

Opis choroby *

Definicja

A severe form of otopalatodigital syndrome spectrum disorder, and is characterized by dysmorphic facies, severe skeletal dysplasia affecting the axial and appendicular skeleton, extraskeletal anomalies (including malformations of the brain, heart, genitourinary system, and intestine) and poor survival.

Dane

Klasyfikacja

Zespół wad wrodzonych OPD II syndrome
OPD syndrome 2
OPD II syndrome
OPD syndrome 2

Kod ORPHA

90652

Kod OMIM

304120

Kod ICD10

Q87.0

Kod ICD11

LD25.1

*Źródło

orphanet