Opis choroby *

Definicja

A rare multiple congenital anomalies syndrome characterized by congenital microgastria and a uni- or bilateral limb reduction defect, that can include absent or hypoplastic thumbs, radius, ulna and/or amelia. Association with other variable abnormalities, including intestinal malrotation, asplenia, dysplastic kidneys, hypoplastic lungs, dysplastic corpus collosum, and abnormal genitalia, has been reported.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
2538

Kod OMIM
156810

Kod ICD10
Q87.8

Kod ICD11
LD2F.1Y

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*Źródło

orphanet