Opis choroby *

Definicja

A rare X-linked syndromic intellectual disability characterized by developmental delay and intellectual disability, early hypotonia, constipation, feeding problems, imperforate anus, characteristic behavior (affable, eager to please), and dysmorphic craniofacial features (such as relative macrocephaly, prominent forehead with frontal hair upsweep, hypertelorism, downslanting palpebral fissures, and open mouth). Additional manifestations are partial agenesis of the corpus callosum, sensorineural hearing loss, joint laxity, cardiac anomalies, and abnormalities of the fingers and toes, among others.

Dane

Klasyfikacja
Choroba

Synonimy
Opitz-Kaveggia syndrome
Zespół Opitza i Kaveggia

Kod ORPHA
93932

Kod OMIM
305450

Kod ICD10
Q87.8

Kod ICD11
LD2F.1Y

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*Źródło

orphanet