Case Findings: Newborn male CXR with cardiomegaly, omphalocele, presumed esophageal atresia as NGT unable to be advanced, L humeral hypoplasia, and hypoplastic vertebral bodies. Patient was then transferred to CHLA for further care.

VACTERL

A defective mesodermal development during embryogenesis before the 35th day of gestation. Non random association of anomalies (3 or more).

Etiology: genetic factors probably not usually involved. Rare familial cases (<1%). Maternal risk factors include diabetes. Associated fetal exposures include lead, lovastatin, dibenzepin, or exogenous sex hormones.

Epidemiology: 1.6:10,000 births. MC in causasian boys. 1/3 are premature, and 13% stillborn.

Differential DX:
- Trisomy 13
- Trisomy 18
- PHAVER syndrome
- Holt-Oram
- Thrombocytopenia absent radius syndrome
- Pseudothalidomide syndrome
- Jarcho-Levin syndrome
- VACTERL-H (Briard Evans syndrome)

Clinical and Radiological Findings (high variability):

Prenatal presentation with polyhydramnios or single umbilical artery. 28% neonatal mortality. 48% mortality in 1st year of life. Intelligence usually normal. Omphalocele associated in 6%. Best clue is vertebral anomaly in presence of other malformation.

Vertebral anomalies (37%): vertebral fusion, hemivertebrae, butterfly vertebrae, segmentation defects, caudal regression, partial or completely absent sacrum

Anal atresia (63%)

Cardiac anomalies (77%) VSD>PDA>ASD, tetralogy of Fallot, transposition.

Tracheoesophageal fistula or esophageal atresia (40%) (¼ have the association), H type fistula.

Renal anomalies (72%): renal agenesis, multicystic dysplasia, hydronephrosis, ectopia, persistent urachus
Limb (58%) (radial array) anomalies: hypoplasia, polydactyly. Other reported skeletal anomalies include humeral aplasia/hypoplasia, rib anomalies, and Sprengel deformity.

Other reported abnormalities: ocular, genital and gonadal anomalies, hydrocephalus and absent corpus callosum, and multiple respiratory anomalies.

REFERENCES:


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Lachman, R.  Taybi and Lachman’s radiology of Syndromes, Metabolic Disorders and Skeletal Dysplasias