Opis choroby *

Definicja

A rare autosomal anomaly defined by the presence of three copies of chromosome 8 in some cells of the body, and clinically characterized by facial dysmorphism, typically deep palmar and plantar creases, mild intellectual deficit and joint, urinary, cardiac and skeletal anomalies.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Mosaic trisomy chromosome 8
Zespół Warkany
Trisomy 8 mosaicism
Warkany syndrome

Kod ORPHA
96061

Kod OMIM
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Kod ICD10
Q92.1

Kod ICD11
LD40.Y

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*Źródło

orphanet