Robinow Syndrome (Foetal Face Syndrome) with Radiological Mesomelic and Rhizomelic Brachymelia of the Lower Limbs: A Case Report

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Summary:

Robinow syndrome was diagnosed in a newborn baby on the basis of clinical and radiological evidence. The syndrome in a rare entity and reportedly the first cure diagnosed in this country.

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Introduction :

Robinow syndrome is a rare inherited disorder. It was described by Robinow in 1969 as a syndrome of characteristic facies, micropenis, cryptorchidism in the male and mesomelic limb shortening[1]. The characteristic facial appearences (foetal face) are frontal bossing, macrocephaly, hypertelorism, wide palpebral fissures with downward slanting, short upturned broad nose with anteverted nares, hyperplastic alveolar ridges, long philtrum, small chin and triangular mouth with downward angles. In addition, hypoplastic genitalia, nevus flammeus, capillary haemangioma on the glabella, mesomelic brachymelia of forearm and small hands with clinodactyly are also noted[2]. The inheritance pattern in Robinow syndrome is not clear. Both autosomal dominant and recessive inheritance have been reported[3-6]. It has been suggested that more severe mesomelic brachymelia and more triangular mouth are indicative of autosomal recessive inheritance[7]. A newborn is described here with radiological rhizomelic and mesomelic brachymelia of the lower limbs. Probably cases with such diagnosis have not been reported before in this country.

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Case report :

A just born male baby, third issue of a consanguineous parents was delivered by caesarean section at term with breech presentation on third September, 2003 at Maternal and Child Health Training Centre (MCHTI), Azimpur. Antenatal history was uneventful. There was no history of the similar problem in the family. On routine examination, the birth weight of the baby was 3000 gm, length was 49 cm and occipitofrontal circumference (OFC) was 38 cm which fell on + 2SD. His face was dysmorphic characterized by prominent eyes with downward slanting and wide palpebral fissures, open posterior fontanelle, macrocephaly, low set ear, triangular mouth with downturned angles, long philtrum, glabeller haemangioma on the central part of the forehead, nevus flameus on the occiput and loose skin around the neck. (Figures 1 and 2). There was micrognathia. Cryptorchidism was evident on the left side (Fig.-3). Limb shortening was not obvious. On the basis of clinical features the baby was diagnosed as a case of Robinow syndrome.

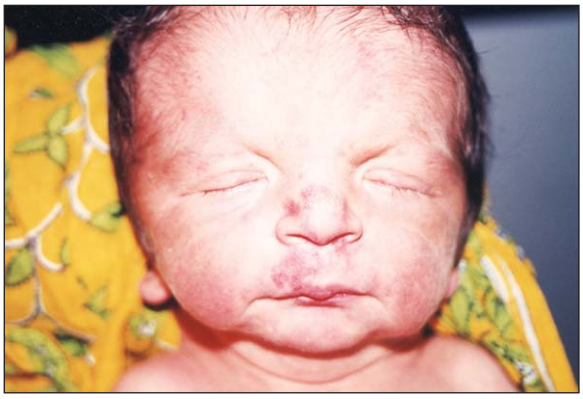

Figure-1 : *Showing downward slanting of palpebral fissure.*

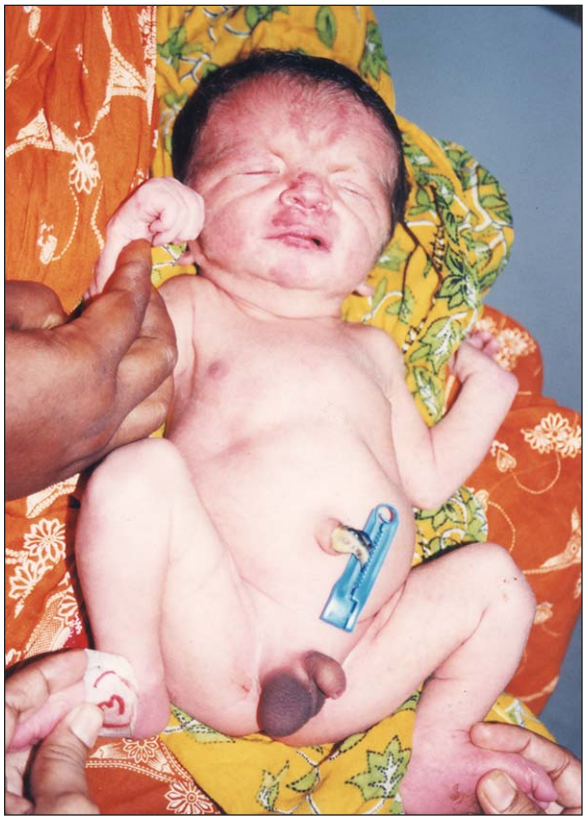

Figure-2 : *Showing glabellar haemangioma on the central part of the head.*

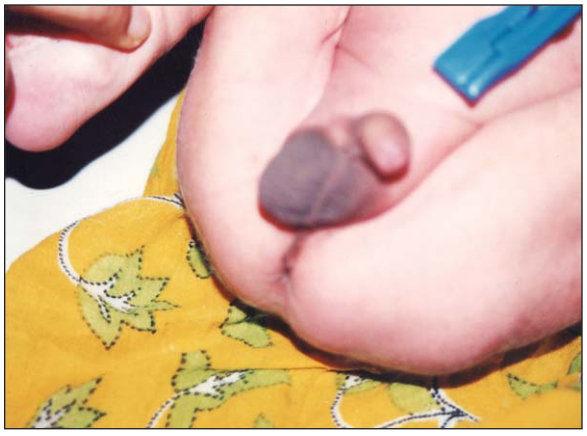

Figure-3 : *Showing left sided cryptorchidism.*

On investigation, his haemoglobin level was 12 gm/dl and random blood sugar 5.7 mmol/litre. Test for VDRL and HBsAg were negative. X-Ray limbs showed short tibias and femora (Fig.-4). Both rhizomelic and mesomelic shortening were found in the lower limbs. With above clinical and radiological findings the diagnosis was established as Robinow syndrome. His postnatal period was uneventful till the second week of life. In follow up visit at the age of three weeks, he was found to have umbilical granuloma and was treated accordingly.

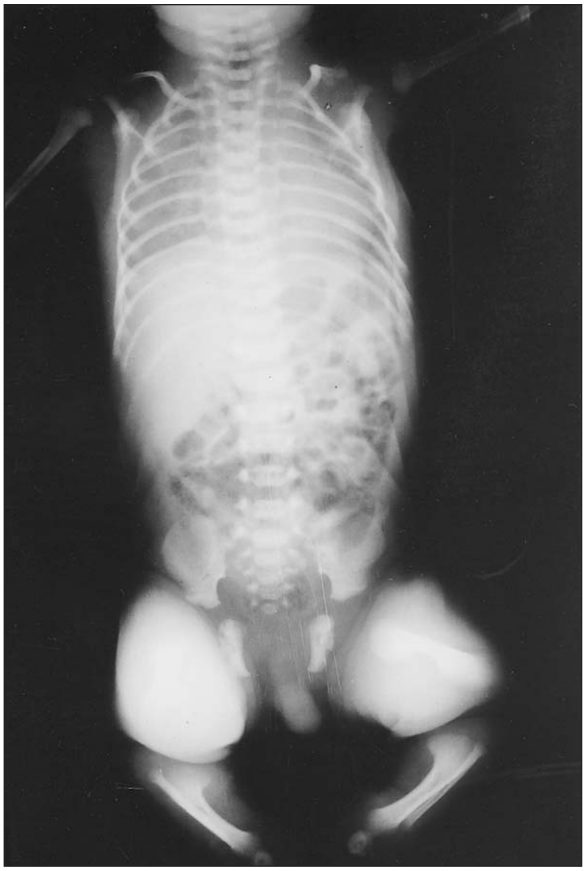

Figure-4 : *X-ray showing rhizomelic and mesomelic braclymelia of the lower limbs (short fibia and femora).*

Discussion:

Langer has divided bony defects of the extremities into four general types: acromelia or distal shortening, mesomelia or shortening of the mid segment, rhizomelia or proximal shortening and proportionate shortening of all segments of the limb [8]. Some of these disorders involve defective growth of both the tubular bones or the axial skeleton or both, and are recognizable at birth. The disorder first described by Robinow et al[1] is evident at birth. So far, over 100 cases have been reported. The gene for the autosomal recessive form was identified as the ROR2 gene on chromosome 9q22. The same gene, ROR2

has been shown to cause autosomal dominant brachydactyly.[9] Along with bony defects the peculiar facies and the genital abnormalities serve to differentiate this condition from other dwarfing syndromes. Affected individuals with Robinow syndrome tend to have short stature but their weight and length may be within normal range at birth[10]. Short stature may appear by three to four years of age[10]. The present case had normal length and weight at birth.

Mesomelic brachymelia was considered almost essential to diagnose Robinow syndrome. Mesomelic shortening of the forearms is one of the cardinal features of the condition but the pattern of limb shortening can be extremely variable. There may be rhizomelic brachymelia[11]. In this case, there was rhizomelic and mesomelic brachymelia. The upper limb deformities were found in the cases described by Wadia et a1[12]. In this case, there was lower limb deformities without any involvement of upper limbs.

The cardinal features of Robinow syndrome are slight to moderate shortness of stature at the postnatal onset, frontal bossing, hypertelorism, macrocephaly, shortening of forearm (mesomelic brachymelia), genital abnormality and nevus flammeus[13,14]. But shortening may be found in lower limbs. Rhizomelic brachymelia (shortening of proximal part of the lower limb) can be a part of this syndrome. These mesomelic and rhizomelic brachymelia may not be evident at birth clinically.

Achondroplasia may mimic this syndrome. Clinically, normal interorbital distance and trident feature in hands is seen in achondroplasia. Radiologically achondroplasia differs from Robinow syndrome by the presence of caudally decreasing lumber interpediculer distances and lordotic lumbosacral angle with shortened sciatic notch[1]. Once the Robinow syndrome is suspected clinically, radiological evaluation is to be carried out to establish the diagnosis.

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