

Opis choroby *

Definicja

A rare partial autosomal trisomy/tetrasomy characterized by global developmental delay, intellectual disability, autistic behavior, muscular hypotonia, macrocephaly and facial dysmorphism (frontal bossing, short palpebral fissures, low set, dysplastic ears, short or shallow philtrum, high arched or narrow palate, micrognathia). Other associated clinical features include sleep disturbances, seizures, aplasia/hypoplasia of the corpus callosum, skeletal abnormalities (large hands and feet, long fingers and toes, talipes).

Dane

Klasyfikacja

Zespół wad wrodzonych Dup(5)(p13)

Synonimy

Dup(5)(p13)
Trisomia 5p13
Trisomy 5p13

Kod ORPHA

329802

Kod OMIM

613174

Kod ICD10

Q92.3

Kod ICD11

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*Źródło

orphanet