

Opis choroby *

Definicja

Primary ciliary dyskinesia - retinitis pigmentosa is an X-linked ciliary dysfunction of both respiratory epithelium and photoreceptors of the retina leading to ocular disorders (mild night blindness, constriction of the visual field, and scotopic and photopic ERG responses reduced to 30-60%) associated with primary ciliary dyskinesia (see this term) manifestations (chronic bronchorrhea with bronchoectasis and chronic sinusitis) and sensorineural hearing loss.

Dane

Klasyfikacja

Choroba

Kod ORPHA

247522

Kod OMIM

300455

Kod ICD10

Q34.8

Kod ICD11

LA75.Y

*Źródło

orphanet