



Dyssegmental Dwarfism

Alternative Names

Dyssegmental Dysplasia
Anisospondylic Camptomicromelic Dwarfism
Lethal Anisospondylic Camptomicromelic Dwarfism
Rolland-Desbuquois Syndrome

WHO International Classification of Diseases

Congenital malformations, deformations and chromosomal abnormalities

OMIM Number

224400

Mode of Inheritance

Autosomal recessive

Description

Dyssegmental dwarfism is an autosomal recessive skeletal dysplasia characterized by anisospondyly and micromelia. Two types are distinguished: a severe, lethal Silverman-Handmaker type and a milder Rolland-Desbuquois form. Although the etiology of the Rolland-Desbuquois form has not been explained, the cause of the Silverman-Handmaker type has been shown to be a lack of extracellular matrix perlecan. Perlecan is a large heparan sulfate proteoglycan present in all basement membranes and in some other tissues such as cartilage, and is implicated in cell growth and differentiation.

Given the recurrence risk of 25% and the lethality of the condition, carrier parents need to receive appropriate genetic counseling for the management of future pregnancies. Prenatal diagnosis of skeletal dysplasia with severe micromelia is usually detectable by in utero ultrasound by 16 to 20 weeks of gestation.

Molecular Genetics

Recently, a mutation has been reported in the heparan sulfate proteoglycan 2 gene HSPG2 as

a possible cause of dyssegmental dwarfism (Silverman-Handmaker type).

Epidemiology in the Arab World

Jordan

Miething et al. (1981) reported male newborn with dyssegmental dwarfism. He died from asphyxia immediately after delivery. The infant has malformations of the first visceral arch (Pierre-Robin-Syndrome) malformations of all extremities (Camptomicromelia) and severe malformations of the spine (Anisospondyly).

Svejcar (1983) observed an offspring of presumably unrelated Jordanian-Palestinians with dyssegmental dwarfism. The proband had hypertrichosis with short and bent limbs with reduced mobility. Svejcar (1983) found abnormal gel electrophoretic patterns of collagen peptides, pointing to a deficiency in alpha-1 chains. This deficiency may be responsible for increased cross-linking and the observed alterations in extractability of collagen.

Palestine

[See also: Jordan > Svejcar (1983)].

References

- Miething R, Stover B, Tuengerthal S, Winterling D, Svejcar J. [Dyssegmental dwarfism--report on two cases] *Radiologe*. 1981; 21(4):190-4.
- Svejcar J. Biochemical abnormalities in connective tissue of osteodysplasty of Melnick-Needles and dyssegmental dwarfism. *Clin Genet*. 1983; 23(5):369-75.

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