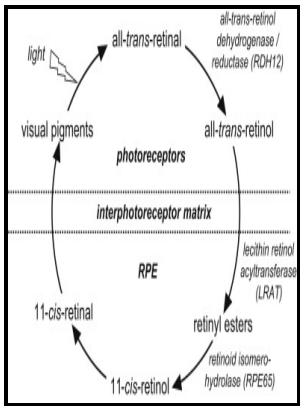
Heredo-retinopathia congenitalis - monohybrida recessiva autosomalis : a genetical-statistical study

[s.n.] - Just



Description: -
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Photography, Artistic.
Sacco, Graciela, -- 1956-
Egypt -- Social life and customs.
Retina -- Diseases -- Genetic aspects
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Hereditas -- 43Heredo-retinopathia congenitalis - monohybrida recessiva autosomalis : a genetical-statistical study
Notes: Bibliography: p. [171]-177.
This edition was published in 1957

Tags: #OMIM #Entry

Heredo

LCA11 613837 is caused by mutation in the IMPDH1 gene 146690 on chromosome 7q32. In addition, a third disease-associated mutant allele at a second locus was identified in 7 12% of the 60 patients. Genotyping microarray disease chip for Leber congenital amaurosis: detection of modifier alleles.

Heredo

The sister died at age 4 months of massive pericardial effusion.

OMIM Entry

Although multiple exonic polymorphisms were determined, no changes were identified that could be causative for the LCA phenotype. Modifier Genes Zernant et al. Heredo-retinopathia congenitalis monohybrida recessiva autosomalis: a genetical-statistical study in clinical collaboration with Olof Olson.

Heredo

However, some authors have reported a small group of children with reasonably good central vision when old enough to be tested, despite apparent blindness and reduced or absent ERG in infancy. Functional phenotyping revealed some patients with and some without detectable cone vision. LCA7 613829 is caused by mutation in the CRX gene 602225 on chromosome 19q13.

Non

Ahi1, whose human ortholog is mutated in Joubert syndrome, is required for Rab8a localization, ciliogenesis and vesicle trafficking. Electroretinogram ERG responses are usually nonrecordable. Three of the 4 children had decreased growth hormone secretion.

Just

Mutations in TMEM216 perturb ciliogenesis and cause Joubert, Meckel and related syndromes. Molecular cloning, chromosomal localization, tissue mRNA levels, bacterial expression, and enzymatic properties of human NMN adenylyltransferase.

OMIM Entry

Twenty-seven of the sporadic cases were from consanguineous families.

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