Opis choroby *

Definicja

A rare genetic disease characterized by congenital oculocutaneous hypopigmentation, visual impairment, generalized osteoporosis with skeletal anomalies such as short stature, short neck and trunk, kyphosis, scoliosis, and platyspondyly, and dysmorphic facial features (including long philtrum, small mouth, micrognathia, and prominent ears). Moderate joint hyperelasticity and muscular hypotrophy have also been reported.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Hernández-Fragoso syndrome
OOCHS
Zespół Hernandeza i Fragoso
OOCHS

Kod ORPHA
2786

Kod OMIM
601220

Kod ICD10
Q87.5

Kod ICD11
LD24.KY

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*Źródło

orphanet