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Gene test information

COMPLEMENT FACTOR H (CFH) AND AGE-RELATED MACULAR DEGENERATION (AMD)

- **Background**

  Age-related macular degeneration (AMD) is a disease that causes progressive damage to the macula. Macula is the central part of the retina that allows us to see fine details. When the macula degenerates, people experience blurring or darkness in the center of their vision. Macular degeneration leads to loss of central vision needed for activities requiring fine vision such as reading, driving and recognising faces.

  Complement factor H (CFH) gene has been determined to be strongly associated with a person's risk for developing AMD. A tyrosine (Y) to histidine (H) change at amino acid 402 in complement factor H results in the formation of a CFH gene variant. People whose genetic makeup includes this variant of the CFH gene are more likely to develop AMD.

- **CFH genotypes**

Genotype	Frequency	Commentary
CFH 402 YY :	63%	Wild-type ("normal") genotype. No increased risk for AMD
CFH 402 YH :	32%	Heterozygous carrier of a 402H allele. Risk for AMD increased about 4-fold compared to wild-type genotype..
CFH 402 HH :	5%	Homozygous carrier of two 402H alleles. Risk for AMD increased about 12-fold compared to wild-type genotype.

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- **Indications for testing**

  - Estimation of individual risk for AMD

  - Optimization of therapy in AMD patients

References:

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Montezuma SR, Sobrin L, Seddon JM. Review of genetics in age related macular degeneration. Semin Ophthalmol. 2007;22:229-40.

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