Opis choroby *

Definicja

A rare, X-linked, multiple congenital anomalies/dysmorphic malformation-intellectual disability syndrome characterized by developmental delay, mild to moderate intellectual disability, speech disturbance, behavioral problems (such as anxiety, hyperactivity, and aggressiveness) and mild facial dysmorphism (including facial hypotonia, thin arched eyebrows, ectropion, epicanthus, malar flatness, thick vermillion of the lips and prognathia). Additional variable manifestations include short stature, skeletal and genital anomalies, seizures, and autism spectrum disorders. Brain imaging may reveal cerebellar vermis hypoplasia, thin corpus callosum, and enlarged subarachnoid spaces.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Dup(X)(q25)
Dup(X)(q25)
Mikrotriplikacja Xq25
Xq25 microtriplication

Kod ORPHA
521258

Kod OMIM
300979

Kod ICD10
Q98.8

Kod ICD11
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*Źródło

orphanet