

## **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 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

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\*[Źródło](#)

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