

Opis choroby *

Definicja

A rare spondyloepiphyseal dysplasia characterized by progressive joint contractures with premature degenerative joint disease, particularly in the knee, hip, and finger joints. Patients are of normal height and present with gait problems, joint pain, and enlarged joints with joint restriction and contractures. Radiological features include generalized platyspondyly, hypoplastic ilia, epiphyseal flattening with metaphyseal splaying of the tubular bones, and broad, elongated femoral necks with marked coxa valga. Histopathologic examination of cartilage shows PAS-positive cytoplasmic inclusion bodies in chondrocytes.

Dane

Klasyfikacja

Choroba

Synonimy

SED, Stanescu type

SED, typ Stanescu

Kod ORPHA

459051

Kod OMIM

616583

Kod ICD10

Q77.7

Kod ICD11

-

*Źródło

orphanet