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

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*Źródło

orphanet