Images in Clinical Practice

Robinow Syndrome

An 8-year-girl, a product of consanguinity, presented with short stature. She had mesomelic dwarfism specially in upper limbs with small hands and short fingers with clinodactyly. The facies was characterized by large head, frontal bossing, hypertelorism, wide palpebral fissure, short upturned nose with anteverted nares, long philtrum, triangular mouth and small chin. (*Fig. 1*). Radiological survey revealed hemivertebrae in mid thoracic region and fusion of few ribs, bifid terminal phalanges in hands. A diagnosis of Robinow syndrome was made.

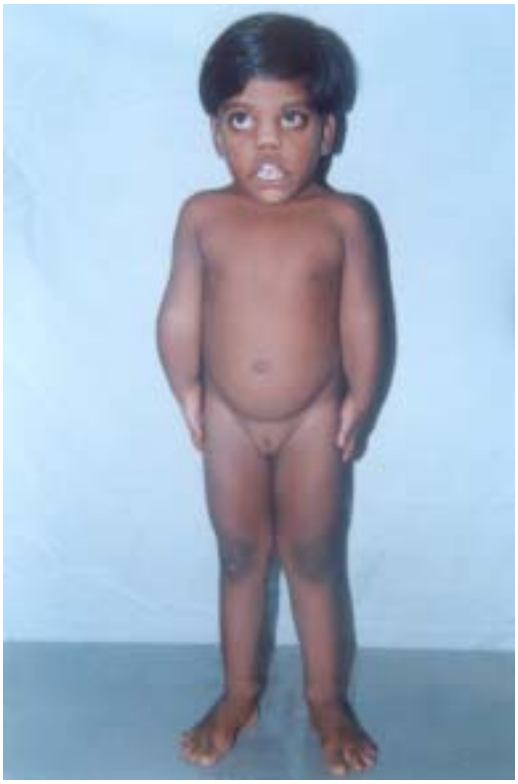

Fig. 1. Short stature, mesomelia specially of upper limbs and characteristic facies in Robinow syndrome.

Robinow syndrome is a mesomelic short limbed dwarfism characterized by abnormal morphogenesis of face, hypoplastic genitalia (small or absent penis, hypogonadism, cryptorchidism, hypoplasia of clitoris and labia minora) and skeletal anomalies. Duplication of kidney and hydronephrosis are occasionally present. Cardiac anomalies constitute part of the syndrome and include right ventricular outflow tract obstruction. Developmental delay and mental retardation occur in about 18% of cases.

Early death secondary to pulmonary and cardiac complication occur in 10 per cent of patients. The gene for autosomal recessive type of Robinow syndrome is located at chromosome 9q 21-23 region.

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