

Opis choroby *

Definicja

Silver-Russell syndrome due to maternal uniparental disomy of chromosome 7 is a genetic malformation syndrome with short stature characterized by severe prenatal and postnatal growth retardation, feeding difficulties, body asymmetry, dysmorphic craniofacial features (triangular-shaped face, relative macrocephaly, frontal bossing, micrognathia, down-turned corners of the mouth) and other anomalies (fifth finger clinodactyly, café au lait macules, male genital anomalies, mild developmental delay and/or speech delay with movement disorders).

Dane

Klasyfikacja	Synonimy
Podtyp etiologiczny	UPD(7)mat UPD(7)mat

Kod ORPHA	Kod OMIM	Kod ICD10
96182	-	Q87.1

Kod ICD11
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*Źródło

orphanet