

## 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

#### Synonimy

#### Kod ORPHA

1133

#### Kod OMIM

207780

#### Kod ICD10

Q87.8

#### Kod ICD11

LD27.0Y

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#### [\\*Źródło](#)

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