

# Genotype-Phenotype Study of Patients With Plaquenil -Induced Retinal Toxicity, With Evaluation of the ABCA4 Gene

NCT01145196

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**Status** RECRUITING  
**Sponsor** National Eye Institute (NEI)  
**Enrollment** 320 participants

## Key Eligibility Criteria

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### Inclusion (12)

- Affected participants must be 18 years of age or older and have:
  - History of systemic lupus erythematosus (SLE), rheumatoid arthritis (RA) or Sj(SqrRoot)(Delta)gren s syndrome, and
  - History of Plaquenil use, and
  - Evidence of Plaquenil -induced retinal toxicity, based on clinical findings.
  - Unaffected volunteers must be 18 years of age or older and have:
- ... and 7 more (see full listing online)

### Exclusion (1)

- \. Participants with other known (genetic) retinal disease including but not limited to: Stargardt s disease and cone or cone-rod dystrophy whose diagnosis preceded their Plaquenil use. Participants with no known previous genetic diagnosis but with clinical findings associated with a genetic diagnosis, such as parafoveal or macular flecks which are associated with Stargardt s disease or fundus flavimaculatus, will also be excluded.

## Locations (1 total)

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National Institutes of Health Clinical Center, Bethesda, Maryland, United States