Opis choroby *

Definicja

A rare genetic disease characterized by lipoatrophic diabetes, mild craniofacial dysmorphism (such as pronounced antitragal incisura and mandibular prognathism), ectodermal dysplasia (generalized hypotrichosis and dental and nail abnormalities), hypoplasia or aplasia of the breasts, and urogenital/renal anomalies. Additional reported manifestations include skeletal abnormalities and hepatosplenomegaly.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Acrorenal defect-ectodermal dysplasia-diabetes syndrome
Defekt kończynowo-nerkowy - dysplazja naskórka - cukrzyca

Kod ORPHA
1133

Kod OMIM
207780

Kod ICD10
Q87.8

Kod ICD11
LD27.0Y

---

*Źródło

orphanet