Opis choroby *

Definicja

A rare, highly variable, multisystemic disorder mainly characterized by short stature, distinctive facial features, congenital heart defects, cardiomyopathy and an increased risk to develop tumors in childhood.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
NS

Kod ORPHA
648

Kod OMIM
616564, PS163950

Kod ICD10
Q87.1

Kod ICD11
LD2F.15

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*Źródło

orphanet