects were detected prenatally in 13 patients (82%). Three babies were diagnosed postnatally. One following failed pulse oximetry screening requiring non-invasive respiratory support and 2 diagnosed following on-going respiratory support on echocardiography. Respiratory support was initiated within the first 24 hours of life in 8 patients; these included all patients with a postnatal diagnosis of truncus arteriosus. Four patients required mechanical ventilation. Whilst in the neonatal intensive care unit, 3 babies developed heart failure and were treated with diuretics and 2 developed necrotising enterocolitis which was managed conservatively. Median length of stay before transfer to our local surgical cardiology centre was 3 days. Fourteen patients (87%) underwent common arterial trunk repair. 11 (78%) babies survived to date. Two patients died in the initial post-operative period and one patient at 14 months of age. The remaining two patients did not receive any surgical intervention and subsequently died (4 and 11 days old).

Conclusion Truncus arteriosus is a rare conotruncal defect with the majority being detected antenatally in our cohort. They