

SPOT DIAGNOSIS (IMAGE GALLERY)



LETHAL SKELETAL DYSPLASIA **Manish Tiwari, Rhishikesh Thakre ,** **Anilkumar Khamkar**

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A third gravid mother with third degree consanguinity delivered vaginally a full term male baby of 2.2 kg. The baby required bag and mask resuscitation with apgar score of 3, 5 and 7 at birth, 1 and 5 minutes respectively. Baby was shifted to nursery. The mother informed that the first baby was a home delivery, male sex, at term and died within one hour of birth with body features similar to the present baby. The second issue was a female, preterm delivery at 6 months of gestation and birth weight of 800 grams. Baby died around 18 hours of age. In this pregnancy, ultrasonography was done twice, first in second trimester which was normal and second time at 36 weeks which revealed polyhydramnios and short femur length. There was no history of bony abnormalities in the family. On admission to nursery, the baby had severe respiratory distress with short limb dwarfism and generalized subcutaneous edema. Anthropometric measurements were weight 2.2 kg { Less than 10th centile}, length 38.4 cm { Less than 5th centile}, head circumference 35.4cm { 90th centile} and upper: lower segment ratio 2.01:1. On physical examination the baby had macrocephaly, large anterior fontanelle, flat occiput, hypertelorism, depressed nasal bridge, anteverted nostrils, long philtrum, deformed low set ears, atretic ear canal, short neck, short thorax, hypertrichosis, protuberant abdomen with huge hydrocele, short limbs {mesomelic} and bilateral club foot. On systemic examination there was respiratory distress with Downes score of 6, no evidence of murmur, abdomen distended due to ascites with no obvious neurodeficit. Radiological survey showed generalised sclerosis of bones. The skull parietal bones were prominent with abnormally based occipital synchondrosis. Long bones appeared short with wide diaphyses and smooth and rounded metaphyses. Shortening was predominantly mesomelic i.e. middle segment. Thorax was short and ribs were normal. Vertebrae were flat and there was absent ossification of pubic rami. Present baby succumbed to respiratory failure at 36 hours of age. Post-mortem bone biopsy done with consent of parents from three different sites was normal. Chromosomal analysis revealed normal karyotype. DNA analysis could not be done.

What is the diagnosis?

Lethal Skeletal Dysplasia -Type Al Gazali. {1,2} The clinical phenotype of the facial features with a flat face and hypertelorism, as well as lowest ears are similar to those seen in achondrogenesis, but the extremities in achondrogenesis cases are much shorter. The radiological picture is completely different from the achondrogenesis group of phenotypes, but shares some common features with mild hypochondrogenesis. However, osteosclerosis, absence of metaphyseal irregularities, adequate ossification of vertebral bodies are not signs found in hypochondrogenesis. Also the bone biopsy, which was normal in our case, is not a feature of achondrogenesis and hypochondrogenesis. In both these condition there are severe changes in growth plate. {1,2} Another differential diagnosis could be dysplastic cortical hyperostosis described by Kozlowski and Tsuruta {3} but microcephaly and the normal size of the hands and feet as well as normal ossification of the pubic bones differentiates this entity.

References-

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