

Table C-34. Ongoing clinical trials examining genetic aspects of retinal degenerative disorders (2 studies)

Clinicaltrials.gov Title Clinicaltrials.gov Identifier Sponsor	Study Design	Purpose	Start Date Expected Completion Date Estimated Enrollment	Primary Outcomes	Secondary Outcomes	Eye-specific Inclusion Criteria	Eye-specific Exclusion Criteria
Molecular Genetics of Retinal Degenerations NCT00231010 National Eye Institute	Diagnostic case series	To investigate in a multinational study the inheritance of genetic retinal degeneration in families of different nationalities and ethnic backgrounds to identify the genes that, when altered, cause retinal degeneration. The findings of this study should help improve diagnosis and methods of treatment for these disorders	September 2005 June 2016 Completed n=119	Linkage will be determined using the LOD score method and mutations in specific genes will be assessed using a combination of residue conservation, BLOSUM score, and molecular modeling. Association will be determined using chi-square and Fisher exact tests. Biochemical, metabolic, and physiological effects will be individualized to the specific assay.	NR	Individuals or family members of individuals with retinal degenerations, either congenital, childhood, or age-related	Diseases, infections, or trauma that mimic primary retinal degenerations

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Natural History and Genetic Studies of Usher Syndrome NCT00106743 National Eye Institute	Prospective, observational study	To explore clinical and genetic aspects of Usher syndrome	March 2005 NR Ongoing but not recruiting n=237	Affected participants will be phenotypically categorized in 1 of the 3 clinical types, based on audiology and vestibular findings.	NR	Documented neurosensory hearing loss and retinitis pigmentosa and fulfill the clinical characteristics for Usher syndrome type 1, 2, or 3 as defined by the Usher syndrome consortium or be unaffected family members of a proband with Usher syndrome, primarily parents and siblings. Family members will be considered unaffected by history if they have had previous normal ophthalmologic and hearing examinations and if they don't have decreased night or peripheral vision.	Had intrauterine infection, perinatal/ congenital infections, or intrauterine and birth complications. These conditions can result in damage to both the auditory and visual system. Have concurrent inherited or acquired conditions that affect the visual and/or auditory system and significantly alter the phenotype

NR=not reported