VATER Association with Multiple Ribs Anomalies

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Abstract

VATER association is an exceptionally rare condition; however it is associated with multiple rib anomalies, which is one of its unique presentation. We are reporting a case of VATER associated with rib anomalies in various forms like bifid rib, ribbon rib and rudimentary rib.

Introduction

VATER association (V-Vertebral defect, A-Anal atresia, C-Cardiac anomalies, TE-Tracheo-oesophageal fistulae, R-Renal anomalies, L-Limb anomalies) is an acronym which has been used to describe above mentioned characteristics. There should be at least three or more of these anomalies together to call it VATER.² It is a rare combination (estimated incidence is 1.6 cases per 10,000 live birth). Regarding etiology, no specific genetic or chromosomal anomalies has been seen with VATER association, although it is shown to be associated with some chromosomal defect such as trisomy 18 and is more frequent in infants born to diabetic mother.¹

Case Report

A 17 years old female presented with breathlessness, palpitation and restlessness for last two days which was aggravated by cough and associated upper respiratory tract infection. Since childhood, she had similar type of episodes intermittently. There was no history of hemoptysis, squatting episodes, cyanotic spells, seizures or loss of consciousness. Her mother was non-diabetic and had no history of any drug intake during pregnancy.

On examination, the patient was short statured (142 cm) without any dysmorphic facies, conscious, oriented with normal intelligence and learning ability. She was mildly dyspnoeic (respiratory rate-32/minute, regular) with blood pressure of 110/72 mmHg, pulse rate of 86/minute, normal jugular venous pressure and without clubbing. She had SPO² of 96%. Clinical examination disclosed pectus excavatum, precordial bulge and laterally shifted apical impulse with systolic thrill at left parasternal space. Cardiac auscultation revealed pansystolic murmur at lower left sternal border, loud P₂ with physiological split. Rest of systemic examination was normal.

Investigations

Complete hemogram revealed: Hemoglobin- 11.5 gm%, total leucocyte count- 7,300/cu mm, differential leucocyte count- N66, L30, E2, M2 and platelets- 2.3 lacs/ mm³. Routine and microscopic examination of urine were normal. Renal and liver function tests revealed no abnormality. Serum electrolytes were within normal limits.

Electrocardiogram showed biventricular hypertrophy with positive Katz Wachtel phenomenon.

X-ray dorso-lumbar spine AP view showed: (Fig. 1)
- Right to left thoraco-lumbar scoliosis
- Reduced disc space between T₉ to T₁₁
- Hypoplastic L vertebræ and destruction of upper margin of L₂
- Left sided rudimentary rib on L₁
- Left sided elongated transverse process on L₅
- Right sided bifid 12th rib.

X-ray chest AP view showed: (Fig. 2)
- Extra numerary ribbon ribs at the level of T₃-T₄ on right side
- Malformed rib at the level of T₄-T on left side
- Bifid 12th rib on right side.
- Cardiomegaly

2D and color Doppler echocardiography showed large perimembranous ventricular septal defect with left to right shunt, severe pulmonary arterial hypertension, with normal biventricular function.

Ultrasonography of abdomen showed left sided renal agenesis.⁴ (Fig. 3)

Discussion

VATER association is a non-random association of birth defects without specific genetic or chromosomal problem. This condition can be seen with some chromosomal defects such as Trisomy 18¹² and is more frequently seen in babies of diabetic mother, and those who had taken cholesterol and lipid lowering drugs (statins) in first trimester of their pregnancy.² A defect in blastogenesis has been suggested as a possible etiology. There are some other syndromes which resemble VATER association like Feingold syndrome, CHARGE syndrome, Townes-Brocks syndrome, 22q11 deletion syndrome and Pallister-Hall syndrome which show some phenotypic overlap with the VATER association.

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Most of the patients of VATER association have congenital heart disease (75%) commonly-ventricular septal defect, atrial septal defect, tetralogy of Fallot. Next common anomaly is vertebral anomalies (70%) like small (hypoplastic) vertebrae and hemivertebrae. Anal atresia or esophageal atresia with tracheo-esophageal fistula is seen in about 55-70% of patients with VATER association. Renal defects are seen in approximately 50% of patients with incomplete formation of one or both kidneys, or urologic abnormalities such as obstruction to outflow of urine or severe reflux of urine. Limb defects occur in up to 70% of babies that include absent or displaced thumbs, extra digit (polydactyly) or fusion of digits (syndactyly). These anomalies are usually noted at birth and require surgical intervention.

Other congenital anomalies which are rarely seen with VATER association are growth abnormalities genital defects, small intestinal atresia, diaphragmatic defects, oral clefts, bladder exostrophy, omphalocele and neural tube defects.

We are presenting this unique case of VATER association with rib anomalies, which is rarely reported. This patient had rare association of rib anomalies in various forms like bifid rib, ribbon rib and rudimentary rib.

References