

Marshall-Smith Syndrome: A Distinct Entity

A.K. Sharma
A. Haldar
S. Phadke
S.S. Agarwal

Marshall *et al.* in 1971(1) described a syndrome of accelerated osseous maturation, relative failure to thrive and unusual facies. Since then 18 cases have been reported in the world literature(2). In this report we describe a 50 days old female infant with characteristic features of Marshall Smith Syndrome. There has been only one earlier report of this syndrome from our country(3).

Case Report

The patient, a female infant, was born to a 24 years old Hindu woman at 41 weeks gestation. The birth weight of the child was 3.2 kg. She was the first and only child of healthy, unrelated parents. The pregnancy had been uneventful. There was no history of exposure to any known teratogen and the family history was non-contributory. The

From the Department of Medical Genetics, Sanjay Gandhi Post Graduate Institute of Medical Sciences, Post Box No. 375, Lucknow, India.

Reprint requests: Dr. Anita K. Sharma, Assistant Professor, Department of Medical Genetics, Sanjay Gandhi Post Graduate Institute of Medical Sciences, Raebareli Road, Lucknow.

*Received for publication: August 23, 1993;
Accepted: March 31, 1994.*

child cried immediately after birth but developed cyanosis and respiratory distress within the next few hours for which she was hospitalized at a neonatology unit for 24 days.

She was referred to the genetic clinic at 50 days of age because of dysmorphic features and failure to thrive. On examination, facial dysmorphism was apparent in the form of prominent forehead, small triangular face, prominent eyes, depressed nasal bridge, posteriorly rotated low set ears and micrognathia (*Fig. 1*). The sclerae were



Fig. 1. Clinical photograph at age of 50 days showing prominent forehead, ocular protrusion, depressed nasal bridge, micrognathia and failure to thrive.

blue. The tongue was posteriorly placed and respiration was noisy and labored with marked recession of suprasternal, substernal and intercostal spaces. A respiratory grunt was audible and the child was having choking spells with cyanosis.

The total body length was 59 cm (90th centile), upper segment 35 cm, lower segment 24 cm, arm span 56.5 cm, weight 2.2 kg, and head circumference 37 cm (75th centile). The hands and fingers were long and thin and so were the feet and toes, there was clinodactyly of little fingers and Sydney line on both sides. The nails were hyperconvex and great toe was smaller than



Fig. 2. X-ray hand, showing advanced osseous maturation.

the second toe with valgus deformity of distal phalanx bilaterally. The feet were high arched and had a vertical crease. There was bilateral genu varum. The child also had clitoromegaly and neonatal ntitis.

A radiographic skeletal survey at the time of examination revealed a markedly advanced bone age (Fig. 2 & 3). Ossification centres were present for four carpal bones in each wrist and the epiphysis for proximal phalanges had appeared in both hands. There were four tarsal bones in each ankle and the ossification centres for femoral and humeral heads were present. Bone age was assessed to be around 3 years.

The chromosomes were normal. The child expired within a few days at home. Autopsy could not be done.

Discussion

We have reported a patient whose salient features were: (i) advanced osseous maturation; (ii) failure to thrive; (iii) marked respiratory difficulty; (iv) unusual facies in the form of prominent eyes, blue sclera, depressed nasal bridge, micrognathia, large posteriorly rotated ears, anteverted nares and prominent forehead; and (v) increased body length, long and slender limbs, fingers and toes.

All these features are characteristic of Marshall-Smith syndrome (MSS)(2,4). Our patient did not have broad phalanges which are also a constant features of MSS(2). Another clinical entity to be considered in the differential diagnosis is Weaver syndrome(5). This is characterized by accelerated skeletal maturity, infants that thrive too well and have height and weight much above normal, increased bifrontal diameter, prominent finger pads and peculiar facies different from the Marshall Smith facies. Although there has been considerable controversy as to whether the Marshall Smith



Fig. 3. X-ray foot lateral view showing advanced osseous maturation.

Syndrome and Weaver Syndrome are one(1,6,7) or separate entities(4,8), our patient does not have any feature of the Weaver syndrome other than accelerated skeletal maturity which is common to both.

In conclusion, this case confirms the Marshall-Smith syndrome as a recognizable, multiple congenital anomalies syndrome and further delineates its facial and radiological appearance. It also supports this syndrome as an entity distinct from Weaver's syndrome.

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