

A Case of Robinow syndrome

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Introduction

Robinow syndrome is a rare autosomal recessive and autosomal dominant disorder affecting the whole skeletal system. Autosomal recessive robinow syndrome is characterized by shortness of the long bones and vertebra anomalies. Less than 200 cases of autosomal recessive robinow syndrome have been reported in the literature.

4 days



History: The newborn was referred to pediatric endocrine clinic for disorder of penis development. He was born 3000 grams by cesarean section at the 32nd gestational week. Family history revealed first degree cousin marriage.

Physical examination: Weight: 4000 grams (0.46 SDS), length: 47 cm (-1.62 SDS) and head circumference: 39 cm (1.58 SDS). On his physical examination, macrocephaly, broad forehead, low set ears, prominent and widely spaced eyes, short nose with upturned tip, broad and triangle-shaped mouth, overgrowth of gums, abnormal short fingers and toes were detected. His genital examination revealed absence of penis, scrotalization occurred and the testes were in scrotum. Scrotal hyperpigmentation was not detected for adrenal diseases.

Clinical Course: Whole blood count, biochemistry analysis and thyroid function tests were normal. Serum FSH, LH and total testosterone levels were found to be compatible with minipuberte. Skeletal radiography showed fusion of hemivertebrae and ribs. Peripheral chromosome analysis revealed 46 XY. Homozygous IVS2-1G> C (c.176-1G> C) mutation was detected in the ROR2 gene in the long arm of the ninth chromosome by using the whole gene sequence analysis for Robinow syndrome.

Conclusion

Robinow syndrome should be considered in infants with mesomelic shortness, brachydactyly, craniofacial dysmorphic findings, gingival hypertrophy and undeveloped genitalia. Aarskog syndrome, I-cell disease, Omodysplasia and Jarcho-Levin syndrome should be considered in the differential diagnosis.

