

Opis choroby *

Definicja

De Barsy syndrome (DBS) is characterized by facial dysmorphism (down-slanting palpebral fissures, a broad flat nasal bridge and a small mouth) with a progeroid appearance, large and late-closing fontanel, cutis laxa (CL), joint hyperlaxity, athetoid movements and hyperreflexia, pre- and postnatal growth retardation, intellectual deficit and developmental delay, and corneal clouding and cataract.

Dane

Klasyfikacja

Choroba

Synonimy

Cutis laxa-corneal clouding-intellectual disability syndrome
Skóra wiotka - zamglenie rogówki - niepełnosprawność intelektualna
Zespół progerii typu De Barsy
Progeroid syndrome, De Barsy type

Kod ORPHA

2962

Kod OMIM

614438

Kod ICD10

Q87.8

Kod ICD11

LD28.2

*Źródło

orphanet