LETTER TO EDITOR (VIEWER'S CHOICE)

THANATOPHORIC DWARFISM: A LETHAL SKELETAL DYSPLASIA

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A $G_2P_1A_0L_1$ woman aged 20 years presented with complaints of abdominal over distention and pain following eight months amenorrhea. She was an un-booked and uninvestigated case first time reporting to hospital in whole duration of pregnancy. She had one, two and half year old alive and healthy female child who was delivered at home without any complication. In present pregnancy, there was no definitive history of any drug intake, radiation exposure, high-grade fever, rashes or fits in the peri-conceptional period. On examination, abdomen was grossly distended and tense. Abdominal veins were prominent. Abdominal girth was 122cms. Fetal parts, lie, presentation could not be appreciated due to distention. Fetal heart sounds were not audible by stethoscope. On vaginal examination, os was one finger dilated, tense bag of membranes felt and presenting part was not appreciable. She was suspected as a case of polyhydramnios or multiple pregnancy. Ultrasound showed a single intrauterine fetus in cephalic presentation of 35 weeks gestational age with biparietal diameter (8.8cm), extremely short femurs (22mm), narrow thorax (23.2 cm circumference) in comparison to abdomen (34.2cm) and polyhydramnios (AFI-28). She spontaneously went into active labor. Baby presented as vertical lie, face presentation, with mentoanterior position. She delivered a moribund short-limbed female of 2.0 kg weight, having severe respiratory distress, who could not be revived despite resuscitation.

At birth, baby's face was congested & swollen probably because of the face presentation. There was macrocephalic head with frontal bossing, flattened nasal bridge, proptotic eyes, protruding abdomen, short proximal upper and lower limb bones, which were abducted and externally rotated and square "trident" hand. (Figure 1)

X-ray showed short ribs with narrow (Bell shaped) chest and protruding abdomen giving the torso a "Champagne Cork" appearance, vertebrae were flat ("platyspondyly") giving "H" shape, with increased inter-vertebral disk spaces, markedly shortened and

Figure 1: Rhizomelic dwarfism

curved proximal long bones (Rhizomelia) especially femur as "French Telephone Receiver". Fibula was shorter than tibia. There were squared very small pelvic bones.

Thanatophoric dysplasia / dwarfism meaning death bearing (Greek) is a lethal congenital form of short-limbed bone dysplasia (1). It clinically presents with polyhydramnios or hydrocephalus. It is diagnosed prenatally, second trimester onwards with the help of ultrasonography and X- Rays. The pregnancy usually results in still-birth or early neonatal death because of pulmonary hypoplasia leading to respiratory failure. (2) Thanatophoric dwarfism in Greek meaning "death bearing" or "death bringing" and was first described in 1967 by Maroteaux et al (3). The estimated frequency is approximately 0.5:10,000 births with equal male - female ratio (4). Two subtypes I & II have been described with relative incidence of 86% and 14% respectively. Our case is of type I variety characterized by extreme rhizomelia, bowed long bones, narrow thorax, a relatively large head, normal trunk length, flattened ribs with increased disk spaces. Prominent forehead, frontal bossing, hyper-telorism, saddle

nasal bridge are the common facial findings. In type II variety, limbs are short but straight, and "Clover leaf skull" is characteristic (5). It is an autosomal dominant genetic defect said to result from de novo mutation of the Fibroblast Growth Factor Receptor gene (FGFR-3) located on the short arm of chromosome '4' affecting the skeletal and craniofacial development. (6) Molecular genetic analysis from cultured amniotic fluid cells, cord blood or fetal tissue can provide the definitive diagnosis. If massive hydrocephalus is present, cephalocentesis be done. Caesarean section should be considered only for obstetrical indications. This has a low risk of recurrence (2%) because condition requires new independent dominant mutation to reoccur.

The incidental discovery of a skeletal dysplasia on routine ultrasound screening, in pregnancy not known to be at risk of a specific syndrome, necessitates a complete evaluation of all limb bones for shortening, shape, mineralization and associated anomalies of head, thorax, spine and face. Therefore, combination of ultrasound, radiological, biochemical and cytogenetic analysis is necessary.

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