

Opis choroby *

Definicja

Cataract-aberrant oral frenula-growth delay syndrome is characterized by cataracts and short stature associated with variable anomalies, including aberrant oral frenula, a characteristic facial appearance (posteriorly angulated ears, upslanting palpebral fissures, small nose, ptosis and epicanthal folds) cavernous hemangiomas and hernias. It has been described in a mother and her two children. It is transmitted as an autosomal dominant trait.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Wellesley-Carman-French syndrome Zespół Wellesley'a, Carmana i Frencha

Kod ORPHA	Kod OMIM	Kod ICD10
1373	115645	Q87.8

Kod ICD11

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*Źródło

orphanet