

Opis choroby *

Definicja

A rare disorder characterized by multiple congenital anomalies. The name is a mnemonic 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

*Źródło

orphanet