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Morphological Features of VACTERL Malformation, an Anatomical Case Report

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ABSTRACT

Introduction: we report a case of congenital anomalies in an embalmed full term fetus, which involved several systems of the body and later it was diagnosed be VACTERL syndrome. On examination, it was observed that the fetus presented with syndactyly in both the upper limbs. On dissection, it was observed that there was presence of tracheoesophageal fistula, as the upper part of the esophagus was terminating as a blind pouch. The fetus also had imperforate anus and single fused kidney. This fetus was donated to us by the department of obstetrics and gynecology of our institution and it was mounted in our anatomy museum. The defective differentiation of mesoderm in the fourth to fifth week of gestation is suggested to be the embryological basis of this syndrome.

Keywords: Anus; Imperforate; Tracheoesophageal Fistula.

Introduction

VACTERL association is characterized by the presence of assembly of congenital anomalies. VACTERL syndrome was initially reported in the early 1970's, with multiple congenital anomalies in the heart, vertebral column, anal canal, trachea, esophagus, kidney and limbs[1,2]. The presence of at least three of these congenital anomalies diagnoses the VACTERL syndrome. This syndrome is actually a primary polytopic developmental field defect[3]. There exist several diagnostic criteria and it is hard to estimate the incidence of this group of malformations. The frequency of this anomalies range between 1 in 10,000 to 1 in 40,000 newborns[4,5]. Most clinicians expect the presence of at least three of the anomalies, however there is more emphasis towards certain features[6]. The etiological factors of this syndrome are deliberated as this is linked to other similar conditions like Prune-Belly syndrome, Klippel Feil syndrome and Goldenhar syndrome. The females taking birth control medication are considered to be at risk to give birth to a child of VACTERL association. However, there are no specific hereditary and chromosomal abnormality in this syndrome. It was also described that, VACTERL can be seen in children diagnosed with Edwards syndrome. The diabetic mothers carry the risk of giving birth to a child with VACTERL. But it was reported that there exist multiple factors in relation to this syndrome and there will be variation in the structures derived from the embryonic mesoderm[7]. Each and every child born with VACTERL syndrome looks distinctive.

Case Report

During the dissection and mounting of a full term embalmed female fetus in the anatomy museum, it was observed that it presented with syndactyly in both the upper limbs. There was absence of thumb and absence of radius bone (radial dysplasia) bilaterally. The hand with four fingers was seen to be placed right angles to the forearm (Fig. 1A). There was also presence of anal atresia and imperforate anus (Fig. 1B). On further dissection, multiple congenital anomalies were observed, which affected the several organs. There was tracheoesophageal fistula, which was associated with the esophageal atresia. The upper part of esophagus terminated as a blind tubular muscular pouch measuring 26 mm. At the bifurcation of trachea, there was an additional trachea-oesophageal fistula (Fig. 1C). In the abdominal cavity, the oesophagus continued down with stomach which was seen to be enlarged abnormally. There was no ascending colon and the caecum with the appendix was present in the left quadrant of abdomen. The caecum continued with the transverse colon and showed a descending colon that continued with sigmoid colon, which was present in the right iliac fossa, and terminated in a diverticulum that would represent a mal-developed rectum. Hence there was no continuation of the rectum with anal canal and the anal opening. Externally there was imperforate anus (Fig. 1B). The liver lobes were seen to be equally large on both sides beneath the diaphragm and the stomach formed a deep visceral impression on the inferior surface of the same. It also presented with malformed kidneys with large supra renal glands. There were no bilateral kidneys, but a single fused renal mass

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(Fig. 1D) was present along the median plane, in front of the vertebral column just above the pelvic brim. The ureter was bilaterally present but were arising from the front of the renal mass.

Discussion

VACTERL is a mnemonic and every alphabet in it denotes a congenital anomaly, which is seen in the affected neonates. It was reported that, only 1% of cases reveal the full range of malformations[5]. The vertebral anomalies in this syndrome are seen in 70% of cases, which include the hypoplasia of vertebral column and hemi vertebra. Initially the vertebral anomalies are not recognized, but later the child may exhibit scoliosis. The imperforate anus and anal atresia are seen in 55% of cases with this syndrome. It was reported that, about 75% of children with VACTERL syndrome have associated congenital heart diseases like interventricular septal defect, atrial septal defect and Fallot's tetralogy. The esophageal atresia and tracheo-esophageal fistula are seen in nearly 70% of children, who are diagnosed with VACTERL association. About 15-33% of children with tracheo-esophageal fistula have co association with congenital heart diseases. Defective kidneys are seen in half of the children with this syndrome. They may have defective development of unilateral or bilateral kidneys and may develop obstructive uropathy. Single umbilical artery was present in about 35% of cases with kidney or urological problems. Therefore, few clinicians have added 'S' to the VACTERL and it becomes 'VACTERLS'. Anomalous development of the limbs is seen in about 70% of babies, which include polydactyly, syndactyly, anomalous forearm like radial aplasia and absence or displaced thumbs. The children with bilateral upper limb or lower limb defects are often associated with defective kidneys and urological problems. It was also observed that, children with unilateral limb defect tend to have renal anomaly on the same side.

The VACTERL are usually small babies with low birth weight and they have difficulty in putting weight and suffer from growth retardation. Gedikbasi *et al.*[8]

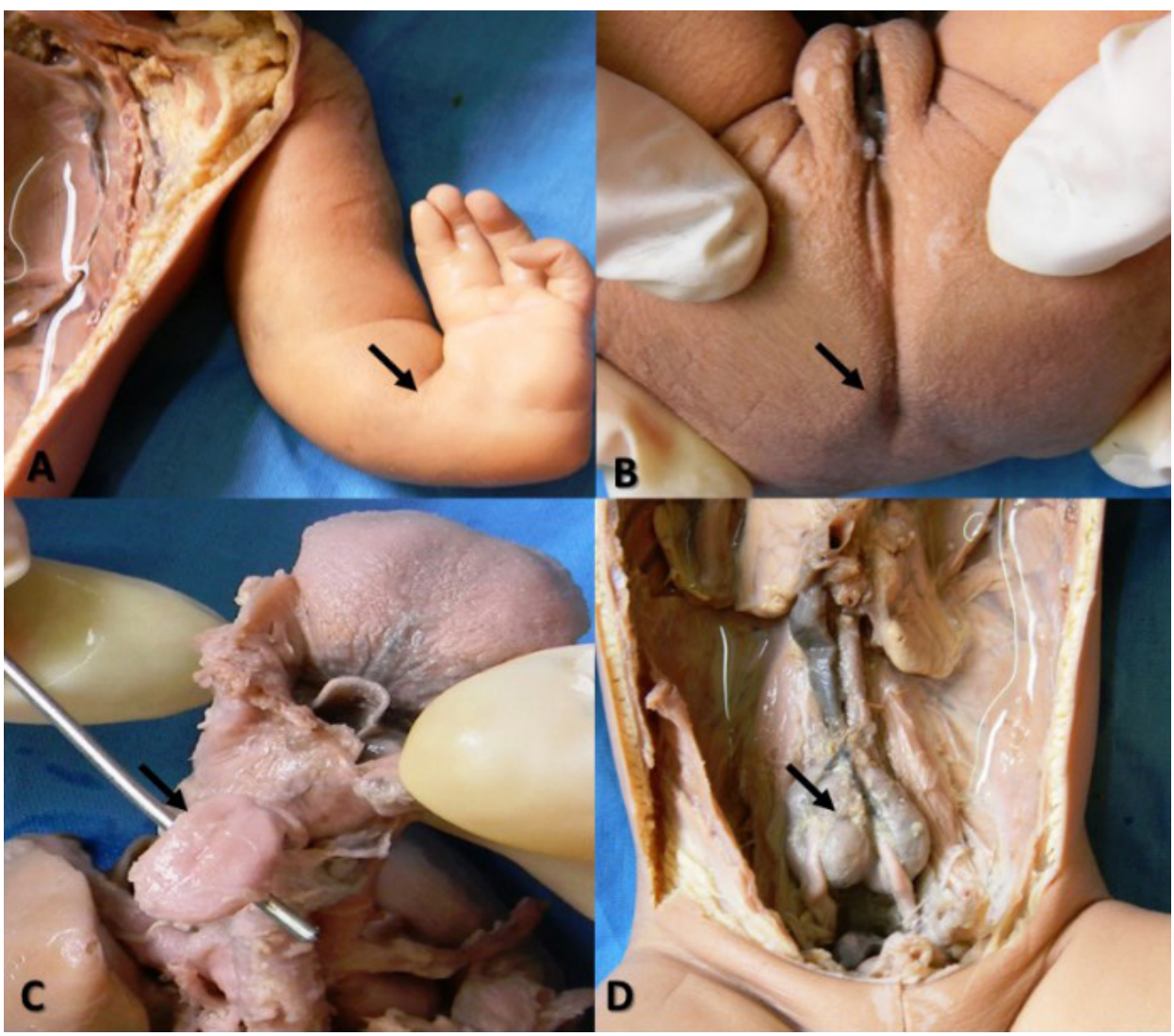

Figure 1. Embalmed fetus diagnosed as VACTERL syndrome showing; A. syndactyly with hand positioned perpendicular to the forearm; B. imperforate anus; C. tracheo-oesophageal fistula; D. single renal mass situated just above the pelvic brim.

observed unilateral caudal regression syndrome in a child of VACTERL. Harris *et al*.[9] observed lumbo-costo-vertebral syndrome associated with the VACTERL, in which there was vestibular fistula, absent right sided ribs, tethered spinal cord and sacral dimples. Komura *et al*.[10] reported a case of VACTERL with hypoplasia of Mullerian duct, renal atrophy and dysplasia of the somites at the cervical and thoracic region. Hemifacial microsomia, external ear malformations, defects of lobes of lung, malrotation of gut and genital anomalies are also associated in few cases[6]. VACTERL have been shown pheonotypic similarity with syndromes like Feingold, CHARGE and 22qll deletion[11]. Shah *et al*.[12] described a case of VACTERL with prune belly syndrome. They suggested that, both these syndromes are due to defect in the mesodermal differentiation and they are incompatible with life. The defective development of differentiating mesoderm in the four to five weeks of intrauterine life have been suggested to be the basis of these syndromes[13]. The VACTERL may be associated with other syndromes due to the common etiology and is termed as the axial mesodermal dysplasia spectrum[14]. It was reported that VACTERL syndrome is more common in babies who have diabetic mothers. The hypo-lipidemic drugs like statins are contraindicated in the first trimester of pregnancy, because there are studies which suggested that these drugs may be the etiological factor in VACTERL syndrome. The overall prognosis is poor for the VACTERL and it depends on the harshness of the anomalies. The combination of malformations and the diagnostic and management facilities available at the hospital will have impact in this association of anomalies.

Conclusion

This fetus is displayed in our anatomy museum, which can be of help to the undergraduate medical students and postgraduates. Reporting of this case is enlightening to the pediatricians, geneticists, clinical embryologists, morphologists and anatomists.

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