Opis choroby *

Definicja

A rare multiple congenital anomalies/dysmorphic syndrome with intellectual disability characterized by mild global developmental delay, intellectual disability or learning difficulties, behavioral problems (like autistic, hyperactive, or aggressive behavior), variable dysmorphic craniofacial features, and abnormalities of the fingers (brachydactyly, tapering fingers, prominent interphalangeal joints). Additional manifestations are highly variable and include recurrent infections and skeletal anomalies, among others.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
476126

Kod OMIM
617061

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet