

Opis choroby *

Definicja

Johanson-Blizzard syndrome (JBS) is a multiple congenital anomaly characterized by exocrine pancreatic insufficiency, hypoplasia/aplasia of the nasal alae, hypodontia, sensorineural hearing loss, growth retardation, anal and urogenital malformations, and variable intellectual disability.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	JBS
	JBS

Kod ORPHA	Kod OMIM	Kod ICD10
2315	243800	Q87.8

Kod ICD11
LD27.0Y

*Źródło

orphanet