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New Genes Involved in Human Eye Color Identified

Three new genetic loci have been identified with involvement in subtle and quantitative variation of human eye color.

The study, led by Manfred Kayser of the Erasmus University Medical Center Rotterdam, The Netherlands, is published May 6 in the open-access journal PLoS Genetics.

Previous studies on the genetics of human eye color used broadly-categorized trait information such as 'blue', 'green', and 'brown'; however, variation in eye color exists in a continuous grading from the lightest blue to the darkest brown.

In this genome-wide association study, the eye color of about 6000 Dutch Europeans from the Rotterdam Study was digitally quantified using high-resolution full-eye photographs. This quantitative approach, which is cost-effective, portable, and time efficient, revealed that human eye color varies along more dimensions than are represented by the color categories used previously.

The researchers identified three new loci significantly associated with quantitative eye color. One of these, the *LYST* gene, was previously considered a pigmentation gene in mice and cattle, whereas the other two had no previous association with pigmentation.

These three genes, together with previously identified ones, explained over 50% of eye color variance, representing the highest accuracy achieved so far in genomic prediction of complex and quantitative human traits.

"These findings are also of relevance for future forensic applications," said Kayser, "where appearance prediction from biological material found at crime scenes may provide investigative leads to trace unknown persons."

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