Schneckenbecken Dysplasia

**Alternative Names**  
Chondrodysplasia, Lethal Neonatal, with Snail-Like Pelvis

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

**OMIM Number**  
269250

**Mode of Inheritance**  
Autosomal recessive

**Description**  
Several types of lethal short-limbed skeletal dysplasia with platyspondylia have been recognized with a different mode of inheritance. Schneckenbecken dysplasia, a rare skeletal dysplasia, is one of them, with an autosomal recessive mode of inheritance. The characteristic radiographic findings consist of flattened, hypoplastic vertebral bodies; short ribs; hypoplastic iliac bones with "a snail-like" configuration; short, broad long-bones with dumbbell-like appearance; short and wide fibula; and precocious ossification of the tarsus. Chondro-osseous histology is characteristic with hypervascularity, increased cellular density, and normal size chondrocytes with a centrally located round nucleus and absence of lacunar space.

**Epidemiology in the Arab World**

**Palestine**  
Chemke et al. (1971) and Graff et al. (1972) described thanatophoric dwarfism in two male offspring of first-cousin Moroccan Jewish parents. In the second-born affected sib the diagnosis was made antenatally by X-ray. However, after review of the radiographs of one, Rimoin (1975) concluded that this was not thanatophoric dwarfism. Thus, Rimoin (1975) concluded that 'there are no well-documented examples of familial thanatophoric dwarfism; a genetically lethal autosomal dominant mutation or an environmental agent could explain' its occurrence. Knowles et al. (1986) and Borochowitz et al. (1986) suggested that the disorder reported by Chemke et al. (1971) and Graff et al. (1972) was the same as the 'new' autosomal recessive dysplasia they described under the designation of Schneckenbecken dysplasia.

**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 one case, born to a consanguineous parents, had Schneckenbecken dysplasia. Al-Gazali et al. (2003) calculated the birth rate of this type of osteochondrodysplasia in the United Arab Emirates to be 0.26/10,000 births.

**References**


Contributors
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