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A Rare Case Of Skeletal Multiple Dysplasia: Lethal Thanatophoric Dysplasia

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Abstract: *We report a case of skeletal multiple dysplasia at term pregnancy delivered vaginally at Regional institute of Medical sciences, Imphal. The skeletal dysplasias are a heterogeneous group of more than 350 disorders frequently associated with orthopaedic complications and varying degrees of dwarfism or short stature. Because of high lethality and morbidity rate as well as high possibility to be found again in the next pregnancy, they need to be actively looked for future ultrasounds.*

Key Words*: Thanatophoric, dysplasia, ultrasound, skeletal*

I. Case Report

A 23 years old woman was admitted on 18th January 2015 at Regional Institute Of Medical sciences, Imphal as G1P0+0 at 40 weeks 2 days for safe confinement. Ultrasound done at 25 week of gestation showed multiple skeletal dysplasia with micromelia and polyhydromnios having AFI 29 cm, narrow thorax, all bones were shortened especially femur, tibia, humerus and fetal weight was 887 grams. Mother was of an average built. There was no history of consanguineous marriage. Patient and her relatives were counselled regarding prognosis, survival of baby and complications. They wanted to continue the pregnancy. She was irregular for check up and did not come for follow up. She was induced at 40 weeks 3 day gestation with prostaglandin E1 and she delivered a live female baby by vaginal delivery. APGAR score was 9/10 and baby had short limbs with narrow thorax. Neonatal resuscitation was done .Baby admitted in NICU but passed away after 24 hours.

II. Introduction

The skeletal dysplasias are a heterogeneous group of more than 350 disorders frequently associated with orthopedic complications and varying degrees of dwarfism or short stature(1,2) . By defination, osteochondrodysplasias, or skeletal dysplasias, refer to disorders with generalized abnormalities of the skeleton, whereas the dysostoses are those disorders that have a single or group of abnormal bones.Although each skeletal dysplasia is relatively rare, collectively the birth incidence of these disorders is almost 1/5000(3). These disorders range in severity from precocious arthropathy in relatively average stature individuals to severe dwarfism with perinatal mortality. These disorders can be associated with a variety of orthopedic, neurologic,

auditory, visual, pulmonary, cardiac, renal, and psychological complications. The four most common skeletal dysplasias are thanatophoric dysplasia, achondroplasia, osteogenesis imperfecta and achondrogenesis.

Thanatophoric dysplasia is characterized by macrocephaly, narrow bell- shaped thorax, shortened ribs, and severe shortening of long bones , short limbs, long narrow trunk, large head with bulging forehead, prominent eyes, flat nose bridge, wide fontanels, and occasionally cloverleaf skull deformity. Thanatophoric dysplasia (TD) occurs in approximately 1/20,000-1/40,000 births(4)

It is diagnosed by ultrasound evaluation,in utero radiography and fetal MRI. The skeletal dysplasias can be inherited as autosomal dominant, autosomal recessive, or X-linked disorders, and some disorders that result from imprinting errors, somatic mosaicism, and teratogen exposure.(5)

III. Discussion

Ultrasound is the most sensitive way of prenatal diagnosis of the skeletal dysplasia anomalies. Maternal habitus, fetal position, alteration of amniotic fluid volume and gestational age can be factors that may aggravate check-ups. A well organized scan of each and every fetus that checks all bones, starting with the skull and finishing with hands and feet should be carried out. In case of a lethal anomaly, an option of pregnancy termination should be considered and if the parents decide against it, the pregnancy should be closely monitored, with parents fully counseled about the prognosis for the child. If an anomaly is found an extensive detailed scan by an expert should be performed, as well as karyotyping and a consultation with pediatrician/surgeon. In ultrasound evaluation, Fetuses with long bone measurements at or less than the 5th centile or >3 SD below the mean should be evaluated in a center with expertise in the recognition of skeletal dysplasias. Lethality should be determined by chest circumference to abdominal circumference ratio and/or femur length to abdominal circumference measurement ratio. A chest-to-abdominal circumference ratio of less than 0.6, or femur length to abdominal circumference ratio of 0.16 strongly_suggests a perinatal lethal disorder.(6)

In our case she came for check up first time at 25 week of gestation. She wanted to continue pregnancy, we could not terminate the pregnancy. At 40 weeks 3 day she delivered female baby having narrow chest, severe shortening of long bones. Short limbs, extra fold of skin. Most probably it looks like thanatophoric dysplasia. If anomaly scan was done earlier ,probably could have been terminated early. She was advised for genetic counselling and karyotype examination as there is a risk to be found again in next pregnancy. This case could not be resolved definitely, for the anomalies were conclusive neither of a specific genetic syndrome, nor of an isolated skeletal dysplasia. Extensive further examination of parents as well as their families is needed in order to be able to give some information about the risks in the next pregnancy.

IV. Conclusion

Prenatal diagnosis of skeletal dysplasias can present a considerable diagnostic challenge. A baby with thanatophoric dysplasia is a painful occurrence to mother and family and recurrence is a painful condition. However, a meticulous sonographic examination yields high overall detection. Although skeletal anomalies are extremely difficult to diagnose alternatively , a detailed scan of a complete fetal anatomy between 20 and 32 gestational weeks with special attention given to the entire skeleton, enables their diagnosis and further adequate plan for pregnancy management and future genetic check up.

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