Opis choroby *

Definicja

Richards-Rundle syndrome is an extremely rare neurodegenerative disorder characterized by progressive spinocerebellar ataxia, sensorineural hearing loss, and hypergonadotropic hypogonadism associated with additional neurological manifestations (such as peripheral muscle wasting, nystagmus, intellectual disability or dementia) and ketoaciduria.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Ketoaciduria-intellectual disability-ataxia-deafness syndrome
Ketoaciduria - niepełnosprawność intelektualna - ataksja - głuchota
Ketoaciduria-intellectual disability-ataxia-hearing loss syndrome

Kod ORPHA
1399

Kod OMIM
245100

Kod ICD10
G60.2

Kod ICD11
LD2H.Y

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*Źródło

orphanet