



### **Case Report:**

#### **VACTERL association**

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#### **Abstract:**

VACTERL association is a useful acronym 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.

**Key Words:** VACTERL association, radial club hand

#### **Introduction:**

VACTERL association is a mnemonically useful acronym for a condition characterised by the sporadic, non-random association of multiple congenital anomalies (1,2). We report one such case which presented with left radial club hand 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 four months old male child was brought to us with deformity of left upper limb noticed at birth. He was born at term by normal delivery following an uneventful pregnancy and there was no significant past medical or surgical history. He was the first child of non-consanguineous parents and there was no family history of congenital anomalies. Physical examination of the baby revealed deformity of left forearm which appeared shorter than contra lateral side with radial deviation at the wrist (Fig. 1). All the digits of the affected hand excluding the thumb were normal; the thumb was however rudimentary. Radiographs revealed features Type 4 left radial club hand (Fig. 2) which included absent left radius with hypoplastic bowed ulna and only four metacarpals corresponding to normal fingers. The thumb appeared as accessory soft tissue density on the radial aspect of the left hand. On further evaluation and imaging, abnormalities involving other systems were detected. Skeletal survey also showed a mild form of vertebral segmentation abnormality with fusion of the posterior elements of C2-C3 (Fig. 3), narrowing of the disc space and partial fusion of the bodies of L1-L2 (Fig. 4). Chest radiogram (Fig. 5) showed features of cardiomegaly (cardiothoracic ratio greater than 50%) and echocardiogram revealed moderate perimembranous ventricular septal defect with right pulmonary vein stenosis and pulmonary hypertension. 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. (Fig. 6) Hemoglobin, total

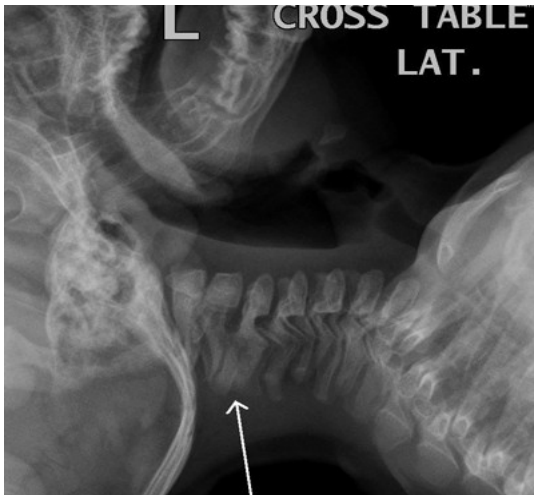
and differential leucocytes count, platelet count, and renal function tests were all within normal limits in more than one occasion. Chromosomal analysis did not reveal any abnormality. Due to simultaneous occurrence of congenital anomalies involving cardiac, skeletal (vertebrae, limb) and renal system, the patient was labelled as a case of VACTERL association. The patient was planned to undergo operative centralization of the wrist on ulna after surgical correction of cardiac defects by pediatric cardiac surgical team.



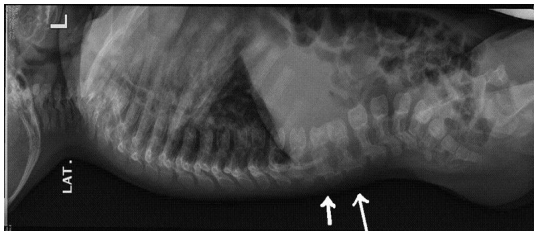
**Fig. 1: Left radial Club hand**



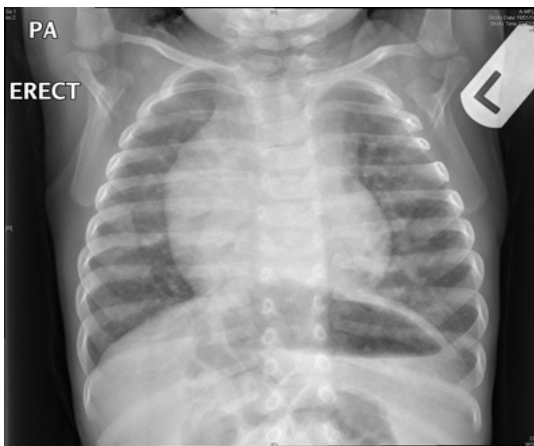
**Fig. 2: Plain radiogram of left upper limb showing Type 4 radial club hand and rudimentary thumb.**



**Fig. 3: Cervical spine radiogram (lateral view) showing fusion of the posterior elements of C2-C3 (white arrow)**



**Fig. 4: Thoracolumbosacral spine radiogram (lateral view) showing narrowing of the disc space and partial fusion of the bodies of L1-L2 (white arrows)**



**Fig. 5: Plain chest radiogram (PA view) showed features of cardiomegaly (cardiothoracic ratio greater than 50%)**



**Fig. 6: Abdominal ultrasonogram showing uncomplicated horseshoe kidney with parenchymal isthmus**

#### **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 with VACTERL association will have vertebral anomalies. In early life these anomalies rarely cause any difficulties, although the presence of these defects on a chest x-ray may alert the physician to other defects associated with VACTERL. 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 with VACTERL association.
- **C - Cardiovascular anomalies:** Up to three-quarters of patients with VACTERL association have been reported to have congenital heart disease. The most common heart defects seen with VACTERL association 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 with VACTERL association.
- **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 to 70 percent of babies include absent or displaced thumbs, polydactyly, syndactyly and forearm (including radial aplasia) and leg defects.

In addition, to the above mentioned features, affected children may also exhibit less frequent abnormalities including growth deficiencies and failure to gain weight and grow at the expected rate (failure to thrive). Furthermore, defects of practically every organ system have been reported in association with

VACTERL in lower frequency (4) like facial asymmetry (hemifacial microsomia), external ear malformations, lung lobation defects, intestinal malrotation and genital anomalies.- VACTERL shows some phenotypic overlap with many other conditions including Feingold syndrome, CHARGE syndrome, 22q11 deletion syndrome. Townes-Brocks syndrome, Pallister-Hall syndrome, Fanconi anemia spectrum, Goldenhar Syndrome, Nager syndrome, caudal regression syndrome, sirenomelia, ectrodactyly-ectodermal dysplasia syndrome, Jarcho-Levin syndrome and Klippel- Fiel syndrome. Some researchers have added an (S) to the VACTERL acronym to represent a single umbilical artery instead of the normal two. Mental functioning and intelligence is usually unaffected; developmental delay/mental retardation should suggest an alternative diagnosis.

VACTERL is seen more frequently in infants born to diabetic mothers. The birth prevalence varies from 1:3,500 to 1:6:10,000 (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.(5) Besides recent research has shown that VACTERL could be caused by defective Shh (Sonic hedgehog pathway) signaling during human embryogenesis. (6) Some reports have also suggested that the VACTERL association may possibly occur with increased frequency in children whose mothers have taken the cholesterol-lowering statin drugs in the first trimester of pregnancy.

Prognosis is overall poor and depends upon the extent and combination of deformities and the quality of available health-care. If detected in utero (by sonography) before viability, termination of pregnancy can be offered.

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