Fibrochondrogenesis

WHO International Classification of Diseases
Congenital malformations, deformations and chromosomal abnormalities

OMIM Number
228520

Mode of Inheritance
Autosomal recessive

Description
Fibrochondrogenesis is a rare disorder with one case recorded among 1,158,067 live-births registered by the Spanish Collaborative Study of Congenital Malformations (ECEMC). Until March 2003, only a limited number of cases with fibrochondrogenesis were recorded worldwide. Fibrochondrogenesis is a neonatally lethal rhizomelic chondrodysplasia distinguished from other forms of lethal dwarfism by broad long-bone metaphyses, pear-shaped vertebral bodies, and characteristic microscopic changes of cartilage: unique interwoven fibrous septa and fibroblastic dysplasia of chondrocytes. Fibrochondrogenesis is distinguished radiologically by the widening of the metaphyses of the long bones, and, on lateral X-ray of the spine, by a median fissure of the body of the vertebra without any loss of vertebral height. Omphalocele is the only internal anomaly known to be associated with this disease.

Epidemiology in the Arab World

Lebanon
Megalbane et al. (1998) reported fibrochondrogenesis in an affected boy whose parents were second cousins. Prenatal ultrasonography performed at 22 weeks of gestation revealed intrauterine growth retardation, an apparently large head, hypoplasia of the thorax, prominent abdomen, rhizomelic limbs, and wide metaphysis. The later characteristic has never been reported before in any osteochondrodysplasia. In addition, the infant presented a small flat nose, protuberant eyes, and the dumbbell-shaped bones characteristic of fibrochondrogenesis. The child died two hours after delivery, due to respiratory distress. An earlier child born to the same couple, was reportedly, similarly affected, and died 30 minutes after delivery.

Oman
Al-Gazali et al. (1997) reported an Omani child with fibrochondrogenesis. This was the first report of fibrochondrogenesis in an Arab patient. The patient was a female child of first cousin parents; making autosomal recessive inheritance likely. Short limbs were diagnosed prenatally in the third trimester. Radiological analysis revealed a large skull with a wide anterior fontanelle and sutures. The thorax was narrow with very short ribs that were cupped anteriorly and posteriorly. Al-Gazali et al. (1999) reported that the couple subsequently had one miscarriage at three months and one normal female child. The fourth pregnancy of the couple was complicated by polyhydramnios. Short limb dwarfism was diagnosed at 24 weeks of gestation. The pregnancy ended in the birth at term of a male child. He had a wide anterior fontanelle, a flat face, large prominent eyes with hypertelorism, a very small/short upturned nose with a depressed nasal bridge, low set ears, a long philtrum, a small mouth with a small mandible. Skeletal X-rays showed the typical changes of fibrochondrogenesis. Al-Gazali et al. (1999) concluded that the presence of consanguinity and the fact that both sexes are affected in this family supports autosomal recessive inheritance of fibrochondrogenesis. However, they did not rule out the possibility of gonadal mosaicism for a dominant mutation in one of the parents.
United Arab Emirates
In a 5-year prospective study for newborns at Al Ain Medical District, Al-Gazali et al. (2003) defined the pattern and birth prevalence of the different types of osteochondrodysplasias in the United Arab Emirates. Among the 38,048 births during the study period, 36 (9.46/10,000 births) had some type of skeletal dysplasia of which 18 were attributed to autosomal recessive genes (4.7/10,000 births). The most common recessive type of skeletal dysplasia in the analyzed group was fibrochondrogenesis. Four cases were identified, including three that came from the same family. Al-Gazali et al. (2003) calculated the birth rate of osteochondrodysplasias in the United Arab Emirates to be 1.05/10,000 births. This is by far higher than the rates described in other world populations.

References

Contributors
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