Opis choroby *

Definicja

A rare disorder characterized by multiple congenital anomalies. The name is a mneumonic for the common features observed in SHORT syndrome that include; short stature, hyperextensibility of joints, ocular depression, Rieger anomaly and teething delay. Other common manifestations of SHORT syndrome are mild intrauterine growth restriction, partial lipodystrophy, delayed bone age, hernias and a recognizable facial gestalt.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Lipodystrophy-Rieger anomaly-diabetes syndrome
Anomalia Riegera - częściowa lipodystrofia
Lipodystrofia - anomalia Riegera - cukrzyca
Zespół Aarskoga,Osego i Pande
Rieger anomaly-partial lipodystrophy syndrome

Kod ORPHA
3163

Kod OMIM
269880

Kod ICD10
Q87.1

Kod ICD11
LD27.6Z

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*Źródło

orphanet