INTERNATIONAL JOURNAL OF MEDICAL AND APPLIED SCIENCES
Earthjournals Publisher
E-ISSN:2320-3137
www.earthjournals.org

Case Report

VACTERL SYNDROME

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Abstract

VACTRL ASSOCIATION was first named in the early 1970's and it included consistent group of anomalies and congenital malformations. Vacterl syndrome stands for vertebral defects, anal atresia, cardiac defects, tracheo-esophageal fistula, renal anomalies and limb abnormalities[1].Presence of any three above mentioned abnormalities confirms the clinical diagnosis of Vacterl syndrome. Incidence is 1 in10000 to 40000 newborns [2].

Key words: VACTERL, trachea-esophageal fistula, ventricular septal defect

CASE REPORT:

We received a fetus with multiple congenital anomalies from Department of OG,SreeBalaji medical college and hospital with parent consent.The anomalies were studied from an embryological perspective.The fetus was dissected after getting proper research and ethical clearance from the university.

As received from the hospital 28 yr old female (PRIMI) in wedlock with a non-consanguineous spouse since 6 months. She was not diabetic,hypertensive,not alcoholic and not used any oral contraceptive pills. No h/o any fever,irradiation,genetic disorder known in the family. The fetus was aborted spontaneously.

MATERIALS AND METHODS:

The fetus was fixed in 10% formalin for 10 days and the same was injected through anterior fontanelle for fixing the brain.After fixation, the fetus was dissected under magnifying lens and the light source.Crown-rump length(CRL) was measured using inch-tape.

ULTRASOUND FINDINGS:

Fetus corresponds to 20weeks of gestation

- ⊡ Placenta and fetal activity appeared normal
- ⊡ SINGLE UMBLICAL ARTERY
- ⊡ PERSISTENT NON VISUALISATION OF STOMACH
- ⊡ ECHOGENIC RIGHT KIDNEY WITH MULTIPLE CYSTS
- ⊡ POLYHYDRAMINOS.

IMAGING OF THE HEART SHOWS:

- ⊡ PERSISTENT LEFT SVC DRAINING INTO DILATED CORONARY SINUS
- ⊡ VENTRICULAR SEPTAL DEFECT

- SINGLE OUTFLOW TRACT SEEN ARCHING TO LEFT(with only one branch of pulmonary artery seen)

OBSERVATION:

Our observation revealed the following in addition to the above,

- EXTERNAL FEATURES:
  -Low set ears, wide set eyes with epicanthic fold
- A)CARDIAC:
  - persistent left SVC draining into dilated coronary sinus (fig 1)
  -single outflow tract seen arching to left
  -ventricular septal defect

Fig 1

B)ANOMALIES OF TRACHEA:

- Agenesis of trachea (Tracheo-oesophageal diverticulum not divided by septum)
- Blind diverticulum dividing into right and left bronchus at its lower end (SECOND TYPE) according to FLOYD ETAL classification.(fig 2)

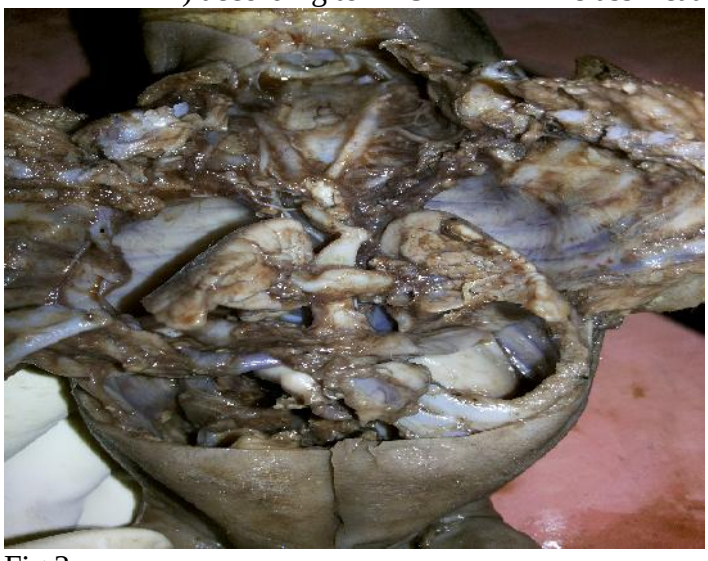

Fig 2

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C)UROGENITAL ANOMALIES:

- Right polycystic kidney and left normal kidney(fig 3)
- Absence of uterus, fallopian tube, upper part of vagina
- external genital organ seen

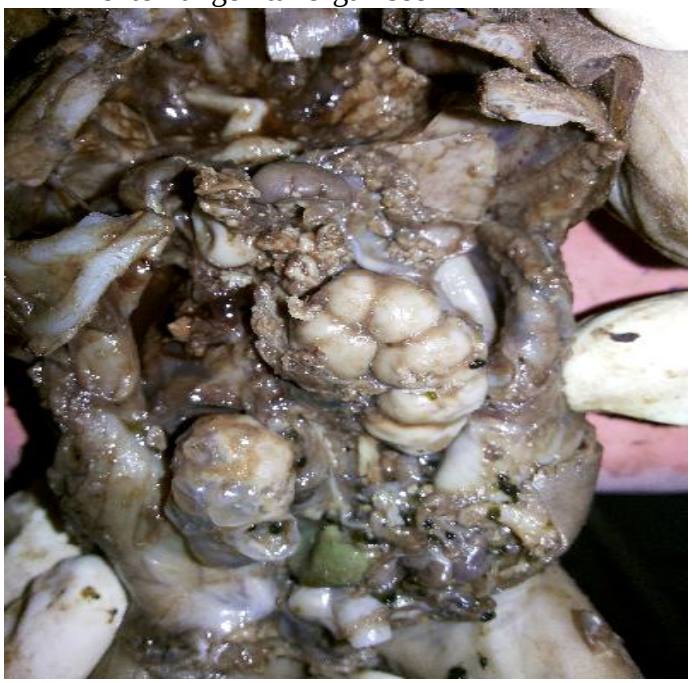

Fig 3

- GASTRO INTESTINAL ANOMALIES:

Contrary to the ultrasound report we could visualize the presence of stomach as an fusiform diverticulum in the midline along with the spleen being attached to the wall of stomach and not seen in left hypochondrium

- Presence of single umblical artery as confirmed by HPE.(fig 4)

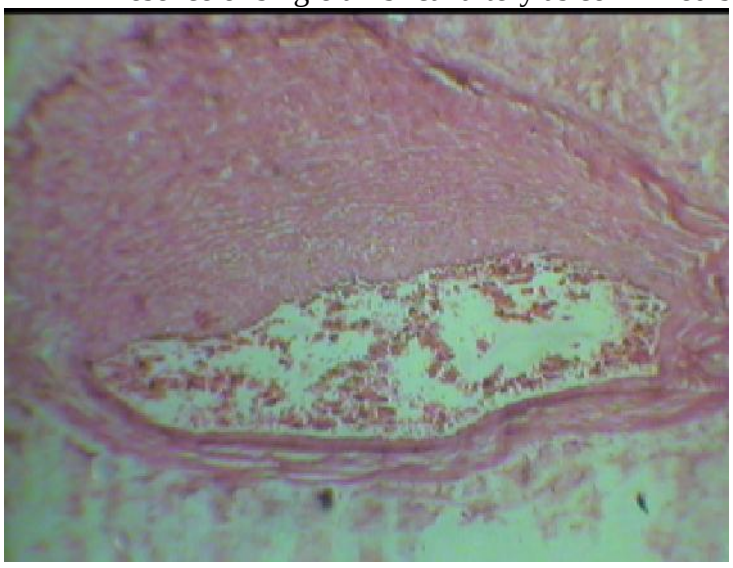

Fig 4

DISCUSSION:

PERSISTANT LEFT SUPERIOR VENACAVA is due to failure of part of anterior cardinal vein caudal to transverse anastomosis and common cardinal vein to retrogress on the LEFT side.Its persistence leads to LEFT SVC draining into coronary sinus[3]. Bulbuscordis is divided into three parts,i.e proximal, middle(conus)and distal(truncusarteriosus).A Spiral septum usually appears within the TA and subdivides it into ascending aorta and pulmonary trunk.It is formed by the union of right superior and left inferior truncusswelling.Single outflow tract is due to non formation of spiral septum.PTA(patent truncus) is always associated with VSD

Tracheo-oesophageal diverticulum gets divided into trachea and oesophagus.At the distal end it becomes bifid to form two bronchi[4].Due to the non formation of the septum,the diverticulum is not divided and leading to tracheal agenesis. TYPE 2,according to FLOYD ET AL classification[5].

Failure of the excretory tubules of the metanephros to establish contact with the collecting tubules derived from ureteric bud leads to the formation of cysts causing the polycystic kidney on the right side[3]

Absence of uterus,fallopian tube and upper part of vagina is due to non formation of paramesonephricduct (Mullerian duct)

CONCLUSION:

Vacterl syndrome specifically refers to the abnormalities in structures derived from the embryonic mesoderm[6]. Multifactorial etiology in which environmental influence triggers with a genetic susceptibility. The causes include maternal diabetics [9], teratogens (OCP,exposure to lead), Infertility treatment, chromosomal anomalies such as deletion 5q11.2, deletion 6q, duplication of 9q, mutation in HOXD 13[7, 8]

Though appropriate measures and management against risk factors decreased the incidence of such syndromes, it does not completely eliminate the possiblity of having the disease. This neccesiates further study into the multifactorial etiologies attributed to it and preventive measures to be taken.

REFERENCES:

1.Lister Hill National centre for Biomedical communication,US National library of Medicine,National Institute of Health,Published july 2014.

2.Botto LD, Khoury MJ, Mastroiacova P, Castilla EE, Moore CA, Skjaerven R et al.The spectrum of congenital anomalies of the VACTER Association:An international study.Am J med Genet 1997;71:08-15

3.IB SINGH Human embryology ninth edition publiushed by Macmillan Publishers India Ltd. Ch 15 page no.235-271.

4.VACTERL Association Sajad Ahmed, Salati Mangalore, South India:ISSN09725997, vol 9 issue 2 April-june 2010

5. Khoury MJ, Cordero JF, Greenberg F, James LM, Erickson JD. A population study of the VACTERL association: evidence for its etiologic heterogeneity. Pediatrics 1983;71:815–20.

6. Harjai MM, Holla RG, Kale CR. Full spectrum ofVACTERL in new born MJAFI 2008;64:84-85

7. Arsic D, Qi BQ, Beasley SW. Hedgehog in the human: a possible explanation for the VATER association. J Paediatric Child Health 2002;38(2):117-21

8. Froster UG, Wallner SJ, Reusche E, Schwinger E, Rehder H. VACTERL with hydrocephalus and branchial arch defects: Prenatal, clinical and autopsy findings in two brothers. Am J Med Genet 1996; 62: 169-72.

9.Genuardi M, Chiurazzi P, Capelli A,Neri G. X-linked VACTERL with hydrocephalus: The VACTERL-H syndrome. Birth Defects Orig Artic Ser 1993; 29: 235-41.