ORIGINAL RESEARCH PAPER
Volume - 15 | Issue - 07 | July - 2026 | PRINT ISSN No. 2277 - 8179 | DOI : 10.36106/ijsr
INTERNATIONAL JOURNAL OF SCIENTIFIC RESEARCH

VACTERL SYNDROME

General Medicine

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ABSTRACT

VACTERL Syndrome is a rare congenital association characterized by the random occurrence of multiple developmental anomalies. The acronym VACTERL represents Vertebral defects, Anal atresia, Cardiac anomalies, Tracheoesophageal fistula with or without esophageal atresia, Renal anomalies, and Limb abnormalities. The condition is usually diagnosed when at least three of these congenital anomalies are present in the same patient. The exact etiology remains unclear, but it is believed to arise from disturbances in embryonic mesodermal development during early gestation. We are reporting a case of a 13-year-old female patient with some features of VACTERL syndrome. Genetic testing could not be done due to resource limited setting. Early diagnosis of VACTERL syndrome is crucial for prompt intervention and improved outcomes especially in the more critical variants. A coordinated multidisciplinary approach is essential for effective management, better outcomes and long-term follow-up of affected individuals.

KEYWORDS

VACTERL SYNDROME

INTRODUCTION

VACTERL/VATER association is typically defined by the presence of at least three of the following congenital malformations: vertebral defects, anal atresia, cardiac defects, tracheo-esophageal fistula, renal anomalies, and limb abnormalities. In addition to these core component features; patients may also have other congenital anomalies. Although diagnostic criteria vary, the incidence is estimated at approximately 1 in 10,000 to 1 in 40,000 new born infants. The condition is ascertained clinically by the presence of the above-mentioned malformations. The caveat here is that there should be no clinical or laboratory-based evidence for the presence of one of the many similar conditions.

Key details regarding the age of presentation:

- Prenatal Diagnosis: The condition can be detected via ultrasound, with some cases identified between 18–34 weeks of gestation.
- Neonatal Diagnosis: Most cases are diagnosed in the first few days of life, as infants often require immediate surgical intervention for issues like anal atresia or tracheoesophageal fistula (TEF).
- Infancy & Early Childhood: While major issues are caught at birth, cardiac or renal anomalies in particular may be identified in infancy, early or rarely late childhood.
- Adulthood: In rare cases, individuals with milder forms of VACTERL association may not receive a formal diagnosis until adulthood, often prompted by long-term issues like chronic constipation (from anal anomalies), severe back pain (from vertebral issues), or the discovery of congenital heart defects.

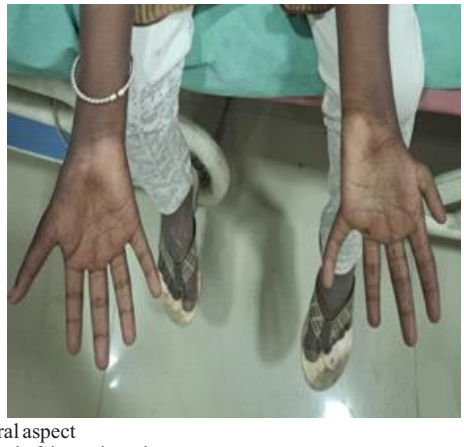
Patient presentation A 13 year old female patient, first child of non-consanguineous marriage, presented to medicine out patient department for fitness for Chronic Suppurative Otitis Media.

On Examination Short stature present
Height-4 feet 2 inch
Weight-23.85 kg
Hypertelorism

Unilateral Maxillary bossing of right side
Depressed nasal bridge
Slanted eyes
Epicanthal fold present

HAND IMAGES-

1)Dorsal aspect



2)Ventral aspect
Left hand of the patient shows-
Bony agenesis of 1 Metacarpo phalynx
Wasting over palm.

INVESTIGATIONS-

Hb	10.7 g/dl
Wbc	6.82*10^3/uL
Platelet	346*10^3/uL
MCV	63.3 fL
MCH	19.7 pg

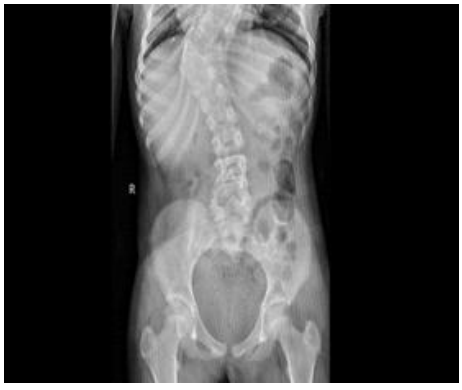
MCHC	31.2 g/dl
Esr	28 mm/hr
HHH	Negative
Sr.Sodium	138.5
Sr.Potassium	4.31
Sr.Chloride	104.7
Urea	28.1
Creatinine	0.66
T.bili	0.27
D.bili	0.11
SGOT	16.8
SGPT	20

Usg (A+P)- Right kidney not visualized in right renal fossa

Another ectopic kidney noted in left illiac fossa measuring 7.1*2.3 cm-
Normal in size, morphology, echogenicity
Left kidney -7.7*4.7 cm

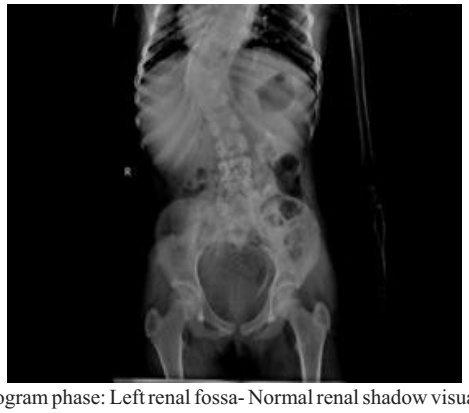
CT INTRA VENOUS PYELOGRAM-

1) Scout film



Scout film
No radio opaque calculus seen.
Bowel shadows are obscuring.

2) Nephrogram phase-



Nephrogram phase: Left renal fossa- Normal renal shadow visualized. Kidney shows normal size ,colour with prompt homogenous nephrogram.

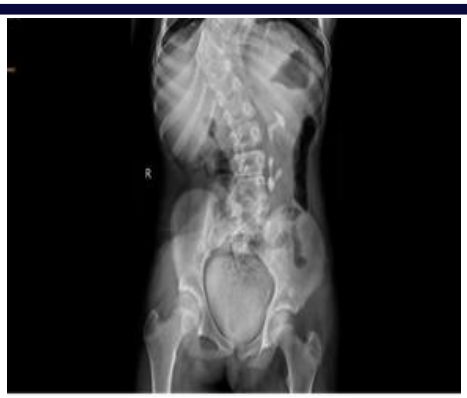
Right renal fossa -no renal shadow identified in right lumbar region
Left illlac fossa -Additional renal shadow seen in left illac fossa showing contrast uptake
S/o ectopic kidney

3) Pyelogram phase
Pyelogram phase -Pelvi Calyceal System

Left kidney-
Normal calyceal pattern& normal pelvis

No e/o dilatation
Normal course of left ureter inserts into urinary bladder at left VUJ

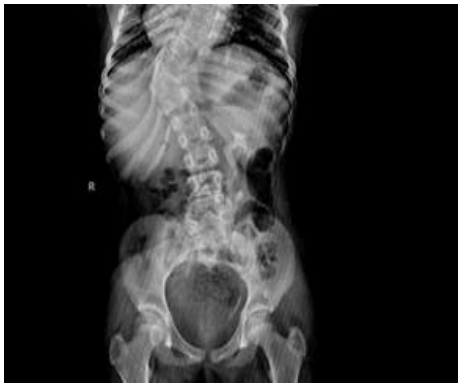
Ectopic Kidney-PC system visualized with mild malrotation & pelvis oriented medially



Ureter arising from ectopic kidney crosses midline & Inserts into urinary bladder on right side at higher than normal position

No evidence of filling defect or obstruction seen in the ureter

Urinary bladder -well opacified with smooth margin
No intraluminal filling defect



IMPRESSION

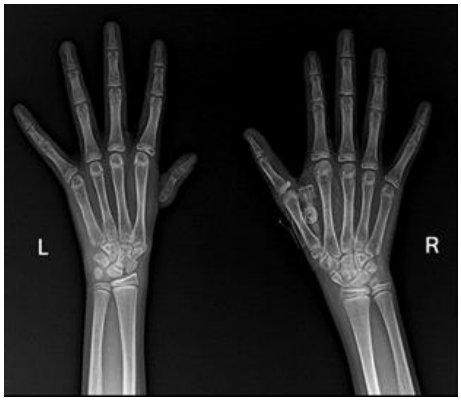
Findings s/o crossed renal ectopic without fusion
Ectopic kidney in left illac fossa & ureter inserting in contralateral side of bladder

2D Echo Ejection Fraction-

60% Dextrocardia noted Morphologically dilated Right Atrium & Right Ventricle Bicuspid Aortic Valve Dilated Aortic Root (Raphe-2 & 7 o clock, Pseudo raphe-11 o clock Pulmonary hypertension

(PASP_33mm hg) Tiny VSD (Perimembranous) Shelfine Aorta
Peripheral pulmonary stenosis

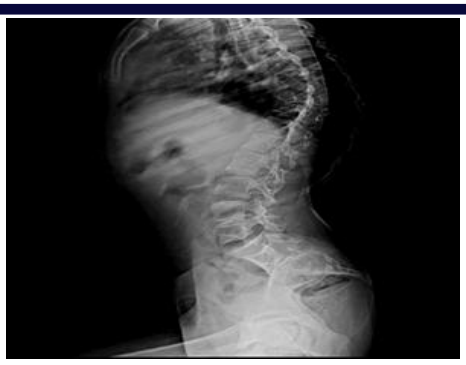
X-RAY REPORTS



X RAY BILATERAL HAND

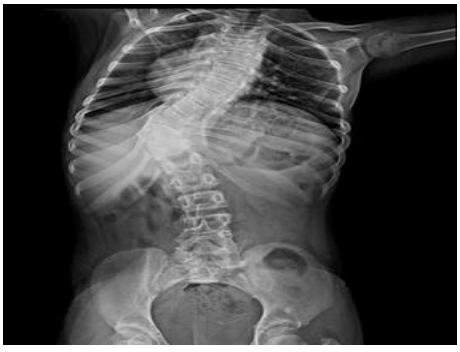
Bilateral hand
1.Severe shortening of 5th metacarpal of right hand.
2.Left hand bone unremarkable.
3.Ossification is appropriate for age.

XRAY LATERAL SPINE



Xray Thoracolumbar spine s/o-

- There is evidence of scoliotic deformity involving 3rd lumbar spine with convexity toward right side.
- Hemivertebrae noted at D11 which is focal point of scoliosis.
- 13th rib noted on right side.
- There is suspicion of fusion of posterior shaft of 10 th & 11 th right rib.
- Incidentally noted Dextrocardia
- No evidence of fracture



X RAY THORRACO LUMBAR SPINE- P A VIEW

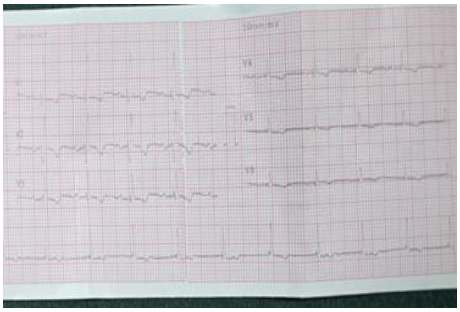
These multiple bony abnormalities described above, suggest possibility of syndromic association.

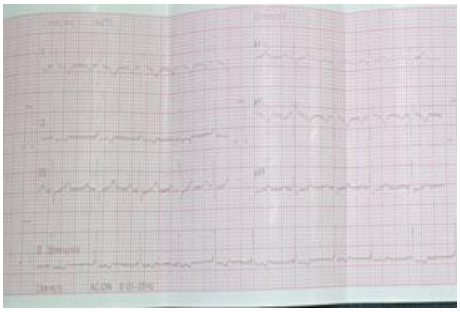
ECG

Lead I Inversion: Inversion of P, QRS, and T waves.
Lead aVR: Upright (positive) P wave, QRS complex, and T wave.
Axis: Extreme right axis deviation

Precordial Leads (V1-V6): Reversed R-wave progression (dominant S waves in all chest leads or V1-V6).

All this ECG changes favour dextrocardia.





ECG OF SAME PATIENT
FINDINGS S/O DEXTROCARDIA

FACIAL FEATURES OF SAME PATIENT-

- Hypertelorism
- Unilateral Maxillary bossing
- Depressed nasal bridge
- Slanted eye
- Epicanthal fold present.

DISCUSSION

VACTERL association is a rare congenital condition characterized by the non-random occurrence of vertebral defects, anal atresia, cardiac anomalies, tracheoesophageal fistula, renal anomalies, and limb abnormalities. The diagnosis is typically established when at least three of these component anomalies are present in the absence of another identifiable syndrome. The estimated incidence ranges from 1 in 10,000 to 1 in 40,000 live births.

In the present case, a 13-year-old female exhibited multiple anomalies consistent with VACTERL association, including vertebral defects (kyphoscoliosis and hemivertebra at D11), cardiac anomalies (dextrocardia, bicuspid aortic valve, perimembranous ventricular septal defect, and pulmonary hypertension), renal anomaly (crossed renal ectopia without fusion), and limb abnormality (bony agenesis of the first metacarpophalangeal bone). The presence of these anomalies fulfil the clinical and diagnostic criteria for VACTERL association.

Most patients with VACTERL association are diagnosed in the neonatal period because of severe anomalies requiring immediate surgical intervention, such as closure of tracheoesophageal fistula and surgery for anal atresia. However, milder or atypical presentations may remain undiagnosed until later childhood or adolescence. In this patient, the diagnosis was made incidentally during evaluation for chronic suppurative otitis media, highlighting the possibility of delayed recognition in individuals with less severe manifestations.

Renal anomalies are reported in approximately 30–50% of patients with VACTERL association and include renal agenesis, dysplastic kidney, horseshoe kidney, and ectopic kidney. Crossed renal ectopia without fusion, as observed in this case, is relatively uncommon but has been described in association with VACTERL. Cardiac anomalies are also frequent, with ventricular septal defect being among the most commonly reported defects. The coexistence of dextrocardia and bicuspid aortic valve further illustrates the wide spectrum of cardiovascular involvement. We also found shelfine aorta which is also seen coarctation of aorta. We also reported peripheral pulmanory stenosis which is found in many syndromic association like WILLIAM, NOONANS SYNDROME.

Vertebral anomalies are considered one of the most consistent findings in VACTERL association. In the present case, kyphoscoliosis associated with hemivertebra and rib anomalies was identified, which is consistent with previous reports describing costovertebral malformations as common skeletal findings. Limb anomalies, particularly reduction defects affecting the upper limbs, are also frequently observed. The metacarpal abnormality noted in this patient supports the involvement of limb developmental fields during early embryogenesis.

The exact etiology of VACTERL association remains unclear, probably sporadic etiology. It is widely believed to result from

disturbances in mesodermal development during early embryogenesis. Although most cases occur sporadically, rare familial occurrences have been reported, suggesting a possible genetic component. Genetic testing could not be performed in this patient due to limited resources, which represents a challenge in establishing a possible underlying genetic etiology.

Management of VACTERL association requires a multidisciplinary approach involving pediatricians, cardiologists, nephrologists, orthopedic surgeons, and other specialists depending on the organ systems involved.

Long-term follow-up is essential to monitor growth, functional outcomes, and complications related to the associated anomalies.

CASE STUDY

A 13 year old female patient presented to our OPD for pre operative evaluation for Chronic Suppurative Otitis Media(CSOM) surgery. At time of admission she was incidentally detected to have some features of VACTREL.

CONCLUSION

VACTERL association represents a complex and heterogeneous group of congenital anomalies with variable clinical presentation. This case highlights a rare presentation of VACTERL association diagnosed in adolescence, characterized by vertebral anomalies, limb abnormality, crossed renal ectopia, and multiple cardiac defects including dextrocardia and ventricular septal defect.

Hence, every patient coming to you for pre operative evaluation, should be evaluated for other system analysis to diagnose multi system association like VACTREL, which may have got undetected at infancy.

The case emphasizes that although most patients are diagnosed early in life, milder forms may remain unrecognized until later childhood or adolescence. Careful clinical examination and thorough radiological evaluation are crucial for identifying associated anomalies and establishing the diagnosis. Early recognition and coordinated multidisciplinary management play an important role in improving long-term outcomes and quality of life for affected individuals.

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