

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
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Kod ORPHA 530983	Kod OMIM -	Kod ICD10 Q87.8
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Kod ICD11
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