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

Synonimy
Plott syndrome
Zespół plotta

Kod ORPHA
2375

Kod OMIM
308850

Kod ICD10
J38.0

Kod ICD11
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*Źródło

orphanet