

# Solitary Kidney with VACTERL-H Association in Children - A Rare Case Report

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**Abstract:** VACTERL-H association is an extremely rare disorder. It includes vertebral anomaly, anal atresia, cardiac defects, tracheoesophageal fistula, renal defects, limbs defects and hydrocephalus. It is diagnosed mainly by clinical examination and a few specialized tests. Multidisciplinary management is required for these cases with staged surgical procedures.

**Keywords:** Anomaly, Clinical examination, Hydrocephalus, Solitary kidney, VACTERL

## 1. Introduction

VACTERL association is a useful acronym for a specific birth defect. It is defined by the presence of at least three of the following congenital malformations:-Vertebral defects, Anal atresia, Cardiac defects, Tracheoesophageal fistula, Renal anomalies, and Limb defects. Incidence is estimated approximately 1 in 10000 to 1 in 40000 live births [1]. VACTERL with hydrocephalus is an extremely rare disorder that affects males and female's child equally [2]. We are reporting a case of female child with VACTERL-H association.

## 2. Case Report

A one-month-old female child presented in our OPD with passage of stool from abnormal site and swelling over back of scalp since birth. On clinical examination baby was active, wt.3.6kg and vital signs were within normal limits. Anterior fontanelle opened and bulged; head circumference was 37cm. A swelling of trilobed shape around 1 x 2 cm present over occipital region of head. Swelling was non transilluminated and non-pulsatile [Figure 1]. Perineal examination, suggestive of recto vestibular fistula [Figure 2]. In ultrasound KUB, there was non-visualisation of right kidney and cranium ultrasonography suggestive of dilated bilateral ventricles and 3rd ventricle with VHR-0.38. Ultrasonography of local swelling revealed bony defect of 11mm with cerebellar parenchyma herniating into lesion of size 23 X 12 mm. Echocardiography of the patient revealed patent foramen ovale. X ray LS Spine were normal. Non contrast CT scan of brain revealed gross asymmetrical dilation of bilateral lateral, third and fourth ventricle with suggestive of communicating hydrocephalus [Figure 3]. Renal scan (DMSA scan) confirmed non visualized non-functioning right kidney and left kidney has normal cortical function with no evidence of scar [Figure 4]. In our case due to simultaneous occurrence of congenital anomalies involving anal atresia, renal anomalies, cardiac defect, and with hydrocephalus the patient was labelled as VACTERL-H syndrome. Patient underwent staged surgical correction for recto vestibular fistula. Sigmoid colostomy was done.

We kept child on regular follow up. Our next planned is anorectoplasty and occipital encephalocele repair. Child on acetazolamide for hydrocephalus.



**Figure 1:** Clinical image of encephalocele



**Figure 2:** Showing recto vestibular fistula with absent anal opening

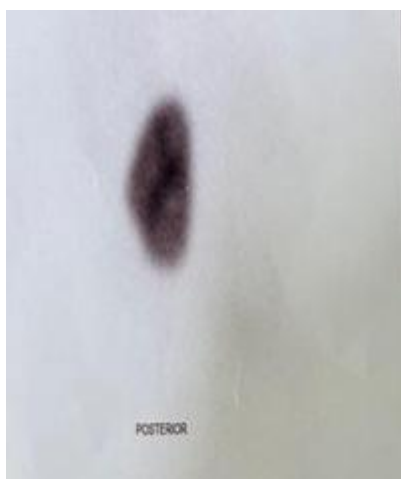
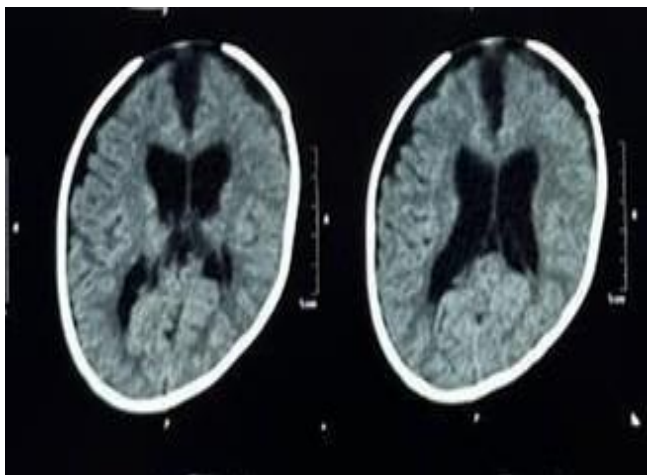


Figure 3: CT head showing dilated ventricles



Figure 4: Renal scan showing right renal agenesis

### 3. Discussion

VACTERL association was first reported by Corcora et al [3]. The full range of anomalies only occurs in 1 % of such cases. In VACTERL each letter represents the first letter of one of the more common findings seen in affected cases. Patient labelled as VACTERL, there should be at least three out of the following seven findings.

**V-Vertebral anomalies:** In vertebral anomalies usually hypoplastic vertebrae or hemivertebra is common. About 70 percent of patients with VACTERL association will have

vertebral anomalies.

**A-Anal atresia:** Anal atresia or imperforate anus is seen in about 55 percent of patients with VACTERL association.

**C-Cardiovascular anomalies:** The most common cardiac defects seen with VACTERL association are ventricular septal defects, atrial septal defects, and tetralogy of Fallot. Three quarters of patients with VACTERL association have been reported to have congenital heart disease.

**TE-Tracheoesophageal fistula:** Esophageal atresia with tracheo-esophageal fistula is seen in about 70 percent of patients with VACTERL association.

**R-Renal anomalies:** Renal agenesis of one or both kidneys or obstructive uropathy are common renal anomalies. It is seen in half of the patients.

**L-Limb defects:** Radial aplasia, polydactyly, syndactyly, leg defects and absent or displaced thumbs are common limb defects. It is seen in up to 70 percent of child.

VACTERL with hydrocephalus is an extremely rare disorder. It is probably inherited as an autosomal recessive trait. Genetic counselling of the family is needed. Fanconi anemia is a major association of VACTERL-H [4]. In addition, to the above-mentioned features, affected children may also exhibit less frequent abnormalities including growth deficiencies, failure to thrive, facial asymmetry (hemifacial microsomia), external ear malformations, lung lobation defects, intestinal malrotation and genital anomalies [5]. VACTERL abnormalities can present with some known chromosomal abnormalities, like trisomy 13, 18, and 5p-syndrome. Interstitial deletion of long arm of chromosome 6 (6q13-15) and long arm of chromosome 13 have been reported in few cases [6]. Diagnosis is mainly by clinically and is based on the phenotypic features. Exact cause of VACTERL association is unknown. Antenatally VACTERL association can be diagnosed through ultrasound or more sophisticated methods such as fetal echocardiogram or fetal MRI. In antenatal scan certain radiological features like polyhydramnios, absence of a gastric bubble, dilated colon, vertebral defects, and limb abnormalities that may suggest an affected fetus with VACTERL anomaly [7]. Management is multidisciplinary team work including pediatric surgeon, pediatricians, cardiologists, and orthopedic surgeons. Treatment is directed towards the specific symptoms that are present in child. The structural abnormalities such as anal atresia, radial defects, and cardiac defects require staged surgical corrections. Prognosis for children with this condition depends on the severity of anomalies.

### 4. Conclusion

VACTERL-H syndrome is very rare clinical entity. The diagnosis of VACTERL-H Syndrome is mainly upon clinical examination and a few specialized tests. Multidisciplinary management is required for these cases with staged surgical therapy. Early diagnosis and interventions are needed to prevent morbidity and mortality.

## References

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