

PRENATAL DIAGNOSIS OF VACTERL ASSOCIATION: USG FINDINGS

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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. Birth defects are associated with an increased risk of preterm delivery. Birth defects are an independent risk factor for neonatal morbidity and perinatal mortality in preterm delivery. I present here two cases of prenatal ultrasounds with VACTERL associations. To examine the potential role of ultrasonography in the prenatal diagnosis of VACTERL association. Patient no. primigravida at 26 weeks ppg with polyhydramnios in preterm labour, patient no. 2 primigravida at 19 weeks 2 days ppg with nil liquor. usg findings:- nil liquor dolicocephaly b/l echogenic kidneys (multiple tiny cortical cysts, suggestive of polycystic kidney disease) dilated sigmoid colon and rectum suggestive of anal atresia single umbilical artery was also seen in this case. 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". (1.) It is important to note that the discovery of a single umbilical artery may be the first indication of the diagnosis. (2.) 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. conclusion: These 2 cases highlight various imaging features of VACTERL association. Early detection is important so that surgical interventions can be undertaken to improve prognosis. Skilful antenatal screening would aid in timely diagnosis.

Is the VACTERL combination inherited - A specific and consistent genetic defect has not been identified in people with VACTERL combination. Very few sporadic cases of VACTERL association have been associated with mutations in FGF8, HOXD13, ZIC3, PTEN, FANCB, FOXF1 and TRAP1 genes and mitochondrial DNA. When a condition is defined as an "association" it means that it is made up of a series of specific characteristics which have been found to occur together more often than would occur by mere chance, but for which no specific cause has been determined (idiopathic). For individuals with a VACTERL combination, the risk of recurrence in a sibling or child is generally estimated to be around 1% (1 in 100). There are very few reports of recurrence of the VACTERL combination in families in the literature. The researchers stated that when dysmorphic features, growth abnormalities and / or learning disabilities are present in addition to features of the VACTERL combination, it may actually be due to a syndrome or chromosomal abnormality; if so, the risk of recurrence for a family member would be the risk associated with that specific diagnosis. Genetic disorders that have common features with VACTERL include Feingold syndrome, CHARGE syndrome, Fanconi anemia, Townes-Brocks syndrome, and Pallister-Hall syndrome. It was also recognized that there is a two to three-fold increase in the incidence of multiple birth defects (with characteristics that overlap with those of the VACTERL combination) in children of diabetic mothers. Are genetic tests available for the VACTERL combination? Because there is no known cause of VACTERL association, there is no specific test to confirm the diagnosis of this condition. If an individual has a specific diagnosis of another syndrome or disease genetics. In addition to the characteristics of the VACTERL combination, genetic tests may be available for this disease. The Genetic Testing Registry (GTR) is a centralized online resource for

information on genetic testing. The target audience for the RTM is healthcare providers and researchers. Patients and consumers with specific questions about a genetic test should contact a healthcare provider or genetics professional. Please consult the list of laboratories testing for the VACTERL association. Currently, the laboratory is collecting samples in order to identify new genes responsible for congenital anomalies, in particular the VACTERL association, caudal dysgenesis syndrome, cloacal exstrophy (OEIS) and bladder exstrophy (BEEC). Are there specific tests available during pregnancy to find out if a fetus has VACTERL combination? There is no diagnostic test for the VACTERL combination during pregnancy. However, many structural abnormalities in the fetus can be seen on an ultrasound in the second trimester, usually between 18 and 20 weeks, when anatomical details are visible. The results of an ultrasound may indicate whether additional ultrasounds (or a more specialized ultrasound) may be indicated, or whether another type of fetal test may be indicated. A normal ultrasound result can help provide relief for people with a family history of a disease such as VACTERL. In addition to a fetal ultrasound, a fetal echocardiogram may be performed, which can provide a detailed picture of the fetal heart.

It can be used to confirm or rule out a congenital heart defect, which is a common finding in people with VACTERL combination. People interested in having a fetal echocardiogram should discuss the availability, risks, and limitations of this test with their health care provider. How do I find a genetics professional in my area - To find a health professional who specializes in genetics, you can ask your doctor for a referral or you can search for one yourself. Online directories are provided by the American College of Medical Genetics and the National Society of Genetic Counselors. If you need further assistance, contact a GARD Information Specialist. You can also find out more about genetic counseling from the Genetics Home Reference.

How VACTERL Combination is Diagnosed Before and After Birth - Prenatal diagnosis of VACTERL Combination can be difficult because some features of the disease may be difficult to detect before birth. Therefore, the diagnosis of VACTERL is usually based on the characteristics observed at the birth of a baby or during the first days of life. The diagnosis is based on the existence of at least three of the following characteristics

(which constitute the acronym VACTERL): spinal defects, usually accompanied by abnormalities of the ribs; imperforate anus or anal atresia; heart defects (heart); tracheoesophageal fistula with or without esophageal atresia; kidney (kidney) abnormalities, including renal agenesis, horseshoe kidney, and cystic and / or dysplastic kidneys; and limb abnormalities. Other types of abnormalities have also been reported in affected individuals and can be used as clues to consider a diagnosis of other conditions with overlapping characteristics. Depending on the characteristics present, some other conditions that may be considered when diagnosing a child with association characteristics VACTERL (differential diagnosis) may include Baller-Gerold syndrome, CHARGE syndrome, Currarino disease, 22q11.2 microdeletion syndrome, Fanconi anemia, Feingold Fryns syndrome, MURCS combination, oculo-auriculo-vertebral spectrum, Opitz G / BBB syndrome, Pallister-Hall syndrome, Townes-Brocks syndrome and VACTERL with hydrocephalus. A woman in her thirties, G2P1, presented to the antepartum unit for preterm labor. Her physical exam predicted that she was three weeks older than the dates of the last menstrual period (PML). She was 32 weeks 3 days old by ultrasound evaluation, including all biometric measurements (biparietal diameter, head circumference, abdominal circumference, femur length, humerus length), which was consistent with her LMP dating of 32 weeks 1 day . The family had recently moved from another state due to her husband's military service. According to her account, she had two previous ultrasound exams at a small community hospital where they said the heart was enlarged but no other abnormalities were noted. Her previous pregnancy was unremarkable, with a normal three-year-old daughter at home. She was in good physical condition, a non-smoker, and had no history of drug addiction. She had no history of diabetes or hypertension. She was referred for an ultrasound evaluation. Obstetric ultrasound was performed using an Acuson Sequoia 512 Imaging Unit (Acuson, Mountain View, Calif.) With a 6 MHz curved linear array. Due to the cardiac findings, a fetal echocardiogram was also ordered and performed by the same sonographer after the obstetric ultrasound. The same machine and the same transducer were used.

Ultrasound findings included hydramnios with an Amniotic Fluid Index (AFI) measured at 31 cm as well as a non-visualization of the stomach in the fetal abdomen (Figures 1 and 2). Further evaluation of the fetal neck and thorax revealed a blind termination esophagus with possible anterosuperior communication with the trachea. VACTERL syndrome is a multisystem birth defect.

Potential findings include vertebral, anorectal, and cardiac abnormalities tracheoesophageal fistula / esophageal atresia; and kidney and limb abnormalities. This case report features a pregnant woman in her early 30s with multiple fetal abnormalities suggesting this diagnosis