Opis choroby *

Definicja

A multiple congenital anomalies syndrome characterized by wormian bones, dextrocardia and short stature due to a growth hormone deficiency. Additional manifestations that have been reported include brachycamptodactyly, kidney hypoplasia, bilateral cryptorchidism, midshaft hypospadias, imperforate anus/anorectal agenesis, body asymmetry, mild developmental delay, hemimegalencephaly and facial dysmorphism (hypotelorism, downslanting palpebral fissures, low-set and posteriorly angulated ears, depressed nasal bridge, and microstomia).

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Stratton-Parker syndrome
Zespół Strattona i Parkera

Kod ORPHA
2863

Kod OMIM
185120

Kod ICD10
Q87.1

Kod ICD11
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*Źródło

orphanet