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?

M A Sabry, Q Al-Saleh, R Al-Saw'an, S A Al-Awadi, T I Farag

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.

Case report
Several patients with complete or partial duplication of a lower limb have been reported. 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.

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


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