

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	Synonimy
Zespół wad wrodzonych	Hernández-Fragoso syndrome OOCHS Zespół Hernandeza i Fragoso OOCHS

Kod ORPHA	Kod OMIM	Kod ICD10
2786	601220	Q87.5

Kod ICD11
LD24.KY

*Źródło

orphanet