Richards-Rundle syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Richards-Rundle syndrome. ORPHA:1399

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.