Right upper limb bud triplication and polythelia, left sided hemihypertrophy and congenital hip dislocation, facial dysmorphism, congenital heart disease, and scoliosis: Disorganisation-like spectrum or patterning gene defect?

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Abstract

A Somali female baby with right upper limb triplication, polythelia, left sided hemihypertrophy, congenital hip dislocation, facial dysmorphism, congenital heart disease, and scoliosis is described. It seems that the above described pattern of anomalies has not been reported before. The possible developmental genetic mechanism responsible for this phenotype is briefly discussed.

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Several patients with complete or partial duplication of a lower limb have been reported.¹⁻¹² In this report, we describe the first case of right upper limb triplication, polythelia, left sided hemihypertrophy, and congenital hip dislocation associated with facial dysmorphism, congenital heart disease, and scoliosis.

Case report

The proband, a female born at term on 7.7.94 with multiple congenital anomalies, was delivered by lower segment caesarian section. Apgar scores were 7 and 8 at one and five

minutes. Pregnancy history was unremarkable. The healthy, young, consanguineous parents have six phenotypically normal daughters with no family history of any congenital anomalies. Birth weight was 3360 g, length 50 cm, and head circumference 35 cm. Placental weight was 750 g and the umbilical cord contained two arteries and one vein. The baby had facial dysmorphism with frontal bossing, left microphthalmia, left microcornea, bilateral iris colobomata, depressed nasal bridge, midfacial hypoplasia, down turned upper lip, high arched palate, short neck, and right torticollis. She had right upper limb triplication including two hypoplastic supernumerary limbs with oligodactyly and an absent thumb on one of them. (fig 1). Right sided polythelia without underlying glandular tissue was present. The right lower limb was shorter than the left with a small right foot and overlapping hypoplastic toes. Two hairy darkly pigmented patches were noted, one on the dorsum of the right foot and the other on the upper part of the back of the forearm of a supernumerary limb. The baby had left hemihypertrophy, scoliosis, and left congenital hip dislocation. The external genitalia were normal female. No CNS, kidney, or other internal organ abnormalities were detected with ultrasound. Skeletal survey showed three right upper limbs including the bones of the shoulder girdle (clavicles and scapulae) and scoliosis (fig 2). Chest x ray showed clear lungs with normal heart size. ECG was normal but echocardiography with Doppler studies showed mild PDA and ASD with a small left to right shunt and mild pulmonary hypertension. Peripheral blood chromosomal study with banding techniques showed a normal female karyotype (46,XX) with no apparent mosaicism. Clinical follow up at the age of 3 months indicated a satisfactory neurodevelopmental status.

Discussion

Limb duplications have traditionally been thought of in terms of incomplete separation

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Figure 2 Skeletal survey of proband showing the bones of three right upper limbs and scoliosis.

report may be the result of a de novo somatic mutation of a pleiotropic gene of the patterning families implicated during early embryogenesis, for example, in limb bud mesoderm/ectoderm signal interaction¹³ and in the specification of the branchial region of the vertebrate head.¹⁴ Alternatively, the present case could fit into the spectrum of the human equivalent of the mouse "Disorganisation locus, DS" (MIM 223200) described in several previous reports.¹⁵⁻¹⁷

It seems that the presence of two supernumerary right upper limbs, polythelia, left

hemihypertrophy, and congenital hip dislocation, associated with facial dysmorphism, congenital heart disease, and scoliosis, has not been described before. However, we cannot claim that the present case represents a new syndrome since phenotypic heterogeneity would be expected among different cases with limb duplications, if the abnormalities were caused by a somatic mutation with consequent mosaic patterns.

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