Tibial Hemimelia-Split Hand/Foot Syndrome with Rare Anomalies

Tibial hemimelia-split hand/foot syndrome (TH-SHFM) is a rare constellation of multiple congenital malformations which includes Ectrodactyly (Lobster claw deformity or Cleft hand/foot) with a wide range of phenotypes of absent tibia/tibial hemimelia or long bone deficiency(1,2). We describe a proposita with these characteristic anomalies in absence of family history of malformations. This was associated with sacral agenesis, anorectal atresia, hemi-vertebra and ventricular septal defect, hither to undescribed.

A 38-week male baby weighing 1980 g was born to a 22-year-old primigravida. The newborn expired 10 minutes after birth. There was no history of consanguinity or exposure to drugs or radiation. Examination revealed a claw like deformity of both hands, phocomelia of right lower limb, a transverse cleft on planter side of left foot, low-set ears, hypoplastic mandible and absence of anal opening. Both hands appeared like lobstar claws (*Fig. 1*).

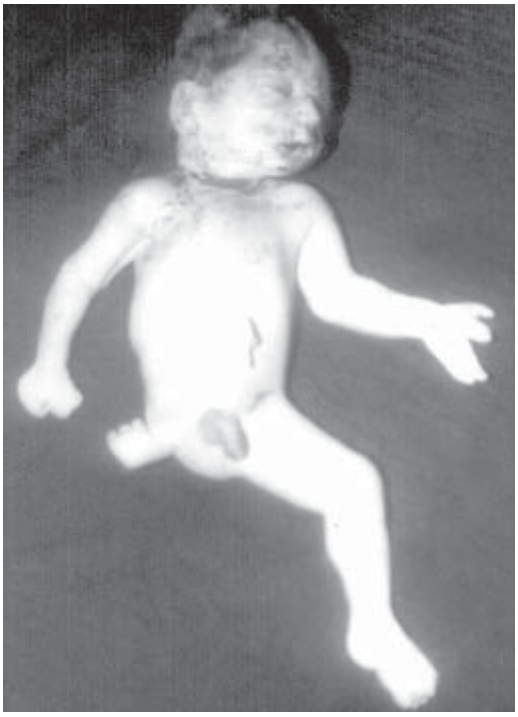

Fig. 1. Claw like deformity of both hands, right sided tibial hemimelia with hypoplastic three toed foot and a transverse cleft on planter side of left foot.

Skeletal survey revealed the absence of the central ray beyond metacarpal with soft tissue median clefts. On the right side the pelvic bones and femur were hypoplastic, tibia and fibula were absent and foot had only three hypoplastic rays. There was hemivertebra at T12 level.

Autopsy confired the above abnormalities. Lungs were hypoplastic and there was a large membranous ventricular septal defect. Karyotyping was 46-XY with G-banding.

The exact prevalence of this rare condition is not known but it has been reported as familial in certain populations. Isolated sporadic cases are also on record(1-3). Malformations in addition to ectrodactyly and bilateral aplasia of the tibias (the full-blown syndrome), include distal hypoplasia or bifurcation of the femurs, hypo- or aplasia of the ulnas, aplasia of the patella, hypoplastic big toes and cup-shaped ears. Mildest visible manifestation is hypoplastic big toes, whereas the severest is tetramonodactyly or transverse hemimelia(1). In a series of 37 patients of TH-SHFM belonging to different families, 15 had tibial hemimelia (9 bilateral, 4 unilateral and 2 with right tibial agenesis and left severe hypoplasia) with foot anomalies ranging from one-toed to 5-toed clubfoot The anomalies ranged from hand or foot anonychia, hand or foot syndactyly and/or preaxial hand polydactyly, to isolated unilateral tibial

agenesis (with or without uni- or bilateral lobster clawhand), to bilateral tibial agenesis and bilateral lobster clawhand(2). Our proposita had unilateral hypoplastic pelvic bones and femur with tibial agenesis, hypoplastic 3-toed foot, and cleft hands. Other associations seen in our case are sacral agenesis, anorectal atresia, hemivertebra and congenital heart disease (CHD).

TH-SHFM associated with fatal pulmonary hypertension and congenital alveolar capillary dysplasia is reported in a consanguineous Turkish family(3). CHD seen in our case was a large membranous ventricular septal defect. Lethal congenital cardiac malformations have been described with "Tibial Hemimelia-Polydactyly syndrome", an entity similar but distinct from TH-SHFM(4). Congenital heart disease has not been reported with TH-SHFM.

Features of Tibial hemimelia-split hand/ foot syndrome, in absence of any family history, in this neonate with anorectal atresia, sacral agenesis, hemivertebra and in particular the CHD may either be a rare association or is it possible that there is an unexplored etiopathogenetic correlation which could explain the presence of all these rare conditions in a single neonate!

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Presence of Thyroid Antibodies in a Child with Systemic Lupus Erythematosus

A 6-year-old girl presented with high grade fever daily and painful swelling of all major joints for two months. She had been treated with various anti inflammatory drugs and corticosteroids as a case of chronic arthritis without much relief. She was investigated in detail, diagnosed and confirmed to have systemic lupus erythematosus (SLE) and lupus nephritis based on the following results obtained. Her laboratory investigations were total WBC count -12,100 / cu.mm, differential count-N 62%, E 2%, L 30%, M 6%, ESR 62 mm at 1 hr, Hb