

Opis choroby *

Definicja

Snowflake vitreoretinal degeneration (SVD) is characterised by the presence of small granular-like deposits resembling snowflakes in the retina, fibrillary vitreous degeneration and cataract. The prevalence is unknown but the disorder has been described in several families. Transmission is autosomal dominant and the causative gene has been localised to a small region on chromosome 2q36.

Dane

Klasyfikacja

Choroba

Kod ORPHA

91496

Kod OMIM

193230

Kod ICD10

H35.5

Kod ICD11

9B70

*Źródło

orphanet