Opis choroby *

Definicja

A rare genetic disease characterized by intrauterine growth restriction, metaphyseal dysplasia, congenital adrenal hypoplasia, and genital anomalies (such as cryptorchidism, posterior hypospadias, and micropenis). Patients may present shortly after birth with severe adrenal insufficiency. Additional manifestations include postnatal growth failure and delayed bone age, mild developmental delay, macrocephaly, mild facial dysmorphism (with frontal bossing, wide nasal bridge, and small, low-set ears), epiphyseal dysplasia, and hypercalcemia/hypercalciuria, among others.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Intrauterine growth retardation-metaphyseal dysplasia-adrenal hypoplasia congenita-genital anomalies syndrome
Wewnątrzmaciczne opóźnienie wzrostu - dysplazja przynasadowa - wrodzona hipoplazja nadnerczy- wady narządów płciowych

Kod ORPHA
85173

Kod OMIM
614732

Kod ICD10
Q87.1

Kod ICD11
5A74.Y

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*Źródło

orphanet