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# BRCA1/2 mutations are not associated with risk of exudative age-related macular degeneration in ashkenazi Jews

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**Purpose:** Age-related macular degeneration (AMD) is pathophysiologically linked to abnormal DNA repair and increased expression of vascular endothelial growth factor (VEGF). The *BRCA1/2* gene is involved in maintaining genome stability and the increase in VEGF in tumors with a BCRA1/2 mutation. Therefore, we hypothesized that *BRCA1/2* mutations may be associated with the development of exudative (wet) AMD and neovascularization together with age-related DNA damage.

**Methods:** Genomic DNA was extracted from peripheral blood leukocytes of 52 Ashkenazi Jewish patients with wet AMD and tested for mutations in BRCA1 (c.5382insC and 185delAG) and BRCA2 (c.6174delT) using high-resolution melt analysis and direct sequencing.

**Results:** No BRCA1 mutations were found and only one patient (1.92%) carried the BRCA2 mutation.

**Conclusions:** The findings suggested that a disruption in the *BRCA1/2* mechanism plays no role in age-related neovascularization of the wet macula in this ethnic group.

## Biography

Shirel Rossnewasser-Weiss is a 3rd year PhD student at the Krieger Eye Research Laboratory, The Felsenstein Medical Research Center, Tel Aviv University. During her studies she was trained for basic science vision research. So far, her studies have focused on genetics of degenerative retinal disease and bioinformatics analysis. She is also involved in ocular and brain tumors studies, pathophysiology of ocular diseases, neuroprotection and stem cell therapy. She participated in national and international conferences including ARVO and her publications include 4 papers in peer review journals in which she was a co-author.

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**Notes:**