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Johanson-Blizzard syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Johanson-Blizzard syndrome. ORPHA:2315*

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.