GP74 VACTERL ASSOCIATION: FROM DIAGNOSTIC TO TREATMENT THROUGH A SERIES OF PITFALL

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Background VACTERL association is a disorder that affects many body systems. VACTERL stands for vertebral defects, anal atresia, cardiac defects, tracheoesophageal fistula, renal anomalies, and limb abnormalities. People diagnosed with VACTERL association typically have at least three of these characteristic features. Other features may include (less frequently) growth deficiencies and failure to thrive; facial asymmetry (hemifacial microsomia); external ear malformations; intestinal malrotation; and genital anomalies. The objective of this case report is to highlight the difficulty of diagnosis and possible complication in management of complex malformation.

Case presentation We present the case of a 10-weeks-old infant from twin pregnancy, born prematurely with C-section at 35 weeks of gestation, with 1890 g. There was no family history of congenital anomalies. His brother was diagnosed after birth with esophageal atresia with tracheoesophageal fistula but died on the second day of life after severe complication during surgery. Our infant with facial asymmetry and breathing problems was investigated in Pediatric ICU for poor feeding, excessive drooling of saliva, fast breathing, cough and intermittent bowel movement disorders (excessive bloating with constipation alternatively with watery stools). Based on the clinical and laboratory findings, a series of imagistic and invasive investigations (including bronchoscopy) they confirmed atrial septal defect, maintain the suspicion of tracheoesophageal fistula, but excluded Pierre-Robin syndrome and other chromosomal disorders, cystic fibrosis, GERD, congenital infections. Because of failure to thrive he was feeding with nasogastric tube. Due to bowel movement disorders he was suspected of Hirschsprung's disease. During surgical intervention the suspected tracheoesophageal fistula was identified and resolved; also, colostomy for segmental bowel resection has been done. The histologic examination excludes congenital aganglionic megacolon, and the anal/rectal stenosis was attributed to VACTERL association. The clinical evolution was favourable with normal weight gain, but the infant underwent two more surgery (removal of the esophageal patch, due to an esophageal stenosis and stoma reversal surgery).

Discussion This case illustrates even if the strict protocol for investigations are followed the clinician experience must remain the most important diagnostic clue. Despite a wide range of diagnostic options for VACTERL described in the literature the final diagnostic tool might be surgical exploration.

GP75 CASE OF A LIFETIME – HOW MOSAICISM MAY MISLEAD

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A patient was referred in consequence of an abnormal microarray finding. The background history was of Left Congenital Diaphragmatic hernia, coarctation of Aorta and PDA. Examined at 5 weeks by Consultant Clinical Geneticist, she was non-dysmorphic, her head circumference was on 25th percentile. Neurological examination was age appropriate.

ACGH showed a mosaic chromosomal imbalance involving chromosome 2q in the form of a ~46.7Mb gain of 2q26.1–q31.2 and a ~17.6Mb loss of 2q36.2–qter, which was estimated to be present in approximate 50% of cells. In contrast G-band karyotype analysis of phytohaemagglutinin stimulated and cultured cells showed no evidence of the abnormal cell line, with only apparently normal female 46, XX metaphases seen. So, there was a data mismatch.

Parental karyotypes were both normal. Due to the peripheral blood karyotype result a skin biopsy was taken for culture and karyotyping, together with a buccal smear for FISH analysis. In addition, a second peripheral blood sample was taken to allow FISH analysis on non-cultured cells. These FISH analyses indicated the presence of an abnormal cell line in 19% (buccal smear) and 35% (whole blood) of the 200 cells analysed. Conversely cultured fibroblasts only resulted in cells with a normal female karyotype. Hence, these results indicate that the level of mosaicism varies dramatically between different cell lineages.

The absence of the abnormal cell line in the cultured cells would support that this cell line is not present in the T-lymphocytes. A FBC and film showed normal white cell counts and morphology. The findings with the cultured fibroblasts may be reflective of either the absence or scarcity of the abnormal cell line in the biopsy.

This most unusual case illustrates that the understanding of how test results are generated and potential limitations of each test is crucial when considering the clinical features of the patient. Further it demonstrates how mosaicism may be unequally distributed between cell types or indeed absent in the actual cell type that is being tested. It is reassuring that the patient is neurologically age appropriate. The management with clinical and developmental observation are warranted to monitor her progress.

GP76 EXPERIENCE IN TREATMENT OF BLADDER EXSTROPHY IN NEWBORN GIRL WITH APERT SYNDROME

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Background and aims Prevalence of bladder exstrophy (BE) is 1 per 30–50 thousand newborns and it is observed in boys 2–6 times more often than in girls. This pathology demands reconstructive surgery at early age. We are presenting the unique clinical case of bladder exstrophy at the girl with the Apert syndrome (its prevalence is 1 per 100–160 thousand newborns) confirmed with molecular genetic testing.

Research is focused on identification of appropriate surgical intervention taking into account anomaly form and anatomic features and child general medical condition.

Methods Case history: the child from intact pregnancy and delivery. Multiple congenital anomalies (MCAs) haven't been

revealed prenatally. Girl was born on 27.05.2018. At birth the diagnosis of BE and multiple dysembryogenic stigmas was made. The girl was transferred to the NMRCCH in an urgent order at first 24 hours of life.

On presentation: BE, total epispadia, divergent labia minora and majora, pubic bones diastasis (up to 4.6 cm). There was a defect on anterior abdominal wall wherethrough bladder mucous membrane was prolapsing. Anus was normal. External sex organs were abnormal, child gender couldn't be identified adequately. The uterus was prolapsing due to anxiety at first 24 hours of life. Acrocephaly, frontal bone persistence, fingers and toes complete syndactyly, uranostaphyloschisis and choanal atresia came under notice.

Results We have performed primary repair of bladder with local flaps on 5th day. Later we performed rehabilitation measures.

Due to molecular genetic testing we revealed heterozygous mutation *c.755C>G (p.Ser252Trp)* in *FGFR2* gene (Apert syndrome, autosomal dominant disorder).

Long-term outcomes were estimated at six months age. The child is tube fed, puts on weight and develops. Urinary system infections took place twice due to vesicoureteral reflux. Note that bladder voiding and storage functions are adequate. Bladder volume is 60 ml and the interval between urinations is about 1 hour. Following step is to perform neurosurgical treatment. Orthopedic surgery should be performed in older age.

Conclusion Bladder exstrophy treatment in newborns even comorbid with other MCAs is optimal and shall be carried out in referral centres. The choice and validation of surgery method for BE treatment demand personalized approach. Multidisciplinary approach in management of these patients is important in the presence of other MCAs.

GP77 OCULOCUTANEOUS ALBINISM IN A NEONATE

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Background Oculocutaneous albinism (OCA) is a genetically heterogeneous group of disorders affecting melanin biosynthesis. It is always an autosomal recessive inheritance and is characterized by decreased or absent pigmentation of hair, skin, iris and retina. Clinical presentation is typically of an infant who does not appear to see at the expected 6–8 week corrected age, often with nystagmus and pale pigmentation. Clinical findings demonstrate grossly obvious iris attenuation and iris transillumination, refractory errors and photophobia. Investigations include genetic testing, Optical Coherence Tomography (OCT) to identify foveal hypoplasia and electrophysiology to establish any excessive crossing of optic nerve fibres at the optic chiasma.

Method A Caucasian female, born to non-consanguineous parents was referred to Paediatric services at one week of age with concerns regarding her pale hair, eyelashes and diffuse pink reflex from both eyes. On examination she had marked iris transillumination without nystagmus and significant hypermetropic astigmatism for which glasses were prescribed. Her father had iris transillumination and nystagmus but met the visual standards to drive. By eight weeks, the infant had

developed nystagmus and was showing some visual awareness. Developmental examination was age appropriate.

Results At initial presentation the expectation of delayed visual maturation was explained to the parents with reassurance that once vision has matured, OCA is a stable condition. Advice was given regarding protection from solar damage as well as the importance of regular skins checks in view of the possible increased risk of skin cancers. The family history and the phenotypic variability between OCA subtypes prompted gene testing to arrive at a molecular diagnosis.

Conclusion The case highlights the importance of early identification so families are aware of the natural history of OCA as well as the need to protect skin from UV damage. Carrier detection and personalised genetic counselling are possible once the disease causing mutations in the family are identified.

GP78

DILATED CARDIOMYOPATHY WITH ROSS III HEART FAILURE INDUCED BY SEVERE IRON DEFICIENCY ANAEMIA POST LONG TERM COW MILK EXCESS

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Aim To present a 1 year and 6 months old girl and 8.2 kg, admitted in the Emergency Department for extremely pale skin, loss of appetite, intense fatigue, muscular hypotonia, tachycardia, polypnea, grade II systolic murmur, hepatomegaly and Ross III heart failure. From the age of 7 months she started to select the food and finally ate only 1 liter/day cow milk. One day before admittance she was not able to wake up from her bed.

Material and Methods The patient performed lab investigations, cardiopulmonary X-ray, cardiac examination and hematology exploration.

Results Cardiomegaly with increased vascular markings was found on X-ray. ECG revealed tachycardia 148 b/min, alternating with bradycardia, 48 b/min, right atrial dilatation, left ventricle hypertrophy and prolonged QTc 0.45 sec. Echocardiography showed dilated cardiomyopathy, tele diastolic diameter 4.02 cm, EF 0.56, SF 29%, grade III mitral regurgitation, grade I aortic regurgitation, minimal pericardial fluid and slight pulmonary hypertension. Cardiac biomarkers as NT pro-BNP was very high, 8 817 pg/mL, with elevated CK MB 2.51 ng/mL and normal Troponin T. Serology for Parvovirus B19 IgM and Coxsackie was negative, excluding the viral infection as etiology for dilated cardiomyopathy. Severe anemia with 1.9 g/dl Hb was detected, that imposed rapid blood transfusion. The Iron level, ferritin was 0.1 ng/ml, extremely low. Seriated blood transfusions were necessary until Hb achieved 10 mg/dl. Concomitant heart failure treatment was started with Furosemide, Spironolactone, Captopril and low dose of Dobutamine. Iron correction was done with oral products. Folic Acid and Vit C was added in treatment. The evolution was slowly good; gradually normal diet was introduced with meat, eggs, vegetables and fruits.

Conclusions Anemia is cited in the literature to induce dilated cardiomyopathy. Our patient had the most decreased reported value of Hb and Iron, due to long duration of exclusive cow milk daily alimentation. This situation