

Opis choroby *

Definicja

A rare head and neck malformation characterized by congenital partial (hypoglossia) or total (aglossia) absence of the tongue. Patients present feeding and respiratory difficulties, as well as delayed speech development and slurred speech. Taste perception is not severely compromised. Associated features include a characteristic facies due to mandibular transverse arch deficiency, oligodontia, and malocclusion, among others.

Dane

Klasyfikacja

Wada morfologiczna

Kod ORPHA

141152

Kod OMIM

612776

Kod ICD10

Q38.3

Kod ICD11

-

*Źródło

orphanet