VACTERL association: A Case Report of a congenital malformations

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Abstract*—VACTERL / VATER association is defined by the presence of at least three of the following congenital malformations: vertebral defects, anal atresia, cardiac defects, trachea-esophageal fistula, renal anomalies and limb abnormalities. It is diagnosed clinically by the above features with presence of core component features like trachea-esophageal fistula or ano-rectal malformations. Etiology is largely unknown. Management centers on surgical correction of the specific congenital malformations in the immediate postnatal period followed by long term treatment of the sequelae. Prognosis is good if surgical correction is achieved but majority continue to be affected by their malformations throughout life.*

Keywords: VACTERL Association, Congenital Malformations.

I. INTRODUCTION

VACTERL Association is a statistically non-random co-occurrence of a group of congenital malformations which include vertebral defects, anal atresia, trachea-oesophageal fistula with/without oesophageal atresia, radial and renal anomalies with limb malformations. Because these malformations were observed to occur more often than would be expected by chance, the condition was termed an association and not a syndrome.[1] The diagnosis requires at least 3 component features for which there is no alternate causal explanation.[2] The incidence varies from 1/10000 to 1/40000 live births.[1]

A case of VACTERL association having anal atresia, vertebral defects , limb anomalies as the main component features along with ambiguous genitalia attended at the SMS Medical College, Jaipur. The case was thoroughly evaluated and a case report was prepared to publish.

II. METHODOLOGY

A case of VACTERL association having anal atresia, vertebral defects, limb anomalies as the main component features along with ambiguous genitalia was presented in NICU of Mahila Chikitsalaya, a hospital attached to SMS Medical College, Jaipur (Rajasthan) India. As it is a rare case so evaluated thoroughly to prepare a detailed case report to publish?

III. CASE REPORT

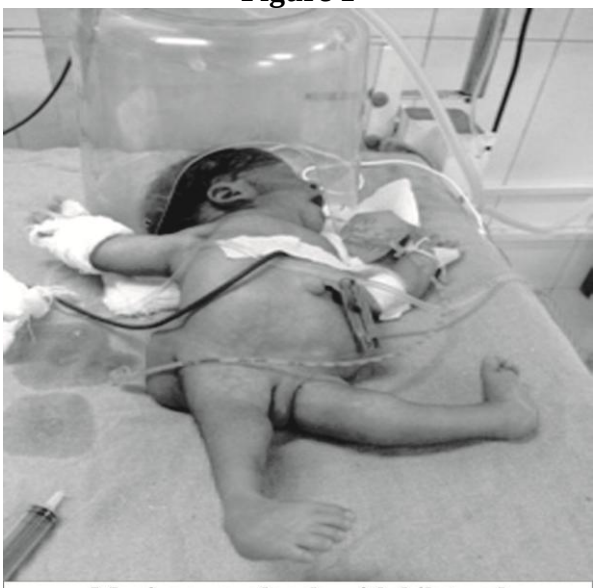
A late preterm (36 weeks) appropriate for age newborn with indeterminate sex having a birth weight 2.75 kg was born vaginally to a fourth gravida mother, aged 28 years at Mahila Chikitsalaya, attached to SMS Medical College, Jaipur, Rajasthan.

It was the product of a non-consanguineous marriage. The first three gravida were live births with no history of any abortions. The antenatal history was uneventful but there had been no proper antenatal check up with no antenatal ultrasonography done.

The baby had immediate cry after birth with an APGAR score of 7/10 at 5 minutes. Baby had respiratory distress immediately after birth, was admitted in the NICU at Mahila Chikitsalaya, SMS Medical College, Jaipur and started on oxygen by hood, intravenous fluids and antibiotics.

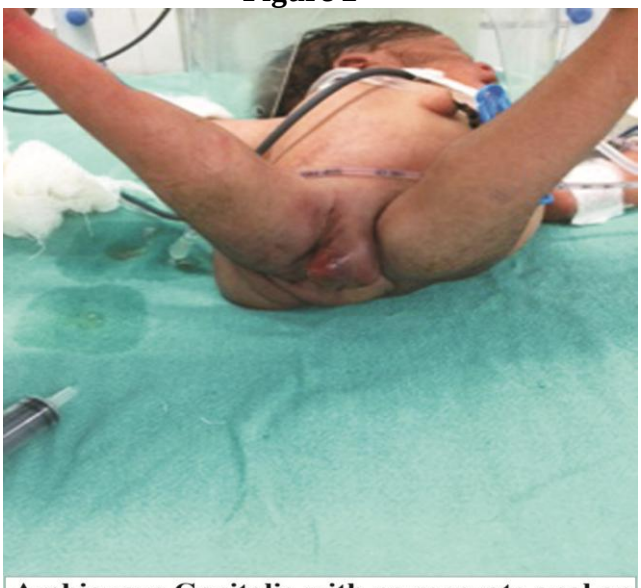
On examination, the baby had a meningo-myelocele (Figure 2) in the lumbar area, bilateral genu recurvatum and club feet , genitalia were ambiguous (Figure 1) and sex could not be determined. There were no separate anal and urethral orifices (Figure 2). The baby passed urine mixed with stool through a common orifice. The infantogram showed scoliosis with hemi-vertebrae (Figure 3).

Figure 1



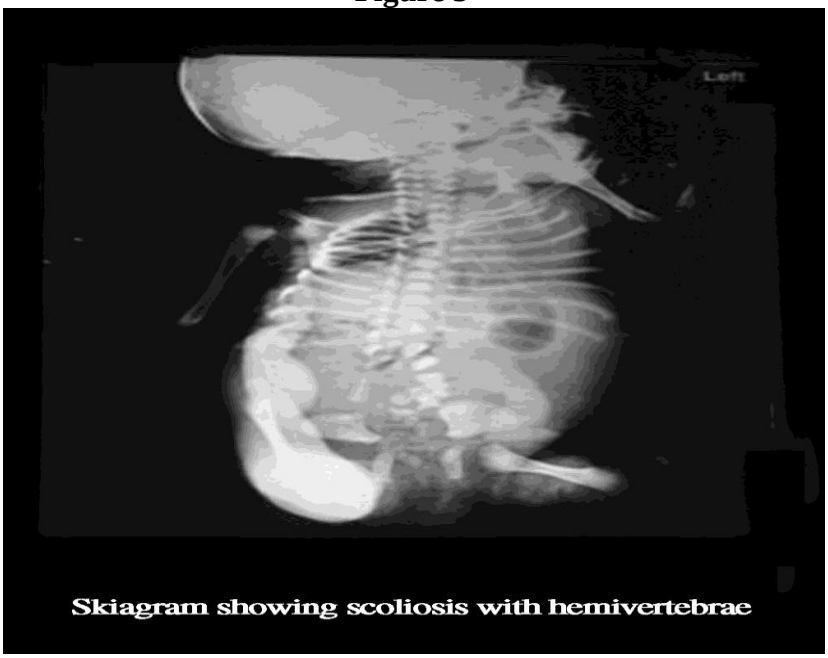
Meningo-myelocele with bilateral genu-recurvatum and clubfeet

Figure 2



Ambiguous Genitalia with no separate anal or urethral orifice and no discernible genitalia

Figure 3



Skiagram showing scoliosis with hemivertebrae

The routine blood investigations and ultrasound examination could not be done as the attendants took the baby against medical advice a few hours after birth.

IV. DISCUSSION

EEC VACTERL / VATER association is typically defined by the presence of at least 3 of the following congenital malformations – vertebral defects, anal atresia, cardiac defects, trachea-oesophageal fistula, renal anomalies and limb abnormalities.[1] In addition to these core component features, patients may also have other congenital anomalies. The condition is ascertained clinically by the presence of the above mentioned malformations, there should be no clinical or lab-based evidence for the presence of one of the many similar conditions as the differential diagnosis is very large.[2]

The association of imperforate anus, trachea-esophageal fistula, vertebral anomalies and polydactyly was first described by Say and Gerald in 1968.[3] The scope of the association was further enlarged to include VSD and single umbilical artery and even all cardiac anomalies. Nora et al used the term VACTERL to include C for cardiac anomalies and L for limb defects.[4] The term VACTERLS is sometimes used to include single umbilical artery. Khaury and Levy have suggested that VACTERL cases can be diagnosed with 3 or more of the six defects.[5]

Vertebral anomalies comprise 60-90% of the affected cases, can affect any vertebrae, involve single or multiple vertebrae, vary in severity and typically include segmentation defects, such as hemivertebrae, butterfly vertebrae, wedge vertebrae and vertebral fusions, supernumerary or absent vertebrae. Abnormal spinal curvature due to underlying costovertebral anomalies is common.[6-8]

Ano-rectal malformations occur in 55-90% of patients and are frequently accompanied by genito-urinary anomalies (including complex cloacal malformations) which occur in upto 25% of patients with VACTERL.[4-6]

Cardiac malformations have been reported in 40-80% of the affected cases and they range from minor clinically insignificant anomalies to complex malformations needing multiple surgical and long term medical interventions. The most common cardiac anomaly is VSD followed by PDA and ASD. Certain variants in isolation (as PDA or PFO) should be considered a normal age based finding rather than a component feature of VACTERL.[4-6]

Tracheo-oesophageal fistula with / without oesophageal atresia is described in 50-80% of the cases. Most common type is upper blind pouch with lower part communicating with trachea. [4-6]

Renal anomalies accompanied by ureteral and genito-urinary anomalies have been reported in 50-80% of cases. Unilateral renal agenesis is the most commonly reported renal anomaly. These can also include horseshoe kidney, cystic or dysplastic kidneys hydronephrosis, ectopic kidney and duplicate ureter. [4-6]

Limb anomalies are reported in 40-55% cases. The most common limb defect observed is radial hypoplasia followed by thumb hypoplasia. [4-6]

Hydrocephalus (VACTERL-H) due to aqueductal stenosis either as a distinct condition or continuum with more general VACTERL.

Etiology remains unknown. It is usually sporadic, a small number of familial cases have been described.[9,10] The etiology has been identified only in a small fraction of patients to date, likely due to factors such as a high degree of clinical and causal heterogeneity, the largely sporadic nature of the

disorder and the presence of many similar conditions. There is evidence for familial clustering suggestive of inherited factors. There is strong clinical and genetic evidence for causal heterogeneity in patients with VACTERL association.[11]

Diagnosis is made on clinical grounds based on the presence of the congenital malformations as above. The requirements for diagnosis vary among clinicians. Many require at least 3 component features for diagnosis while others need certain component features like trachea-oesophageal fistula or ano-rectal malformation[12]

Antenatal diagnosis can be challenging as certain component features can be difficult to diagnose before birth. The presence of a single umbilical artery may be the first clue to diagnosis. It should always result in a careful antenatal examination for features of VACTERL association.

Differential Diagnosis is broad and includes a number of conditions for which genetic testing is available. The presence of other features not typically seen in VACTERL association may suggest other disorders such as pigmentary abnormalities in Fanconi Anemia or hypocalcemia in 22q 11.2 syndrome. Ruling out these conditions is a challenging but critical part of the diagnostic work up and is essential for proper genetic counseling as there is genetic testing available for a number of these overlapping disorders. To rule out these disorders we should look for certain features that are not typical of VACTERL association such as brain malformations, ophthalmological anomalies and hearing deficits.

Management is very complex. In conditions which are incompatible with life like severe cardiac malformations, imperforate anus and tracheo-oesophageal fistula are managed with surgery in the immediate neonatal period as soon as circumstances allow. Many of the congenital malformations may result in long term sequelae.

Prognosis- Such patients face considerable medical challenges throughout life. Referral to a highly experienced centre with a co-ordinated, multidisciplinary team can greatly improve outcomes. Despite the significant associated morbidity, patients with VACTERL association do not typically display neuro-cognitive impairment.

V. CONCLUSION

A case like VACTERL / VATER association having multiple congenital malformations are only able to manage at an experienced centre having a co-ordinated multi-disciplinary team. So such type of cases should be referred to these higher centers to have better results. The patients, families and treating clinicians should be encouraged to seek care through such specialty clinics in order to achieve best possible outcome.

CONFLICT OF INTEREST

None declared till now.

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