

Opis choroby *

Definicja

Microcephalic osteodysplastic dysplasia, Saul-Wilson type is a skeletal dysplasia characterized by a distinct facial phenotype, short stature, brachydactyly, clubfoot deformities, cataracts, and microcephaly. It has been described in four patients. Facial features include frontal bossing with a depression over the metopic suture, a narrow nasal root with a beaked nose, and midfacial hypoplasia with prominent eyes. Characteristic radiographic findings are observed (irregularities of the vertebral bodies, hypoplasia of the odontoid process, short phalanges, coning several epiphyses *etc.*).

Dane

Klasyfikacja

Choroba

Kod ORPHA

85172

Kod OMIM

618150

Kod ICD10

Q78.8

Kod ICD11

LD24.D

*Źródło

orphanet