

## 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

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### \*Źródło

orphanet