VACTERLS ASSOCIATION WITH PULMONARY ARTERIAL HYPERTENSION AND LEAST VERTEBRAL ANOMALY

*DR. BRAJA KISHORE BEHERA, **DR.SAILABALA SHAW, *DR.RISHAV RAJ
*P.G (PAEDIATRICS), **P.G (O&G)
UTKAL UNIVERSITY

INTRODUCTION
In early 1970’s VATER association was first named. Clustering of features involves the idea of the “developmental field defect”, in which malformations that occur in blastogenesis tend to result in polytopic anomalies, or birth defects affecting multiple organ systems. The clinical manifestation of VACTERLS association includes vertebral anomalies, anal atresia, congenital heart disease, tracheoesophageal fistula, renal dysplasia and limb abnormalities, Single umbilical artery. As initially described, the condition included the statistically non-random co-occurrence of a group of congenital malformations. Because these malformations were observed to occur together more often than would be expected by chance, the condition was termed an association. However, there was not (and still remains no) evidence for a single, unifying cause that would result in the condition being termed a syndrome. As per Some authors VACTERLS association would be more accurately described as a “primary polytopic developmental field defect” rather than an association. Congenital malformations are a major cause of fetal loss. Most of these fetuses are aborted at an early gestational age. The etiology of these defects may be genetic, environmental or multifactorial. No definite etiology has been proved, but a defect in mesodermal differentiation, in early first trimester, has been suggested.

CASE REPORT
A term baby weighing 2.1Kg was born at 36 week gestation from a 35 year old second gravid, consanguineous marriage with no significant history of any infection, drug intake, hypertension or diabetes mellitus, regular antenatal check up in a peripheral hospital, with history of previous abortion. There was no history of exposure to any known teratogen. The antenatal USG (done at 30 and 34 weeks) showed oligohydramnious with limb anomaly. The baby was delivered with severe respiratory distress HR 140/min with oral secretion after birth. A single umbilical artery was noted at the time of cord ligation. Endotracheal intubation and resuscitation was carried out. The baby put under ventilation for 3 days, mild cyanosis and saturation was around 78-82 %. On echocardiography the child have pulmonary arterial hypertension. Baby was planned for trachea-esophageal fistula repair. But the baby could not revived and died. The anomalies were oesophageal atresia (inability to pass OG tube in oesophagus), right side tri radiate foot, absent anal opening, thoracic hemivertebra, unilateral right dysplastic kidney, ASD, PDA, PAH on echo cardiography.

ON EXAMINATION
Vertebral-Thoracic hemivertebrae
Anal atresia
Cardiac-ASD,PDA,PAH
Esophageal atresia
Renal-dysplastic rght kidney
Single umbilical artery
Limb-tri-radiate foot
DISCUSSION:
VACTERL association is a nonrandom co-occurrence of birth defects. It is called association rather than syndrome because the defects are not patho-genetically related but tend to occur more frequently than expected and in general the etiology of association is not clearly known. Incidence of this association is approximately 1 in 10,000 to 1 in 40,000 live births.[1]
Our patient had vertebral anomaly, anal atresia (imperforate anus), cardiovascular anomalies (ASD, PDA, PAH), renal anomalies like dysplastic right kidney and limb deformities like tri-radiate foot.
VACTERL association is diagnosed by presence of any three of the classical findings. Vertebral anomalies commonly accompanied by rib anomalies have been reported in approximately 60-80% of patients. Vertebral anomalies include hemivertebrae, butterfly vertebrae, “wedge vertebrae, vertebral fusions, and supernumerary or absent vertebrae and other forms of vertebral dysplasia.
Imperforate anus/anal atresia as part of an ARM occurs in approximately 55-90% of patient. A complete imperforate anus is often discovered in the immediate postnatal period, typically through routine examination..
25% of cases GU anomaly detected in patients with VACTERL association, such as is the case with fistulae connecting the GU and anorectal tracts [4,5]
VACTERL association has a defect in the differentiating mesoderm occurring between 6th and 10th weeks of gestation [2]. Most of these cases are sporadic. No clear pattern of inheritance has been demonstrated.
It is suggested that multiple genetic and environmental factors may play a role in determining the risk of developing and inheriting this disorder. The effects of teratogens, maternal diabetes, in-utero exposure to estrogen and/or progesterone containing compounds, statins, lead and infertility treatment have been variably
documented as one of the multifactorial causes of VACTERL association. A defect in blastogenesis may be a possible etiology of this malformation. As per Martinez-Frias et al combinations of anomalies of blastogenetic origin, such as VATER/VACTERL should be considered and called “polytopic field defects” instead of the generic term “association” [3].

Certain clues can suggest VACTERL-type anomalies, such as lack of a gastric bubble due to TEF, and a dilated colon due to imperforate anus vertebral anomalies, cardiac malformations, renal anomalies, and limb abnormalities, may be ascertained more easily by antenatal ultrasound [6,7].

And also presence of single umbilical artery (SUA) in the umbilical cord was missed during USG which is an important clue for antenatal diagnosis. The presence of Single umbilical artery should always result in careful antenatal examination to look for features. In this case antenatal ultrasound could not identify the single umbilical artery. Up to 35% of patients with this anomaly have a single umbilical artery.

Many babies with VACTERL association may not have facial dysmorphic features, learning disability or abnormalities of growth. Some babies with VACTERL are born small and have difficulty with gaining weight although development and intelligence in survivors is usually normal.

CONCLUSION
Prenatal ultrasound plays a key role for diagnosing any congenital defect in. Utmost care should be given to every pregnancy and slightest doubt regarding any fetal defect should be thoroughly investigated. A proper counseling of the parents regarding birth defect and its outcome and regular ultrasonographic study in subsequent pregnancies is mandatory.

REFERENCES