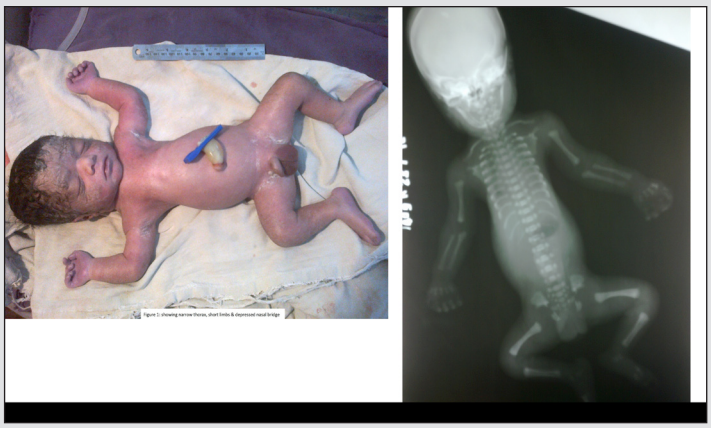
SPOT DIAGNOSIS (IMAGE GALLERY)



SKELETAL DYSPLASIA

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A 28 year old healthy non-consanguineously married woman with 34 weeks gestation presented to our hospital. Her first child is healthy and one year old. Antenatal ultrasound scans of the present pregnancy done at 12 and 20 weeks were reported as normal. A repeat ultrasound scan done at 32 weeks revealed single live foetus with tetramicromelia, with femur length corresponding to 25 weeks, macrocephaly (34 weeks) and a narrow thorax. At birth, the baby weighed 2000 grams, short in length, poor Apgar score and poor respiratory effort. Physical examination revealed macrocephaly, frontal bossing, depressed nasal bridge, funnel shaped chest with narrow thorax, protuberant abdomen and short limbs (Fig 1). Radiographic evaluation revealed long narrow thoracic cage, shortening of all the long bones, short ribs with wide cupped costo-chondral junction with short and small iliac bones and short tubular bones of the hands (Fig 1). The baby died within 20 minutes. A clinical autopsy revealed no internal anomalies.

What is the diagnosis?

Thanatophoric dysplasia. It is the most common type of lethal neonatal skeletal dysplasia with an incidence of 1:20000 – 1:50000 births, characterized by marked underdevelopment of the skeleton and short-limb dwarfism. (1) The etiology is due to the mutation in the fibroblast growth factor receptor 3 gene (FGFR 3), located on the short arm of chromosome 4. (1) Two forms of thanatophoric dysplasia have been identified. (1) Type I is characterized by marked under-development of the skeleton, short and curved long bones, metaphyseal flaring, underdeveloped pelvic bones and roof of the acetabulum is flat. The vertebral bodies are flat and underdeveloped. In type II, the long bones are not as short as in type I, they are not bent or bowed. There is metaphyseal flaring and cupping. The vertebral bodies are not as flat as in type I. Skull is clover-leaf shaped due to premature closure of coronal and lambdoid sutures. Other features are relatively large head with frontal bossing, depressed nasal bridge, prominent eyes and hypertelorism. Brain malformations are also associated such as megalencephaly in 100 percent due to temporal lobe enlargement. Hippocampus is dysplastic. (1) Encephalocele and holoprosencephaly (2) and chorioangioma of placenta (3) producing cardiac failure in fetus with thanatophoric dysplasia have been described. Accurate diagnosis of thanatophoric dysplasia is important for parental counselling and management. A definite diagnosis can be established by molecular genetic analysis to find out the abnormal mutations in the FGR3 gene. (4)

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E-published: September 2012 . **Art**#61, **DOI No. :** 10.7199/ped.oncall.2012.61

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