

Genetics and Age Related Macular Degeneration

Ruchi Shrestha^{1*}

¹Department of Ophthalmology, Vitreoretina Surgeon, Reiyukai Eiko Masunaga Eye Hospital, Banepa, Kavre, Nepal

ABSTRACT

Age related macular degeneration is a multifactorial disease involving genetic and environmental factors. Newer drugs targeting complement pathway (Eculizumab, Lampalizumab), gene therapy and stem cell therapy are recent innovations for the treatment of age related macular degeneration.

Keywords: Age related macular degeneration (ARMD), Genetics

INTRODUCTION

Age-related macular degeneration (ARMD) is a multifactorial disease involving genetic and environmental factors. A genetic understanding of ARMD allows for early diagnosis and treatment. Risk factors are old age, smoking, obesity, hypertension, cardiac disease and sedentary life style.

20 percent ARMD have positive family history. First degree relatives of patients with late AMD have four fold increased risk of ARMD. ARMD has racial association. Caucasians, Whites, Asians appear more susceptible to ARMD and lower in Blacks.

Two specific genetic locus ARMD 1 and HTRA 1 located on chromosome 1q and 10q respectively likely accounts for more than 50% of AMD. HTRA1 is one of the most significant gene that may weaken Bruch's membrane and promote angiogenesis in advanced choroidal neovascularization (wet ARMD). Inhibition of complement cascade plays a great role in the genetic treatment of AMD.

The management of AMD varies from reducing genetic risk, anti-vascular endothelial factor, gene and stem cell therapy. High dietary intake of nutrients along with anti-oxidants (zinc and omega -3 fatty acids) reduces the risk of early AMD. Rotterdam study and Blue mountains eye study showed that weekly consumption of fish was associated with 40% reduced risk of late AMD. Newer drugs like Eculizumab and Lampalizumab (Anti-vascular endothelial growth factor) targets the complement pathway and are shown to be effective for the treatment of British woman has become the first in the world to undergo gene therapy for wet ARMD in Oxford university, UK AMD. Implantation of a specially engineered patch of retinal pigment

epithelium cells derived from stem cells has been described successful for the treatment of treat wet AMD.

CONCLUSION

In conclusion Genetic research on AMD has made enormous progress over the last decade. Lifestyle modification may help prevent AMD in genetically susceptible individuals. Currently there are no studies firmly demonstrating that genetic testing should influence AMD treatment selection. In future it is likely that genetic testing will be more practical and helpful in guiding treatments for AMD.

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Correspondence to: Ruchi Shrestha, Department of Ophthalmology, Vitreoretina Surgeon, Reiyukai Eiko Masunaga Eye Hospital, Banepa, Kavre, Nepal, Tel: 9847027650; E-mail: dr_ruchishrestha@gmail.com

Received: December 20, 2021; **Accepted:** August 30, 2021; **Published:** September 09, 2021

Citation: Shrestha R (2021) Genetics and Age Related Macular Degeneration. *J Clin Exp Ophthalmol*. 12:p115.

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