Age-related macular degeneration associated polymorphism rs10490924 in ARMS2 results in deficiency of a complement activator.

Micklisch S, Lin Y, Jacob S, Karlstetter M, Dannhausen K, Dasari P, von der Heide M, Dahse HM, Schmölz L, Grassmann F, Alene M, Fauser S, Neumann H, Lorkowski S, Pauly D, Weber BH, Joussen AM, Langmann T, Zipfel PF, Skerka C (2017) Age-related macular degeneration associated polymorphism rs10490924 in ARMS2 results in deficiency of a complement activator. *J Neuroinflammation* 14(1), 4. PubMed

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Abstract

Age-related macular degeneration (AMD) is the leading cause of blindness in developed countries. The polymorphism rs10490924 in the ARMS2 gene is highly associated with AMD and linked to an indel mutation (del443ins54), the latter inducing mRNA instability. At present, the function of the ARMS2 protein, the exact cellular sources in the retina and the biological consequences of the rs10490924 polymorphism are unclear.

Identifier

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