Opis choroby *

Definicja

A rare, genetic, multiple congenital anomalies/dysmorphic syndrome characterized by the association of short stature and progressive discrete subaortic stenosis. Additional variable manifestations include upturned nose, voice and vocal cord abnormalities, obstructive lung disease, inguinal hernia, kyphoscoliosis and, occasionally, epicanthus, strabismus, microphthalmos and widely spaced teeth. There have been no further descriptions in the literature since 1984.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Onat syndrome
Zespół Onata

Kod ORPHA
3191

Kod OMIM
271960

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet