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Case Report

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A CASE REPORT OF VACTERAL ASSOCIATION

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ABSTRACT

VACTERL association is a acronym used for a condition characterised by the sporadic, non-random association of specific birth defects of multiple organ systems. We present one such case which had congenital abnormalities of renal, skeletal and cardiac system.

KEYWORDS: VACTERL association, radial hand deformity.

INTRODUCTION

VACTERL association is a acronym used for a condition characterised by the sporadic, non-random association of multiple congenital anomalies.[1,2]We report one such case which presented with absent thumb of both hands and weakness of all limbs since birth and delayed milestones but on further work-up was found to have anomalies of vertebrae, kidneys and cardiac and hence was labelled as a case of VACTERL association

Case Report: A nine months old male baby was brought to us with deformity of left upper limb with absent thumb noticed at birth. The baby was born third order birth to a non consanguineous parents with no family history of any congenital anomalies. He was born by full term vaginal delivery following an uneventful pregnancy with no significant past medical or surgical history. Physical examination of the baby revealed deformity of both limbs with absent thumb of both hands and small rudimentary thumb seen on left side other findings included low set ears and head lag present. (Fig. 1) **Radiographs** revealed absence of right thumb and metacarpal bone with absent left thumb and metacarpal, radius is shortened on both side(Fig. 2). On further evaluation and imaging, abnormalities involving other systems were detected. Chest radiogram (Fig. 3) showed features of cardiomegaly (cardiothoracic ratio greater than 50%) hemivertebra seen at level D7 and delayed ossification centre of left humeral head and infantogram revealed pelvic bone appears normal with delayed appearance of bilateral femoral head ossification centre and right foot shows shortening of great toe with normal phalanges with abduction deformity between great toe and fourth toe. Echocardiogram revealed moderate perimembranous ventricular septal defect 7mm with left to right shunt. Ultra sonogram of abdomen found both kidneys to be fused across the midline in keeping with horseshoe kidney with parenchymal isthmus and no obvious hydronephrotic changes. Hemoglobin, total and differential leucocytes count, platelet count, and renal function tests were all within normal limits. Chromosomal analysis did not reveal any abnormality. Due to simultaneous occurrence of congenital anomalies involving cardiac, skeletal (vertebrae, limb) and renal system, the patient was labelled as a case of VACTERL association. The patient was referred to higher centre as per attender s will.

fig 1

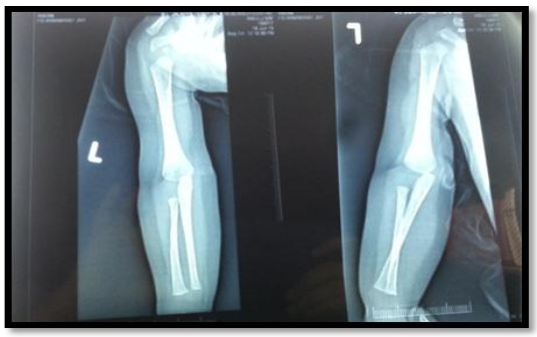

fig 2

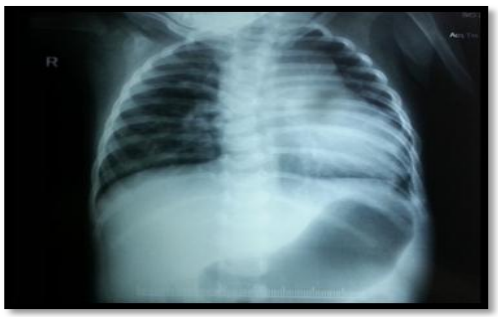

fig 3

DISCUSSION: VACTERL association is a mnemonically useful acronym for a condition characterised by the sporadic, non-random association of specific birth defects in structures derived from the embryonic mesoderm.Each letter in VACTERL represents the first letter of one of the more common findings seen in affected cases. VACTERL association was first reported by Corcora et al. in 1975,[1] but only 1.0% of such cases present the full range of anomalies.[2]. For getting labelled as VACTERL ,there should be atleast three out of the following seven findings.[3]

• V - Vertebral anomalies: Vertebral anomalies usually consist of hypoplastic(small) vertebrae or hemivertebra (where only one half of the bone is formed). About 70 percent of patients have vertebral anomalies. Later in life, these vertebral anomalies may put the child at risk for developing scoliosis. • A - Anal atresia: Anal atresia or imperforate anus is seen in about 55 percent of patients. • C - Cardiovascular anomalies: Up to three-quarters of patients with VACTERL association have been reported to have congenital heart disease. The common heart defects seen are ventricular septal defects, atrial septal defects and Tetralogy of Fallot. Less common defects are truncus arteriosus and transposition of the great arteries. • T-E - Tracheoesophageal fistula: Esophageal atresia with tracheo-esophageal fistula (TE fistula) is seen in about 70 percent of patients. • R - Renal (Kidney): Renal defects are seen in half the patients with malformation of one or both kidneys or obstructive uropathy. • L- Limb defects: Limb defects seen in up 70 percent of babies include absent or displaced thumbs, polydactyly, syndactyly and forearm (includng radial aplasia) and leg defects. In addition, affected children may also exhibit less frequent abnormalities including growth deficiencies and failure to thrive.

VACTERL in lower frequency.[4] like facial asymmetry (hemifacial microsomia), external ear malformations, lung lobation defects, intestinal malrotation and genital anomalies. VACTERL shows some pheonotypic overlap with many other conditions including Feingold syndrome, CHARGE syndrome, 22 qll deletion syndrome. Townes-Brocks syndrome, Pallister-Hallsyndrome, Fanconi anemia spectrum, Goldenhar Syndrome, Nager syndrome, caudal regression syndrome, sirenomelia, electrodactyly-ectodermal dysplasia syndrome, Jarcho-Levin syndrome and Klippel- Fiel syndrome. The birth prevalence varies from 1:3,500 to 1.6:10,000, more incidence seen in infants born to diabetic mothers.[3] and is rarely seen more than once in one family. The reason it is called an association, rather than a syndrome is that while all of the birth defects are linked, it is still definitely unknown which genes or sets of genes cause these birth defects to occur. A disruption in differentiating mesoderm in first 4-5 weeks has been suggested to be the basis for such a non-random association. Besides recent research has shown that VACTERL could be caused by defective Shh (Sonic hedgehog pathway) signalling during human embryogenesis.[5] Reports have also suggested that the VACTERL association may possibly occur with increased frequency in children of mothers have taken the cholesterol-lowering statin drugs in the first trimester of pregnancy.[6] Prognosis is overall poor and depends upon the extent and combination of deformities and the quality of availability of a multidisciplinary approach. If detected in utero during anomaly scans before viability, termination of pregnancy can be offered.

REFERENCES

1. Corcoran R, Entwistle GD. Letter: polycystic ovarian disease and oral contraception. Lancet, 1975; 21:1386–87.
2. Khoury MJ, Cordero JF, Greenberg F, James LM, Erickson JD. A population study of the VACTERL association: evidence for its etiologic heterogeneity. Pediatrics, 1983; 71: 815–20. 3. Harjai MM, Holla RG, Kale CR. Full spectrum of VACTERL in new born MJAFI, 2008; 64: 84-85.
3. Kallen K, Mastroiacovo P, Castilla EE, Robert E, Kallen B. VATER non-random association of congenital malformations: Study based on data from four malformation registers. Am J Med Genet, 2001; 101: 26-32.
4. Botto LD, Khoury MJ, Mastroiacovo P , Castilla EE, Moore CA, Skjaerven R et al. The spectrum of congenital anomalies of the VATER association: An international study. Am J Med Genet, 1997; 71: 08-15.
5. Arsic D, Qi BQ, Beasley SW. Hedgehog in the human: a possible explanation for the VATER association. J Paediatr Child Health, 2002; 38(2):117-21.