

Opis choroby *

Definicja

A very rare syndrome of congenital hypothyroidism characterized by thyroid dysgenesis (in most cases athyreosis), cleft palate and spiky hair, with or without choanal atresia, and bifid epiglottis. Facial dysmorphism and porencephaly have been reported in isolated cases.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Athyroidal hypothyroidism-spiky hair-cleft palate syndrome
	Niedoczynność przytarczyc - rozszczep podniebienia
	Zespół Bamfortha i Lazarusa
	Bamforth syndrome
	Hypothyroidism-cleft palate syndrome

Kod ORPHA
1226

Kod OMIM
241850

Kod ICD10
E03.1

Kod ICD11
5A00.0Y

*Źródło

orphanet