

# Zespół Lamba i Shaffer

Kod Orpha: 530983 Kod OMIM:

## 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

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#### Kod ICD10

Q87.8

#### Kod ICD11

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#### [\\*Źródło](#)

orphanet

## Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.