MEIER-GORLIN SYNDROME - A VERY RARE CONGENITAL MALFORMATION

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ABSTRACT

The Meier-Gorlin syndrome or ear, patella, short stature syndrome (MIM 224690) is a rare autosomal recessive disorder. It is characterized by severe intrauterine and postnatal growth retardation, microcephaly, bilateral microtia, and aplasia or hypoplasia of the patella. Despite the presence of microcephaly, intellect is usually normal (1). This case study discusses a case on Meier-Gorlin Syndrome.

INTRODUCTION

The Meier-Gorlin syndrome is a rare autosomal recessive disorder. It was first described by Meier and Rothschild (2) and the second case reported by Gorlin et al. 1975 (3) so named after the two. While almost all cases have primordial dwarfism with substantial prenatal and postnatal growth retardation, not all cases have microcephaly, and microtia and absent/hypoplastic patella (4).

CASE REPORT

A newborn born by nonconsanguineous marriage to phenotypically normal parents, He was born normally at 37-week gestation and weighed 2.1 kg, small for his gestational age, antenatal history was not significant. On examination, He had microtia and microcephaly with his head circumference of 30 cm and length being 46 cm at birth, metopic sutural prominence and hyper extensibility of joints with typical nose, which is more prominent and narrow, with a convex in profile view. On day 4th of life he had systolic murmur but no signs of congestive cardiac failure. No cyanosis and spo2 was maintained and other systems were within normal limits. Routine blood investigations were normal. 2 D ECHO showed, small VSD.
DISCUSSION
The Meier-Gorlin syndrome is a rare autosomal recessive disorder. It was first described by Meier and Rothschild and the second case reported by Gorlin et al. 1975 so named after the two. Cohen et al. (1991) used the designation ear, patella, short stature syndrome (EPS) for a condition they observed in 2 sisters who had bilateral microtia, absent patella, short stature, poor weight gain, and characteristic facial features. Other skeletal anomalies included complete habitual dislocation of the elbow, slender ribs and long bones, abnormal modeling of the glenoid fossa with hooked clavicles, and clinodactyly. Bone age was significantly delayed and there was flattening of the epiphyses. Hurst et al. (1988) found 2 males with similar characteristics. Few studies have documented variable results of endocrine work-up, including growth hormone assays. Loeys et al. [1999] reported two brothers with MGS, delayed bone age, one of whom was subjected to glucagon stimulation test with subnormal GH and borderline Somatomedin C. Bongers et al. (2001) reported 6 female and 2 male patients from 7 families with Meier-Gorlin syndrome and reviewed the literature on this condition. Most of their patients had bilateral small ears, patellar aplasia/hypoplasia, and short stature, except for monozygotic twins who had normal patella on physical examination. Radiographic studies of the patellae were recommended in patients with this condition to understand the patellar abnormality better.

Guernsey et al 2011(9) had done genetic work-up of 45 individuals with MGS and found mutations in five genes from the pre-replication complex (ORC1, ORC4, ORC6, CDT1, and CDC6), crucial in cell-cycle progression and growth. In our case, physical characteristics like microtia, typical nose, microcephaly and IUGR are present. Which is reported in many cases (10). But we got VSD in addition to typical features.

REFERENCES
