

Opis choroby *

Definicja

SRD5A3-CDG is a rare, non X-linked congenital disorder of glycosylation due to steroid 5 alpha reductase type 3 deficiency characterized by a highly variable phenotype typically presenting with severe visual impairment, variable ocular anomalies (such as optic nerve hypoplasia/atrophy, iris and optic nerve coloboma, congenital cataract, glaucoma), intellectual disability, cerebellar abnormalities, nystagmus, hypotonia, ataxia, and/or ichthyosiform skin lesions. Other reported manifestations include retinitis pigmentosa, kyphosis, congenital heart defects, hypertrichosis and abnormal coagulation.

Dane

Klasyfikacja

Choroba

Synonimy

CDG syndrome type Iq

CDG1Q

CDG-Iq

Zespół CDG typu Iq

Wrodzone zaburzenie glikozylacji typu 1q

Wrodzone zaburzenie glikozylacji typu Iq

CDG-Iq

CDG1Q

Congenital disorder of glycosylation type 1q

Congenital disorder of glycosylation type Iq

Kod ORPHA

324737

Kod OMIM

612379

Kod ICD10

E77.8

Kod ICD11

5C54.0

*Źródło

orphanet