

## 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

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#### \*Źródło

orphanet