PB-044

Thanatophoric displasia

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Objectives: Thanatophoric dysplasia (TD) is a lethal congenital anomaly with skeletal dysplasia. It is charecterized by short limb dwarfism, enlarged head with frontal bossing, short neck, protuberant abdomen because of thorasic dysplasia. Fibroblast growth factor receptor 3 (FGFR3) is the only gene known to cause thanatophoric dysplasia. We report a prenatal diagnosis of TD to highlight the importance of an accurate diagnosis in counselling and plan of management.

Case: A 27-year-old G3P2 woman in a non-consanguineous marriage was referred at 18 weeks' gestation for fetal skeletal evaluation. Ultrasound examination showed markedly shortened long bones and bowed femur, bilateral clubfeet, short ribs, narrow thorax and platyspondyly and increased nuchal fold (7.4 mm). The head was enlarged with bulging of forehead, eyes were prominent and were widely spaced, Thanatophoric dysplasia was suspected. The couple were counselled regarding the lethality of the condition. Amniocentesis was accepted by the couple, performed during the 18th gestational week showed normal karyotypes (46, XY) We counseled the couple extensively, and explained that this genetic condition causes type-1 TD. Thereafter, they decided to proceed with fetal termination. At the 19th gestational week, pregnancy was terminated. Laboratory investigations: After termination, fetal radiologic surveying and autpsy was performed. The findings on fetal autopsy were short and stubby femur, tibia, fibula, humerus, ulna and radius, curved femora, bilateral talipes, micrognathia. The ribs were short and horizontally placed. To further support the diagnosis, histopathological examination of section from femur revealed disorganized physeal growth zone with disordered proliferation and hypertrophy of chondrocytes and peripheral band of horizontally oriented fibrosis. The final diagnosis with the anatomical abnormalities, radiological and histopathological findings was TD type 1. Histopathology: The epiphyseal cartilage in the long bones did not differ clearly from those of the age-matched controls. The growth plate showed a variable disorganization, the rows of the proliferative and the columns of the hypertrophic chondrocytes were well formed in some areas, but nearly absent in other areas. The primary and secondary trabeculae of the metaphysis were plump and haphazardly arranged. Most striking, however, were tufts of fibrous tissue scattered along the growth plate, especially at the periphery of the growth plate, as an extension of the so-called ossification groove of Ranvier. This mesenchymal tissue penetrated the growth plate and transformed into irregularly formed plump bony trabeculae. At the periphery of the growht plate, the periosteal ossification far exceeded the plane of the endochondral ossification. This tissue penetrates the growth plate and shows atypical ossification.

Conclusion: Skeletal dysplasias constitute a heterogeneous group of bone growth disorders resulting in abnormal shape and size of the skeleton. TD is the most common lethal osteochondrodysplasias with a prevalence of 1:20,000 to 1:40,000 births. It is characterized by marked underdeveloped skeleton and short-limb dwarfism. The term 'thanatophoric' derived from the Greek word "thanatophorus", means "death bringing" and was first described by Maroteux. TD is caused by activation of FGFR3 gene leading to negative regulation of bone growth. The activation of FGFR3 in a majority is due to denovo mutations. There are two clinical types of TD which closely resemble in their clinical features; however have distinct radiological features and genetic mutations. Type 1, the more commonly encountered form is characterized by a normal-shaped skull and curved (telephone receiver shaped) long bones, especially seen in femur bone, whereas type II is associated with a cloverleaf-shaped skull and straight femurs. The close differential diagnoses of TD, includes other skeletal dysplasias characterized by micromelia and severe thoracic hypoplasia,namely osteogenesis imperfecta type II, which is characterized by fracture of long bones and achondrogenesis characterized by extreme hypomineralization and micrognathia. Hypophosphatasia is characterized, in addition to micromelia and thoracic hypoplasia, by ubiquitous hypomineralization. Other rarer differentials include achondroplasia, perinatal hypophosphatemia, campomelic dysplasia and hypochondrogenesis. Accurate diagnosis of fetal skeletal abnormalities is important for patient counselling and to plan the management. In spite of the very poor prognosis, some patients might opt to continue with the pregnancy. In such patients, ultrasound follow-up examination is important to detect early changes that might complicate delivery. Thanatophoric dysplasia is always lethal, due to the severe pulmonary and thoracic hypoplasia. Both types are due to de novo mutations. Therefore, the recurrence risk is trivial.

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İntrauterin ikiz eşi ölümü olan gebeliklerin koryonisite ve ikiz eşi ölüm zamanına göre gebelik sonuçlarının değerlendirilmesi

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Amaç: Çoğul gebelikler, tekil gebeliklere kıyasla artmış perinatal mortalite riskine sahiptir. Monozigotik ikizlerde, dizigotik ikiz gebelikle kıyasla her iki fetusun kaybı rölatif riski 20, bir