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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.

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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.

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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