Opis choroby *

Definicja

A rare genetic syndromic intellectual disability characterized by global developmental delay and speech delay, variable degrees of intellectual disability, and dysmorphic facial features (such as frontal bossing, epicanthal folds, strabismus, depressed nasal bridge, short philtrum, auricular abnormalities, micrognathia, or crowded teeth, among others). Additional reported manifestations are behavioral problems (stereotypies, aggression, anxiety, autism spectrum disorder), skeletal anomalies (scoliosis, pectus carinatum, clinodactyly of fingers and toes, among others), and seizures.

Dane

Klasyfikacja
Choroba

Synonimy
SOX5 haploinsufficiency syndrome
Zespół haploinsuficjencji SOX5

Kod ORPHA
530983

Kod OMIM
-

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet