

New genes involved in human eye color identified

May 6 2010



Three new genetic loci have been identified with involvement in subtle and quantitative variation of human eye colour. 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 colour used broadly-categorized trait information such as 'blue', 'green', and 'brown'; however, variation in eye colour exists in a continuous grading from the lightest blue to the darkest brown. In this genome-wide association study, the eye colour 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 colour varies along more dimensions than are represented by the colour categories used previously.

The researchers identified three new loci significantly associated with quantitative eye colour. 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 colour 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".

More information: Liu F, Wollstein A, Hysi PG, Ankra-Badu GA, Spector TD, et al. (2010) Digital Quantification of Human Eye Color Highlights Genetic Association of Three New Loci. PLoS Genet 6(5): e1000934. doi:10.1371/journal.pgen.1000934

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