VACTERL- a rare syndrome associated with a many birth defects (case report)

V. Johannova, D. Smetanova

GENNET- Center for Genetics, Fetal Medicine and Assisted Reproduction, Czechia

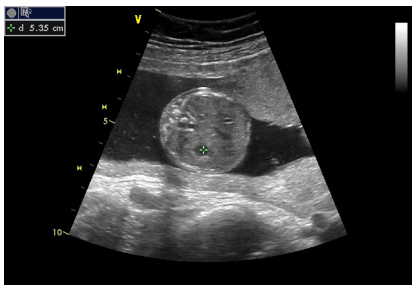
OBJECTIVE

Esophageal atresia is an incomplete formation of the esophagus. It is the most frequent anomaly of the esophagus frequently associated with tracheoesophageal fistula. The estimated incidence is 1 in 3500 live births (1 in 4000 in the U.S.). Other congenital malformations are present in up to 50 % of cases. Two syndromes in particular are associated with esophageal atresia: - VACTERL (vertebral anomalies, anal atresia, cardiac malformations, tracheoesophageal fistula, esophageal atresia, renal anomalies and limb anomalies) – CHARGE (coloboma, heart defects, atresia of the chloane, retardation of mental/or physical development, genital hypoplasia and ear abnormalities).
About 19 % of infants with esophageal atresia meet criteria for VACTERL

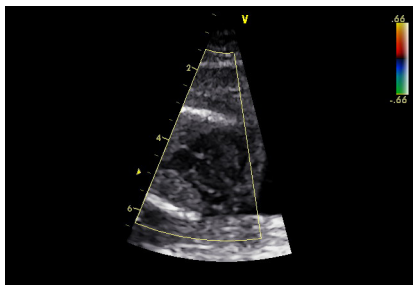
METHODS

Case report:

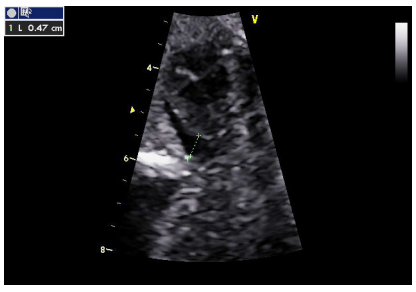
- 33 years old secundipara, spontaneous pregnancy
- 1st trimester ultrasound scan at 13th week with normal sonoanatomy
- combinated test negative, RT21 1/4750, RT13+18 1/940, very low values of both markers (PAPP-A 0,35MoM, FbhCG 0,32MoM!)
- integrated screening positive for NTD- R NTD 1/12! (AFP 3,24, uE3 0,74, hCG 5,66 MoM), RT21 1/730 (neg)
- US scan at 17th week- the finding: pericardial effusion, symetric 4 chamber view, cardiac outflow tracts is seen normally. The heart is stored more to the right side, apex of the heart to the left. Myocardial hypertrophy of both chambers. The right kindey is dystopic stored under the bifurcation of the aorta. The rest of the sonoanatomy looks normally. Fetal biometry was corresponding.
- based on the US finding on fetal heart consular examination was indicated at the pediatric cardiac center of University Hospital Prague Motol with results- normal basal anatomy of the heart with good function of both ventricles chambers, tiny pericardial effusion, relative hypertrophy of chamber‘s myocard
- complete serological examination including TORCH antibodies was performed with negative results, just toxoplasmosis - IgG positive, KFR posit., IgM and IgA negative
- AMC was recommended, the patient doesn’t accept
- next US control after 3-4 weeks, without progress of the pericardial effusion, repeatedly detected fetal tachycardia about HR 160-170, but from 20th week border fetal biometry, about 28th week the polyhydramnios appears
- progress of the polyhydramnios (suspected for stenosis or atresia GIT) and FGR at 31st week, abnormal CPR from Doppler AU, ACM
- the patient was followed up in the perinatal center from 32nd to 35th week
- The pregnancy was terminated at 35th week with an acute cesarean section for fetal hypoxia.



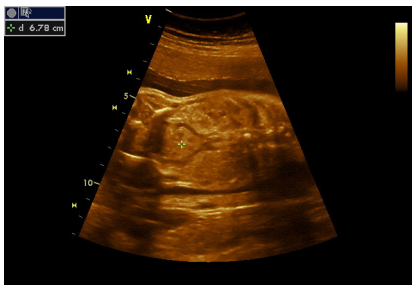
stomach
18w



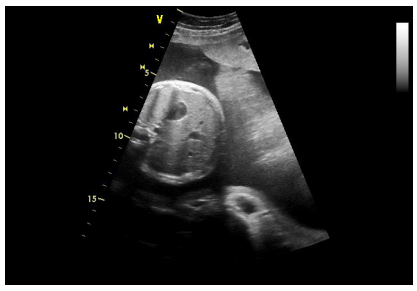
pericardial effusion
18w



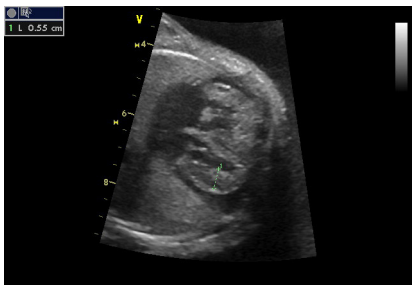
pericardial effusion
22w



dystopic kidney
18w



stomach
30w



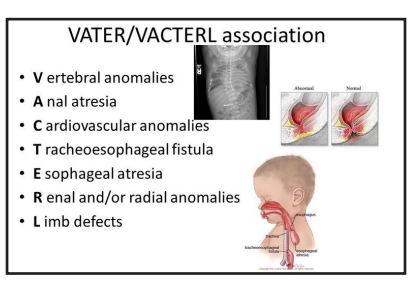
heart
30w

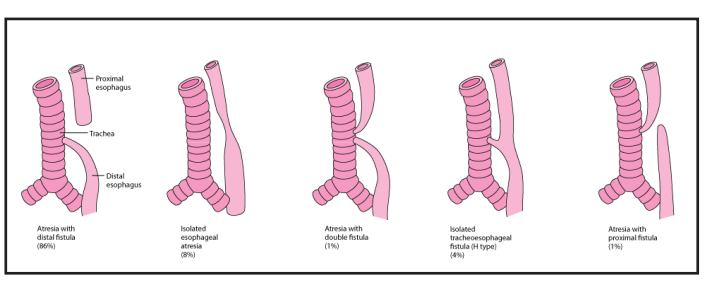
RESULTS

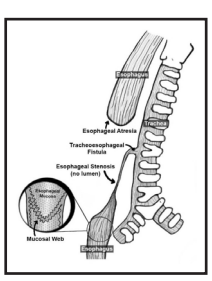
Male newborn BW 1890g, BD 46cm, the Apgar score 6-8-9. Pelvic dystopia of the right kidney was diagnosed prenatally. Hypospadias and anus perinealis ventralis were diagnosed after birth. Esofageal atresia with tracheooesophageal fistula was confirmed by x-ray with the contrast material. Pleural affusion and pulmonary hypertension appeared afetr birth. Immediate transfer to NICU under mechanical ventilation. Echocardiography with the finding partial anomalous pulmonary venous (VPDx to VCSup), small VSD subaort. and patent Ductus Arteriosus. Pericardial effusion wasn’t present after birth. The findings were closed like an atypical form of Scimitar syndrome.
The genetic examination was performed after birth, karyotype 46XY, SNP array normal. The biochemical examination for CDG syndromes was also completed after birth with negative results.

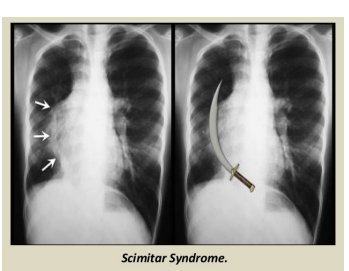
CONCLUSION

Esophageal atresia is a rare birth defect in which a baby is born without a part of the esophagus. The exact cause of esophageal atresia is still unknown but it appears to have some genetic components. Up to half of all babies born with esophageal atresia has one or more other birth defects, such a other digestive tract problems, kidney and urinary tract problems, heart defects or sceletal problems. Diagnosis occurs prenatally but in more cases (like this case) at birth. Scimitar syndrome is a rare congenital disorder.
The syndrome consists of an anomalous pulmonary venous return in which the right pulmonary vein is not connected to the left atrium but to the inferior vena cava.
Scimitar syndrome is named after the crescent-or Turkish sword-like shadow in the chest radiography, it is often associated with hypoplasia of the right lung and pulmonary artery and dextroposition of the heart. It has a varied presentation, from asymptomatic, severe pulmonary hypertension and/or heart failure.
Even though the patient did not accept an invasive examination prenatally, the karyotype and kardioarray CGH did not show any pathology after birth.









Scimitar Syndrome.