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A Rare Congenital Anomaly Involving Skeletal, Cardiac and Genitourinary System

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Abstract: We present a rare case of association of congenital anomaly that involves skeletal, cardiac and genitourinary anomalies in 10 month old female child. It is a rare entity with conglomeration of a lot of congenital anomalies. Association of absent ribs, renal agenesis, ovarian agenesis, ostium secundum and supernumary nipples could not be matched with any syndrome using morphologic database.
Keywords: Congenital anomalies, Supernumary nipples, Renal agenesis.

INTRODUCTION

Congenital anomalies (CA) are defined as structural or functional abnormalities including metabolic disorders, present at birth. These anomalies of prenatal origin occur due to defective embryogenesis or intrinsic abnormalities in the process of development and are an important cause of neonatal and infant morbidity and mortality [1, 2].

Supernumerary nipples are common minor congenital malformations. They consist of nipples and/or related tissue in addition to the nipples on the chest. Supernumerary nipples are found to be located along the embryonic milk lines. The ectopic supernumerary nipples are present beyond the embryonic milk lines [3].

CASE REPORT

A 10 month old female child, born out of non-consanguineous marriage was admitted with fever, cough and respiratory distress. She was born at term with birth weight of 2.3 kg and had history of multiple similar episodes since birth.

On examination, it was found that there was a depression over left side of chest with bilateral wheeze. There was also an ejection systolic murmur over left parasternal region. Detail clinical examination revealed microcephaly (39 cm), cleft palate, supernumary nipples (Fig. 1.), and left sided lipomatous swelling over scapular region (Fig. 2) and hepatomegaly. There was no facial dysmorphism.

Chest x-ray showed absent 4th, 5th and 6th ribs on left side (Fig. 1), bifid cervico-dorsal spine with upper dorsal block and hemivertebra. Echocardiography revealed a large ostium secundum atrial septal defect. Ultrasonography of whole abdomen revealed left sided renal agenesis and left sided ovarian agenesis.

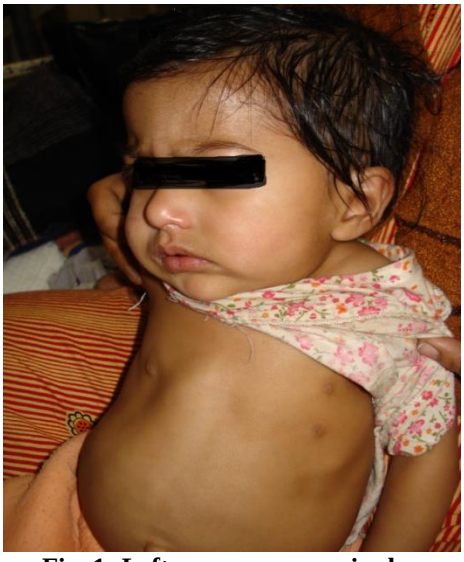

Fig. 1: Left supernumary nipples

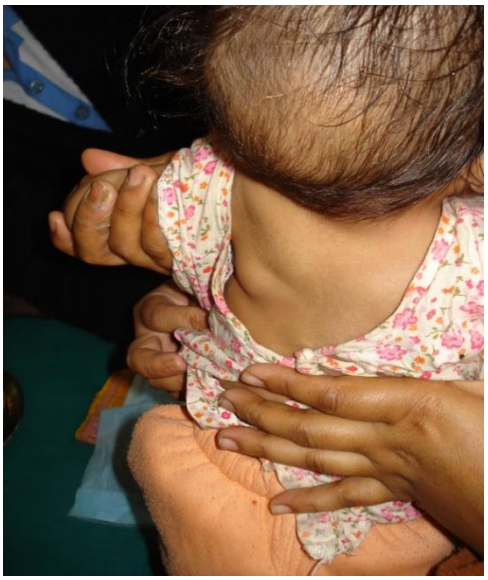

Fig. 2: Lipomatous swelling over left scapula

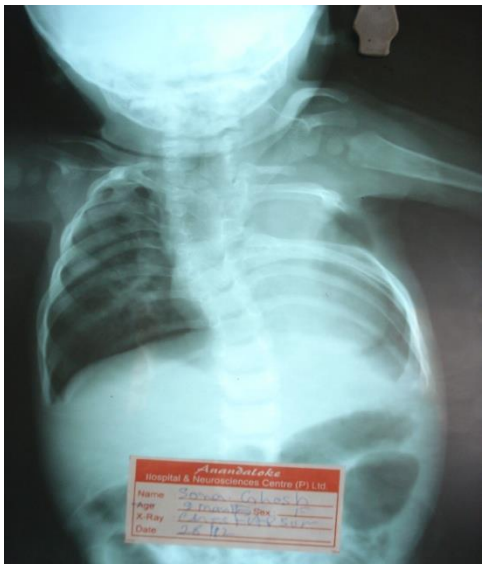

Fig. 3: Absent ribs on left side

DISCUSSION

The embryonic milk line extends bilaterally from beyond the axillae on the arms, down the chest and the abdomen toward the groin reaching the inner sides of the thighs [3, 4].

Supernumerary nipples appearing with breast tissue and ducts are called polymastia [3].

Supernumerary nipples have been found in various syndromes like Turner syndrome, Fanconi anemia, ectodermal dysplasia, Kaufman-McKusick syndrome and Char syndrome [5]. Association of supernumery nipples and renal involvement have been reported by many authors [6-11]. These include renal malformations, urinary tract anomalies, renal agenesis and renal adenocarcinoma. Association with cental nervous system (neural tube defect, developmental delay, epilepsy etc), gastrointestinal system (peptic ulcer, pyloric stenosis), respiratory system (laryngeal web, ear abnormalities, accessory lung lobe), skeletal (vertebral anomaly, absence of rib, hemihypertrophy, arthrogryposis, scalp defects, microcephaly etc) and cardiovascular system (patent ductus arteriosus, atrial septal defect, ventricular septal defect, conduction defect etc.) have also been notified [3].

Congenital unilateral renal agenesis have been frequently associated genitourinary anomalies like vesicoureteral reflux, ureterovesical junction obstruction, ureteropelvic junction obstruction, unicornuate or bicornuate uterus, uterus didelphys, double or absent vagina, absent or hypoplastic ovary, absent fallopian tube, persistent Gartner's duct cyst, and abnormal external genitalia [12-15].

CONCLUSION

Our case is a rare entity with conglomeration of a lot of congenital anomalies. Association of absent ribs, renal agenesis, ovarian agenesis, ostium secundum and supernumary nipples could not be matched with any syndrome using morphologic database. Such a rare association has not been reported in any world literature to the best of our knowledge.

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