

## Pulmonary hypoplasia in an infant with VACTERL association: A case report

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**Abstract:** VACTERL association is a set of congenital anomalies occurring in 1 in 10,000 to 1 in 40,000 infants. Other defects in conjugation with VACTERL are known to occur rarely. Lung hypoplasia in infants with VACTERL poses a unique challenge, as these patients have reduced pulmonary reserve and high susceptibility to chest infection owing to cardiac defects and tracheoesophageal fistula. These infants have poor quality of life and delay in diagnosis further adds to morbidity. Here we are presenting a case of VACTERL with lung hypoplasia, diagnosed owing to high suspicion by treating team.

**Key words:** VACTERL, Lung hypoplasia.

Date of Submission: 10-04-2020

Date of Acceptance: 24-04-2020

### I. Background:

VACTERL association is a set of congenital anomalies involving multiple systems in body. It includes, vertebral anomalies, anal atresia, cardiac defects, tracheoesophageal fistula (TEF), renal and limb abnormalities(1). Diagnosis requires any 3 of the preceding anomalies to be present in a patient. Its incidence varies from 1 in 10,000 to 1 in 40,000 infants(2). Chromosomal micro-aberrations and familial tendencies of certain defects have been observed on rare instances(3). Rarely, there may be other abnormalities seen in patients along with this association, such as caudal regression, airway abnormalities, lung hypoplasia or urogenital septal abnormalities(4-7). Here we are presenting a rare case of VACTERL association with pulmonary abnormalities.

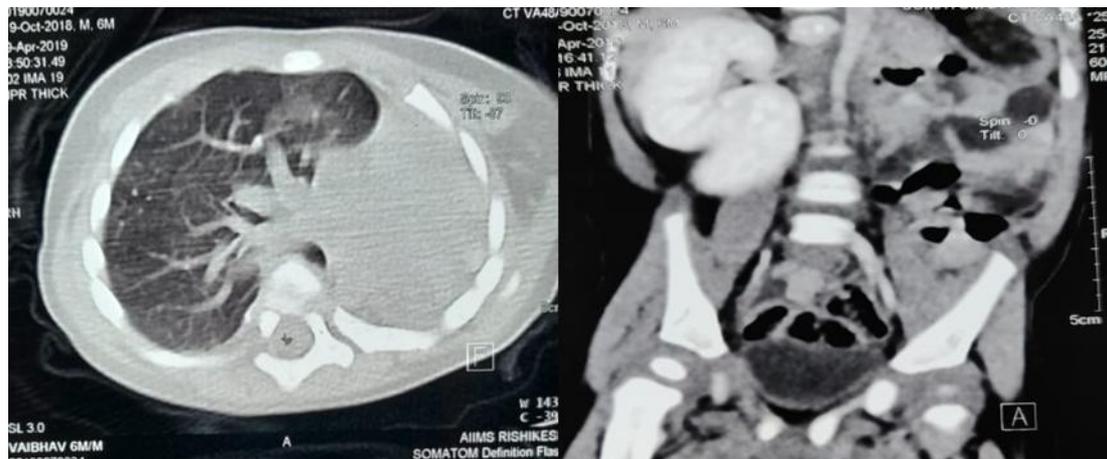
### II. Case Report:

6-month-old male child presented with complaints of fever on and off for past 1 month associated with noisy breathing. Despite seeking treatment on OPD basis from various doctors, fever spikes persisted and patient increasingly became sick. He was admitted at nearby hospital, where he received IV antibiotics. Chest x ray done at this center revealed a homogeneously opaque left side of chest, and hence he was referred to our center for further management. There was no history of tubercular contact or blood transfusions however, patient had been hospitalized 2 times for chest infections. Birth history is non-contributory, immunization is complete for age and development is normal for age.

On examination, patient was febrile (temperature 103°F) and in respiratory distress (PR- 130/min, RR- 54/min, SpO<sub>2</sub>- 97 % at room air). On general physical examination, other than pallor there was no other significant finding. Systemic examination revealed tachypnea, subcostal, intercostal and suprasternal retractions, nasal flaring, reduced expansion and decreased air entry on left side. On right side, entire chest field was full of crepitations and conductive breath sounds. CNS examination was unremarkable apart from irritability. Per abdomen examination was unremarkable except for umbilical hernia (0.5mm diameter defect, reducible). CVS examination was within normal limits.

After initial supportive management (supplementary oxygen, empirical antibiotics and IV fluids) and stabilization in emergency, patient was shifted to pediatric ward. Initial investigations revealed a TLC of 19750 cells/cumm, with neutrophilic predominance, mild anemia (Hb 9.3 g/dl, MCV- 76.2 fl, MCH- 24.1 pg, MCHC- 31.6 g/l), normal platelet count and elevated CRPH. Chest radiograph was obtained which showed opacity on left side with mediastinal shift to left side and hyper-inflated right lung. CT thorax with pulmonary angiography was done which showed left lung aplasia with left main bronchus ending into a blind pouch with absent left pulmonary artery. Vertebral anomalies were also noted- D4, D5 and D6 were butterfly shaped (hemivertebrae).

With these new findings a syndromic diagnosis was suspected and further investigations were done. USG whole abdomen revealed left sided crossed and fused renal ectopia, which was confirmed by CT urography. Cardiac evaluation through 2D ECHO revealed a 3mm PDA with PFO (left to right shunt), with good biventricular function and no evidence of PAH.



**Figure 1:** CT scan of thorax and abdomen showing left sided pulmonary hypoplasia with compensatory hyperinflation on right side and left ectopic kidney.

Patient responded to antibiotic therapy and symptomatically improved. Oxygen supplementation was gradually tapered and removed entirely by day 10 of admission. Parents were counselled regarding patient's condition and possibility of multiple chest infections, renal complications that could arise in future, and advised regular follow up.

### III. Discussion:

On account of multiple system involvement, VACTERL can have debilitating limitation on quality of life. This is further accentuated by delay in diagnosis and lack of awareness and follow up with appropriate specialist. Nearly 25% of malformations are identified late into childhood, and in over 50% of patients, the diagnosis of VACTERL association is made only in adulthood(8). It is also observed that these patients have short stature, more incidence of end stage renal disease (ESRD) and worse transplant outcomes as compared to general population(9). Patients with anal atresia and limb defects have higher morbidity. Our patient had congenital heart disease (with left to right shunt) along with left lung hypoplasia. Hence, he is especially vulnerable to chest infections. Such patients should be immunized against pneumococci, meningococci, and influenza. With high risk of recurrent infections and renal abnormalities, these patients are also at risk of growth failure. As such, significance of proper nutritional assessment and rehabilitation cannot be overemphasized. Apart from ensuring a balanced diet and proper feeding practices in pediatric age group, supplementation of micronutrients like Vitamin A, Vitamin D and Zinc should be mandated. With vertebral anomalies, these patients are at higher risk of developing scoliosis, abnormal gait and restrictive lung disease. Thus, regular spinal and gait examination should be done without fail at each visit. Regular follow up, hand hygiene, regular assessment of pulmonary, cardiac and renal function is the key to provision of a better quality of life. Parents need to be counselled about the disease and prognosis.

### IV. Conclusion:

VACTERL association is a relatively rare finding in clinical practice and as such clinicians should have a high index of suspicion for early diagnosis. Association of pulmonary hypoplasia is a very rare finding. Pulmonary hypoplasia increases the morbidity. Children with this finding are more vulnerable and at higher risk of respiratory failure in event of respiratory infection. Intrauterine diagnosis by ultrasonography provides option of termination of pregnancy. Early diagnosis can improve outcomes by surgical intervention before development of complications(10). Regular follow up, family counselling cornerstones of management.

**Funding:** The authors received no funding/grant from any external agencies.

**Conflict of Interest:** The authors declare that they have no conflict of interest.

**Ethical approval:** All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional research committee (Institute ethics committee vide letter no.

AIIMS(R)- ethics/1995) and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards.

**Consent to participate:** Written and informed consent were taken from the patient and his family members for using his individual and clinical data for publication and research purposes.

**Availability of Data and Materials:** Data sharing not applicable to this article as no datasets were generated or analyzed during the current study.

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Anchal Kumar Tripathi, et al. "Pulmonary hypoplasia in an infant with VACTERL association: A case report." *IOSR Journal of Dental and Medical Sciences (IOSR-JDMS)*, 19(4), 2020, pp. 18-20.