

## 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

|                       |                 |
|-----------------------|-----------------|
| <b>Klasyfikacja</b>   | <b>Synonimy</b> |
| Zespół wad wrodzonych | JBS             |
|                       | JBS             |

|                  |                 |                  |
|------------------|-----------------|------------------|
| <b>Kod ORPHA</b> | <b>Kod OMIM</b> | <b>Kod ICD10</b> |
| 2315             | 243800          | Q87.8            |

**Kod ICD11**  
LD27.0Y

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### \*Źródło

orphanet