

What is the Cause of Blue Eye Color in Humans?

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University of Southern Maine

Thinking Matters

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What is the Cause of Blue Eye Color in Humans?

Approximately 16 different genes are responsible for human eye color. This literature review will discuss some changes in one of these genes, OCA2, a major contributor to eye color. One interval on chromosome 15 which contains the OCA2 gene and the OCA2 locus is found to be responsible for an estimated 74% of the variance in human eye color. Expression of OCA2 produces P protein in melanocytes. Melanocytes produce melanin, the substance that gives eyes their color. Two adjacent genes on chromosome 15, HERC2 and OCA2, are so close together that an intron of HERC2 contains the promoter region for OCA2, affecting its expression. Linkage analysis of a Danish family identified two SNPs within a region of HERC2 that are associated with blue and brown eye colors: rs12913832 and rs1129038. Rs12913832 was found to be located upstream from the OCA2 promoter in a conserved sequence in intron 86 of HERC2. This conserved region around rs12913832 controls the necessary expression of OCA2 and the C allele at rs12913832 reduces the activity of the OCA2 promoter specifically within iris melanocytes. We discuss how blue eye color in humans is due to a common founder mutation in an OCA2 inhibiting regulatory element dating back approximately 10,000 years.

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