

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 Synonimy

Zespół wad wrodzonych 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

*Źródło

orphanet