

Opis choroby *

Definicja

A rare X-linked syndromic intellectual disability characterized by congenital and permanent vocal cord paralysis causing severe congenital laryngeal stridor, associated with intellectual disability in male patients. Other presenting symptoms may include weak cry, cough, cyanosis, neonatal asphyxia, feeding difficulty, aspiration, and bronchiectasis. Microcephaly, tone abnormalities, visual and hearing impairment may also be associated features.

Dane

Klasyfikacja

Zespół wad wrodzonych Plott syndrome
Zespół plotta

Synonimy

Kod ORPHA

2375

Kod OMIM

308850

Kod ICD10

J38.0

Kod ICD11

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*Źródło

orphanet