Opis choroby *

Definicja

A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by the association of pancreatic agenesis and lobar/semilobar holoprosencephaly. Insulin-dependent diabetes mellitus and pancreatic exocrine deficiency manifest early after birth. Additional reported manifestations include intrauterine growth retardation, muscle weakness, seizures, mild intellectual disability and dysmorphic craniofacial features, and agenesis of the gallbladder.

Dane

Klasyfikacja
Choroba

Kod ORPHA
556955

Kod OMIM
618500

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet