



Rhizomelic Chondrodysplasia Punctata, Type 1

Alternative Names

RCDP1
Chondrodysplasia Punctata, Rhizomelic Form
CDPR
Chondrodystrophia Calcificans Punctata
PTS2 Deficiency
Alkylglycerone Phosphate Synthase Deficiency

WHO International Classification of Diseases

Congenital malformations, deformations and chromosomal abnormalities

OMIM Number

215100

Mode of Inheritance

Autosomal recessive

Gene Map Locus

6q22-q24

Description

Rhizomelic chondrodysplasia punctata is a rare, multisystem, developmental disorder, characterized by the presence of stippled foci of calcification in hyaline cartilage, coronal vertebral clefting, dwarfing, joint contractures, congenital cataract, ichthyosis, and severe mental retardation. The cataracts are present in about 72% of cases, and skin changes in about 27%. The coronal cleft of the vertebral bodies is demonstrable radiologically and appears to represent embryonic arrest with cartilage occupying the cleft between the anterior and posterior parts of the vertebral bodies. Biochemically, RCDP patients have subnormal levels of red cell plasmalogens and progressive accumulation of phytanic acid starting from normal at birth and increasing to levels more than 10 times normal by age 1 year. Specifically, involvement of the vertebral bodies has been described.

Molecular Genetics

Most patients with rhizomelic chondrodysplasia punctata type 1 have mutations in the PEX7 gene, which codes for the peroxin 7, the cytosolic PTS2-receptor protein required for peroxisomal import of proteins containing a peroxisomal targeting signal type 2 (PTS2). These enzymes are deficient in cells of patients with RCDP, because of their mislocalization to the cytoplasm.

Epidemiology in the Arab World

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 three had autosomal recessive rhizomelic and non-rhizomelic chondrodysplasia punctata. In two of these cases, the parents were consanguineous. Al-Gazali et al. (2003) calculated the birth rate of this type of osteochondrodysplasia in the United Arab Emirates to be 0.78/10,000 births. This is higher than that reported for other world populations.

References

Al-Gazali LI, Bakir M, Hamid Z, Varady E, Varghes M, Haas D, Bener A, Padmanabhan R, Abdulrazzaq YM, Dawadu A, Abdulrazzaq YM, Dawodu AK. Birth prevalence and pattern of osteochondrodysplasias in an inbred high risk population. *Birth Defects Res Part A Clin Mol Teratol.* 2003; 67(2):125-32.

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