Case Report

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ANATOMY

# VACTREL Association with Less Common Cardiac and Tracheal Defects - A Case Report

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## Abstract

VACTERL/VACTER Association is defined by presence of at least three of the malformations: vertebral defects, anal atresia, cardiac defects, tracheo- esophageal fistula, renal anomalies and limb abnormalities. The type of defects and incidence of occurrence of each defect in this association may vary. Vertebral, anal, esophageal defects are reported by many, but reported incidence of truncal defect and tracheal agenesis are less. Here is a case which fits in to this association presenting anal, renal, limb and cardiac defects. Less commonly occurring persistent truncaus arteriosus, tracheal agenesis, absence of fibula on right side instead of radial anomalies of upper limb are the interesting features in this case. Our attempt is to present this case and highlight these less common features.

***Key Words:*** **VACTERL association, Tracheal agenesis, Truncal defect, Aanal agenesis**

## Introduction

Developmental defects are a major cause for fetal loss. Most defective fetuses are aborted at an early date. The etiology of the defects may be genetic, environmental or multifactorial. Defects may be in the form of malformation which is seen due to defect in developmental process itself, or there may be syndromes which present with multiple related defects. Apart from them there are some cases which show abnormalities in systems that occur in association with each other. An example for such association is VACTERL /VACTER association. This was first named in the early 1970's and the condition included statistically non- random co- occurrence of a group of defects namely: vertebral defects, anal atresia, cardiac defects, tracheo-esophageal fistula, renal anomalies and limb abnormalities. Incidence of this Association is estimated at approximately 1 in 10,000 to 1 in 40,000 live born infants [1]. Many conditions fall under differential diagnosis for this defect. A few among those are– Baller-Gerold syndrome, CHARGE syndrome, Currarino syndrome, deletion22q11.2 syndrome, Fanconi anemia and MURCS association etc. [1]. It was proposed later to include vascular anomalies including Single Umbilical Artery (SUA) as part of V in VACTERL, cardiac malformations for C and additional limb anomalies for L [1]. It was also proposed to include tracheal agenesis as a component feature with VACTERL association [2]. Tracheal agenesis and truncal defect of heart are comparatively less common defects in this association.

Much discussion and debate is going on the causative factors of this defect [1,3,4,5,6,7].The role of genetics in causation of the defect is controversial. Since any genetic cause for this defect is not yet definitely established, the latest

diagnostic genetic investigations may offer a better basis for this disorder.

Accurate prenatal diagnosis and proper management of pregnancy, appropriate counseling for the parents,and early post natal surgical correction of these defects where ever possible will greatly help the affected child, its parents and ultimately society.

## Case report

A male fetus of 22weeks gestation, with a short and defective right lower limb was received in the department of Anatomy, NRI Medical College from Obstetric department of NRI General Hospital for study. There was neither history of maternal diabetes or hypertension nor family history of birth defects in the family

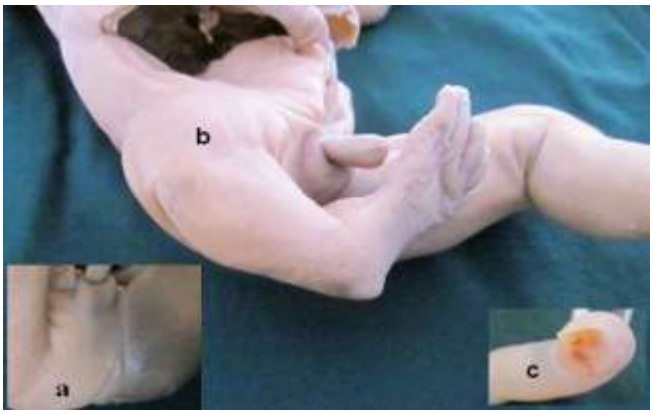
The study was carried out after ethical committee clearance and with a proper written consent from parents.

External and internal defects were recorded after proper fixation with formalin.

External examination revealed

a) Absence of anal orifice b) short and defective right lower limb c) single umbilical artery (Fig 1).

**Figure 1: Anal agenesis, limb defect and single umbilical artery**




Internal examination:

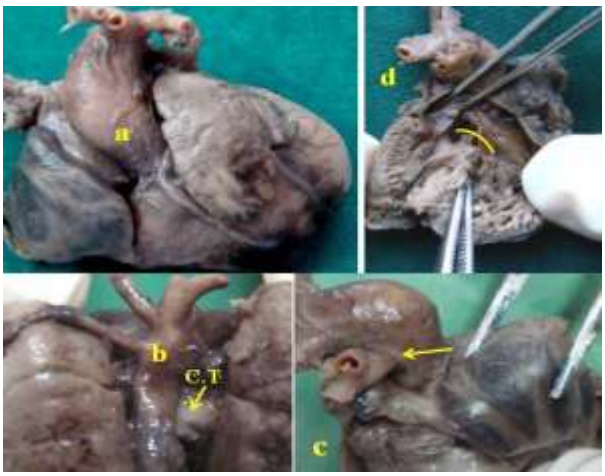
Vertebral Defects: No vertebral defects are seen

GIT: Distension of large intestine with meconium+; Rectum-moderately dilated

Anal atresia +. Imperforate anus [Fig.1a]

**Cardio vascular defects**

**Figure 2: Cardiac defects**



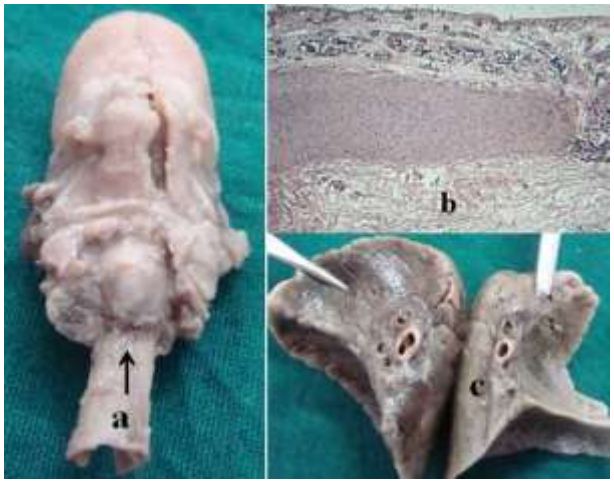

- Right ventricle was large. A single large dilated vessel originated from this chamber. [Fig.2a] This vessel, truncus arteriosus or truncal artery replaced the aorta and continued as aortic arch and descending thoracic aorta posterior to a horizontal cartilaginous tube (C.T) [Fig 2b].
- Branches originating from this vessel included: a single vessel (? Pulmonary trunk) from its left lateral aspect, brachiocephalic trunk, left common carotid and left subclavian arteries and two coronary arteries near its commencement
- Truncal orifice presented anterior, left and posterior cusps
- Right coronary orifice was related to left cusp and left coronary orifice was related to posterior cusp.

- A single pulmonary artery originated from left lateral aspect of truncal artery on a slightly posterior plane, divided immediately into right and left branches to be distributed to lungs [Fig.2 c]
- Left ventricle was small in size.
- Inter-ventricular septal defect was seen in upper part of septum [Fig 2d]
- Single umbilical artery was present in umbilical cord [Fig1c]

Tracheal and Laryngeal Defects

- Larynx terminated as a blind tube at lower border of cricoid cartilage (Fig 3a)
- Tracheal agenesis +
- A single horizontal cartilaginous tube (? a common bronchus to two lungs) was located anterior to descending aorta [Fig 2 b; C.T]and was entering the lungs on either side through their hila . Histological examination of this tube confirmed its structure as bronchus [3b].No fistulous connection between this tube and esophagus existed

**Figure 3: Tracheal and Laryngeal defects**



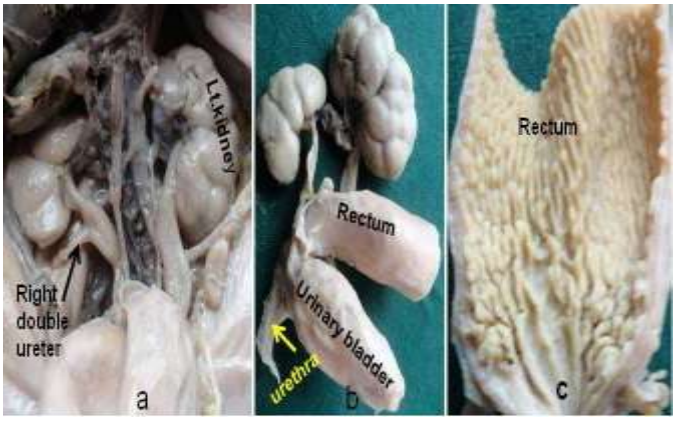

**Esophagus :**

No esophageal atresia or tracheoesophageal fistula

Rectum and Anal canal: Rectum terminated blindly behind the urinary bladder merging with its lower posterior wall [Fig 4.b]. Interior shows few longitudinal folds of mucosa in a much narrower gut region which has no external opening (Atretic anal canal with imperforate anus)[Fig 4.c].No fistulous communication between rectum and any other organ present.

**Renal defects**

**Figure 4: Renal and anal defects**




Right kidney:-Small. Length, width and thickness - 22.58X10.96X6.5 mm [Fig 4.a]

Left kidney: - Larger than right. Length, width and thickness 29.89 X 14.33X 13.96mm. [Fih 4.a]

Right Ureter:- Initial part was double ureter. Upper division was continuing with the right kidney.

Lower division was narrow at commencement and continuous with an irregular small mass of tissue. - ? renal tissue

[Fig4a]

Left ureter: Dilated (hydroureter). Both ureters were opening into postero lateral part of urinary bladder.

Urethra. Normal. No hypospadiasis nor epispadiasis nor any fistulous communication seen

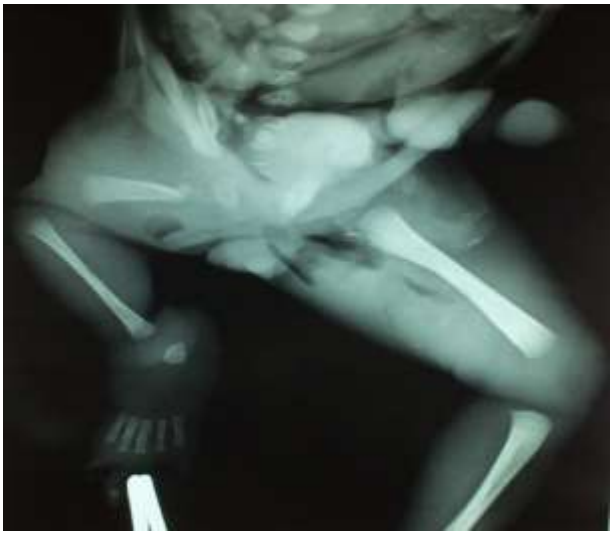
Limbs: Right lower limb was short. Right Foot: club foot

X-ray of limbs and pelvis

- short right femur
- absence of right fibula (Fig.5)

Umbilical Vessels: Abdominal aorta continued as a single umbilical artery. [Fig.1c] A single branch has originated from it. This branch has trifurcated. Out of the three branches, one continued as accessory renal artery and entered lower pole of left kidney. The other two branches proceeded to rectum. Right external iliac artery was replaced by a vascular plexus from surrounding vessels.

**Figure 5: X-ray lower limbs and pelvis**



Discussion

As there is no single definitive causative factor for VACTERL association, many hypotheses have been put forth for its causation. One among such is that VACTERL association represents a dysmorphogenetic response of primary developmental field, i.e., polytopic developmental field defects (DFD) [3].

A definite role for genetic factors in its causation is also controversial [1,4,5,6,7,8]. Studies were carried on twins, familial cases and family studies [1,3,4,5,6]. Although familial occurrence indicates possible genetic involvement no definite causative genes have been identified yet. According to development theory VACTERL may result from mesodermal deficiency caused by abnormal blastogenesis [8].

One of the less commonly observed features in VACTREL association is truncal defect of heart (persistant truncus arteriosus) .Individuals with chromosomal defects (22q11.2) may also present truncal defects .This chromosomal defect may form an overlapping condition for VACTERL association and differentiation has to be made between these two. Neural crest cells from hindbrain region may migrate to out flow region of heart through $3^{rd}$, $4^{th}$, $6^{th}$ pharyngeal arches and contribute to endocardial cushion formation in conus and truncus arteriosus of developing heart. Truncal defects are usually due to abnormal migration, proliferation or differentiation of these cells [9], but the defect in the present case is associated with other anomalies which are common in VACTERL association. A definite chromosomal defect cannot be attributed in this particular case as genetic studies were not carried out.

Tracheal agenesis is another less commonly reported entity. This was reported both in VACTERL and TACARD associations [10]. In our case though the trachea is not developed, there is a horizontal cartilaginous tube entering both lungs. As a deviation from classic description, esophagus is absolutely normal without any atresia or fistulous communication with this cartilaginous tube. In typical cases of VACTERL association, esophageal atresia and/or tracheo-esophageal fistula are reported

Infants with VATER association with both renal anomalies and anorectal atresia were significantly more likely to have genital defects [11]. An analysis of genitourinay anomalies has been undertaken in patients with VACTERL association. It was reported that Genito Urinary anomalies

were noted in patients without malformations of renal, lower vertebral or lower G I systems. [12] In the present case though there are no genital anomalies, urinary anomalies are seen in the form of small kidney and double ureter in upper part of right side

Single umbilical artery has also been observed as an accompaniment of this condition. In present case, replacement of external iliac artery of right side by a plexus of vessels might have been one of the causes for short limb on right sided (probably due to reduced blood supply)

Review of literature revealed reports of many cases with different combinations of defects, and also its occurrence with other syndromes like Prune Belly Syndrome [13]. The basic element of three or four defects was present in all. There is a reported case of a twenty three year married female patient with 3 months amenorrhea [14]. This shows that VACTERL patients with milder form of defects can lead a normal life.

Prenatal ultra sound plays a major role in diagnosing and management of this disorder. Tongsong T.et al diagnosed a case of VACTERL association and an elective termination of pregnancy was done after proper counseling of parents because of malformations in fetus fitting into VACTERL association and also severe oligohydromnios in mother [15]

## Conclusion

Present case may fit into VACTERL association as there are vascular, anal, cardiac, tracheal, renal anomalies and limb defects. As the etiology of VACTERL association is debatable and wide array of defects are included in this association, emphasis should be made on diagnosis and correction of the defects at the earliest. Utmost care should be given to every pregnancy and pregnant woman with smallest iota of doubt of any fetal defect should be thoroughly investigated for possible anomalies. A perfect prenatal diagnosis of defects, proper counseling of parents, appropriate management of pregnancy and earliest treatment to offspring is most desirable.

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