

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

Synonimy

Zespół wad wrodzonych Stratton-Parker syndrome

Zespół Strattona i Parkera

Kod ORPHA

2863

Kod OMIM

185120

Kod ICD10

Q87.1

Kod ICD11

-

*Źródło

orphanet