

## Opis choroby \*

### Definicja

Cone dystrophy with supernormal rod response (CDSRR) is an inherited retinopathy, with an onset in the first or second decade of life, characterized by poor visual acuity (due to central scotoma), photophobia, severe dyschromatopsia, and occasionally, nystagmus. Night blindness usually develops later in the course of the disease, but it can also be apparent from childhood. A hallmark of CDSRR is the decreased and delayed dark-adapted response to dim flashes in electroretinographic recordings, which contrasts with the supernormal b-wave response at the highest levels of stimulation.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

Cone dystrophy with supernormal rod ERG

Dystrofia stożka z ponadnormalnym ERG pręcików

Cone dystrophy with supernormal rod electroretinogram

Cone dystrophy with supernormal scotopic electroretinogram

#### Kod ORPHA

209932

#### Kod OMIM

610356

#### Kod ICD10

H35.5

#### Kod ICD11

9B7Y

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#### \*Źródło

orphanet