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Case Report

A Case Report of VACTERL Association and Management of Its Renal Component

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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 report such case having congenital Renal, Vertebral and Limb defect; and management of its renal component at our institute.

Key words: VACTERL, VATER, Renal component.

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. [1,2] Although diagnostic criteria vary, the incidence is estimated at approximately 1 in 10,000 to 1 in 40,000 live-born infants. Antenatal diagnosis can be challenging, as certain component features can be difficult to ascertain prior to birth. The management of patients with VACTERL/VATER association typically centers around surgical correction of the specific congenital anomalies (typically anal atresia, certain types of cardiac malformations, and/or tracheo-esophageal fistula) in the immediate postnatal period, followed by long-term medical management of sequelae of the congenital malformations. If optimal surgical correction is achievable, the prognosis can be relatively positive, though some patients will continue to be affected by their congenital malformations throughout life. Importantly, patients with VACTERL association do not tend to have neurocognitive impairment. We report such case which presented with Lumbar scoliosis and with left radial club hand but on further work-up was found to have anomalies of vertebrae, kidneys and hence was labelled as a case of VACTERL association.

CASE REPORT

A three years old female child was brought to us with complaining of failure to thrive and excessive crying during

micturation noticed by parents. she was born at term by normal delivery following an uneventful pregnancy and there was no significant past medical or surgical history.she was the first child of non-consanguineous parents and there was no family history of congenital anomalies.History of oral contraceptive pills taken during pregnancy.

On physical examination there was scoliosis at lumbar vertebral region with no other genitourinary anomalies. Plain X ray KUb was showing Hemi vertebra at L3 with lumbar scoliosis(fig 1).

- HER lab reports showed
- Hb 9.5gm
- Urine NAD
- Blood urea 20mg%
- S creatinine 0.9%

USG was done which was suggestive of absence of Right kidney at normal position and Massive Hydronephrosis with Megaureter on left side (fig 2).

IVP was advised which shows Right hypo plastic unascended kidney with Hydronephrosis and Hydroureter (megaureter) on left side (fig 1).

So diagnosis of VACTERL association was done and patient was planned to take for surgery for salvaging Left Kidney and its function. Patient was taken for surgery after fitness. Left extended McBurney's incision was taken, peritoneum was reflected and Megaureter located it was traced upto junction with bladder (UV junction)(fig 3). Ureter was clamped and divided at UV junction. Lower end was transfixed. Then ureter was open longitudinally till L3 level. The redundant portion of ureter was excised so as to refashion a ureter of normal calibre on infant feeding tube of 6 fr size. Ureteric wall was closed with continuous Vicryl 4-0 suture. Excess length of ureter was excised in such way that remaining ureter was brought to bladder without tension. This newly refashioned ureter was reimplanted into the bladder by LEADBETTER POLITANO TECHNIQUE. [3] Infant feeding tube was brought out suprapubicaly and bladder was closed with Foley's in situ.

Infant feeding tube was removed after 8 days and Foley's was removed after 10 days. Post operative recovery was uneventful.

Regular follow up was taken. Follow up IVP done after 6 months which was showing normal functioning of left kidney (fig 4).

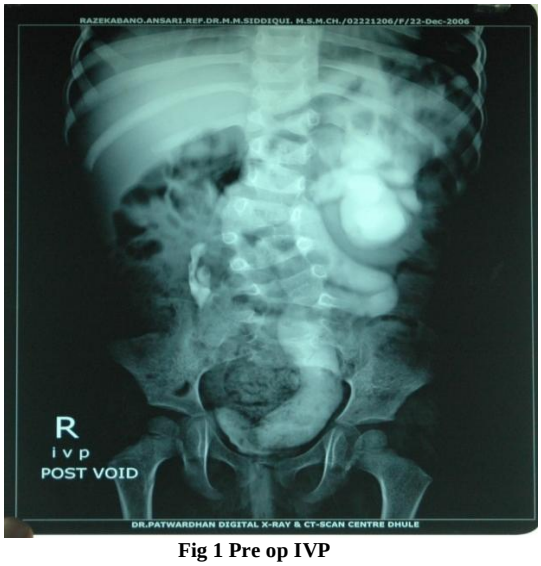

Fig 1 Pre op IVP

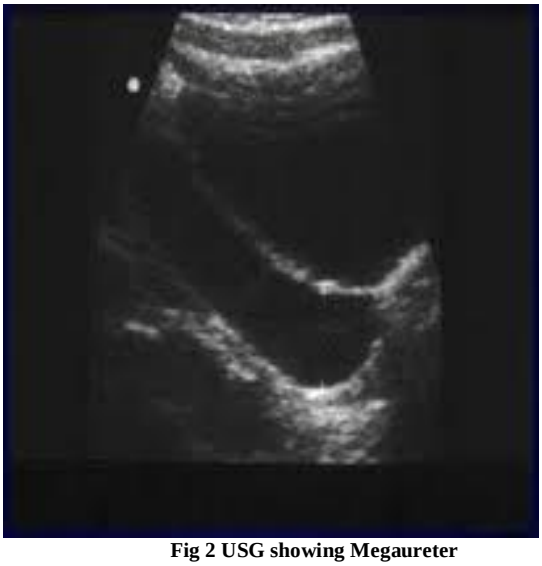

Fig 2 USG showing Megaureter

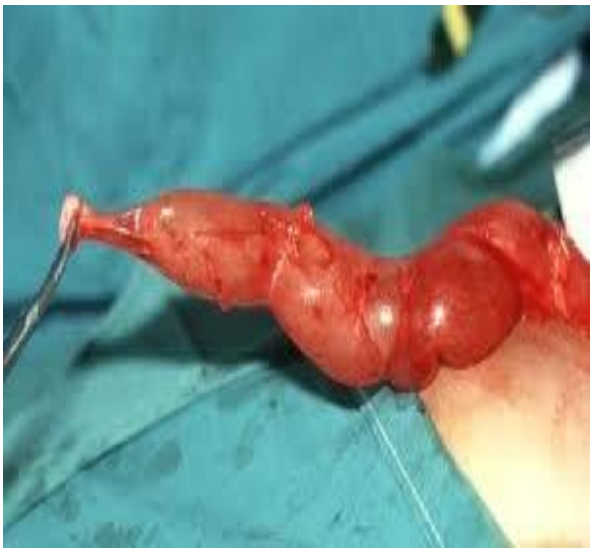

Fig 3 Intra op

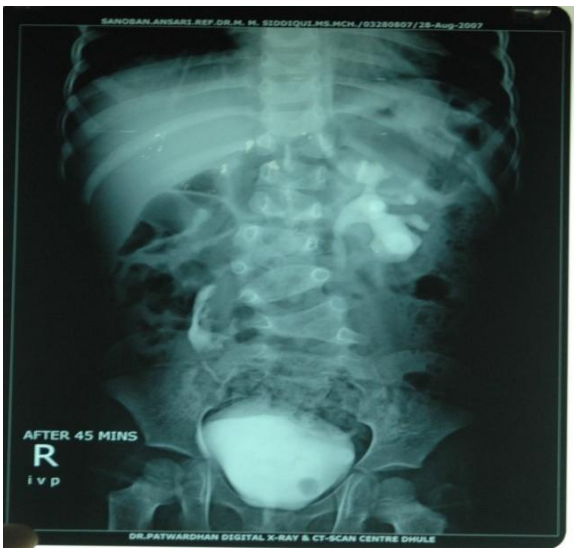

Fig. 4 Post op IVP

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. [4]

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 70 percent of babies include absent or displaced thumbs, polydactyly, syndactyly and forearm (includng 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 theexpected rate (failure to thrive). Furthermore defects of practically every organ system have been reported in association with VACTERL in lower frequency [5] like facial asymmetry (hemifacial microsomia), external ear malformations, lung lobation defects, intestinal malrotation and genital anomalies.- VACTERL shows some pheonotypic overlap with many other conditions including Feingold syndrome, CHARGE syndrome, 22qll deletion syndrome. Townes-Brocks syndrome, Pallister-Hallsyndrome, Fanconi anemia spectrum, Goldenhar Syndrome, Nager syndrome, caudal regression syndrome, sirenomelia, electrodactyly-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 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. [6] Besides recent research has shown that VACTERL could be caused by defective Shh (Sonichedgehog pathway) signaling during human embryogenesis. [7] 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 healthcare. If detected in utero(by sonography) before viability, termination of pregnancy can be offered.

CONCLUSION

Knowledge of VACTREL anomaly has initiated us in investigating the patient and diagnosing the renal anomaly early. Early diagnosis has led to early management, thus saving the child from becoming a "RENAL CRIPPLE"

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