

Opis choroby *

Definicja

A rare congenital disorder of copper metabolism that is principally characterized by bony exostoses (including the pathognomonic occipital horns), and connective tissue manifestations with cutis laxa and bladder diverticula. Central nervous system involvement is variable.

Dane

Klasyfikacja

Choroba

Kod ORPHA

198

Kod OMIM

304150

Kod ICD10

E83.0

Kod ICD11

LD28.2

[*Źródło](#)

orphanet