Opis choroby *

Definicja

A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by global developmental delay, intellectual disability, and dysmorphic facial features (such as facial asymmetry, prominent forehead, short palpebral fissures, low nasal bridge, smooth and long philtrum, thin upper lip, and low-set, posteriorly rotated, dysplastic ears), exclusively affecting females. Additional reported manifestations include short stature, choanal atresia, scoliosis, congenital ocular, dental, cardiac, and urogenital anomalies, as well as hypotonia, seizures, and structural brain abnormalities, among others.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
X-linked facial dysmorphism-short stature-choanal atresia-intellectual disability syndrome limited to females
X-linked facial dysmorphism-short stature-choanal atresia-intellectual disability syndrome limited to females

Kod ORPHA
480880

Kod OMIM
300968

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet