Opis choroby *

Definicja

A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by overgrowth and macrocephaly with megalencephaly apparent at birth, global developmental delay, intellectual disability, and dysmorphic facial features (including frontal bossing, long face, sparse eyebrows, hypertelorism, downslanting palpebral fissures, and prognathism). Patients may exhibit tall stature with dolichostenomelia, arachnodactyly, kyphoscoliosis, and joint laxity, as well as neurologic manifestations, such as hypotonia, gait ataxia, or seizures. Brain imaging may show increased white matter volume, thick corpus callosum, or small cerebellum.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
457359

Kod OMIM
617011

Kod ICD10
Q87.3

Kod ICD11
LD2C

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*Źródło

orphanet