Prenatal Diagnosis Of VACTERL Association: Usg Findings

Shristy Mohanty
India

VACTERL is an abbreviation for the congenital group of abnormalities, including vertebral / vascular anomalies, anal atresia, cardiac defects, tracheo-oesophageal fistula / oesophageal atresia, renal defects and limb abnormalities. Most cases of VACTERL association are sporadic, which means they occur in people with no history of the condition in their family. Rarely, families have multiple people affected with VACTERL association. The exact cause is unknown; however, several environmental and genetic factors are included in literature. Three components out of seven are used to label the case as VACTERL. The combination is necessary, but other congenital malformations may be present as well. Congenital anomalies are associated with increased risk of preterm birth. Congenital anomalies are an independent risk factor for neonatal morbidity and perinatal mortality in preterm birth.

I present here two cases of antenatal ultrasound scans with VACTERL associations. VACTERL association is reported to be uncommon. It is a complex entity with multi system congenital malformations. It has been proposed that the diagnostic criteria should also include single umbilical artery”. It is important to note that the discovery of a single umbilical artery may be the first indication of the diagnosis. Some components of it can be diagnosed quite early by antenatal imaging, but ano-rectal and tracheo-oesophageal anomalies may remain undetected.

The treatment of VACTERL association includes surgical correction of severe cardiac defects, imperforate anus, trachea-oesophageal fistula and limb defects. Prognosis is not good. Hence, if detected in-utero, termination of pregnancy is advisable. In post-natal period, early detection and surgical intervention along with lifelong rehabilitation can improve the outcome. Patients with VACTERL association do not tend to have neuro-cognitive impairment.