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

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*Źródło

orphanet