Genotyping IDs long-term risk of macular degeneration

13 November 2012

Genotyping of two genetic risk alleles can be used to estimate the long-term risk of early and late age-related macular degeneration (AMD), but knowing the phenotype is important in assessing risk when early AMD is present, according to a study published online Nov. 9 in the *Archives of Ophthalmology*.

(HealthDay)—Genotyping of two genetic risk alleles can be used to estimate the long-term risk of early and late age-related macular degeneration (AMD), but knowing the phenotype is important in assessing risk when early AMD is present, according to a study published online Nov. 9 in the *Archives of Ophthalmology*.

Ronald Klein, M.D., M.P.H., from the University of Wisconsin School of Medicine and Public Health in Madison, and colleagues described the relationships of risk alleles in complement factor H (*CFH*) and age-related maculopathy susceptibility 2 (*ARMS2*) with the incidence and progression of AMD in a cohort including 4,282 individuals followed during a 20-year study. Genetic risk was defined as low (presence of zero to one risk allele; 2,820 individuals); intermediate (two risk alleles; 1,129 individuals); and high (three to four risk alleles; 333 individuals).

The researchers found that the five-year incidence of early AMD was 9.1 percent and of late AMD was 1.6 percent, both of which increased with age. Of the participants aged 45 years with no AMD, 33.0, 39.9, and 46.5 percent, respectively, in the low, intermediate, and high genetic risk groups were estimated to develop early AMD, while 1.4, 5.2, and 15.3 percent, respectively, were estimated to develop late AMD by age 80 years.

"The value of risk assessment will be determined as the pathogenesis of the disease becomes better understood and new evidence emerges to support cost-effective interventions before onset or at the earliest stages of the disease," the authors write. "For now, knowing the phenotype when early AMD is present contributes more to risk assessment than knowing the genetic risk based on the two AMD candidate genes with the largest attributable risk."

More information: Abstract
Full Text (subscription or payment may be required)