Opis choroby *

Definicja

A rare genetic disease characterized by lethal non-spherocytic, non-immune hemolytic anemia, in association with abnormalities of the external genitalia (such as micropenis and hypospadias). Reported dysmorphic features include flat occiput, dimpled earlobes, deep plantar creases, and increased space between the first and second toes. There have been no further descriptions in the literature since 1995.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Water-West syndrome
Zespół Watera i Westa

Kod ORPHA
1046

Kod OMIM
600461

Kod ICD10
D58.8

Kod ICD11
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*Źródło

orphanet