

## Opis choroby \*

### Definicja

A rare genetic disease characterized by intrauterine growth retardation, permanent neonatal diabetes mellitus, and congenital hypothyroidism. Additional manifestations include congenital glaucoma, hepatic disease (hepatitis, fibrosis, and cirrhosis), polycystic kidneys, exocrine pancreatic dysfunction, sensorineural hearing impairment, developmental delay, and mild facial dysmorphism (such as flat nasal bridge, epicanthal folds, long philtrum, and low-set ears), among others.

### Dane

### Klasyfikacja

Choroba

#### Kod ORPHA

79118

#### Kod OMIM

610199

#### Kod ICD10

P70.2

#### Kod ICD11

GB8Y

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### \*Źródło

orphanet