Opis choroby *

Definicja

A rare multiple congenital anomalies/dysmorphic syndrome characterized by profound intellectual disability, choreoathetosis, progressive spastic diplegia, progressive tapetoretinal degeneration with loss of retinal vessels, and glomerulopathy resulting in death late in the first or early in the second decade of life. Absence of the cerebellar granular layer has been reported. There have been no further descriptions in the literature since 1982.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Hunter-Jurenka-Thompson syndrome
Zespół Huntera, Jurenka i Thompson
ORC syndrome
Oculorenocerebellar syndrome

Kod ORPHA
2715

Kod OMIM
257970

Kod ICD10
Q04.8

Kod ICD11
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*Źródło

orphanet