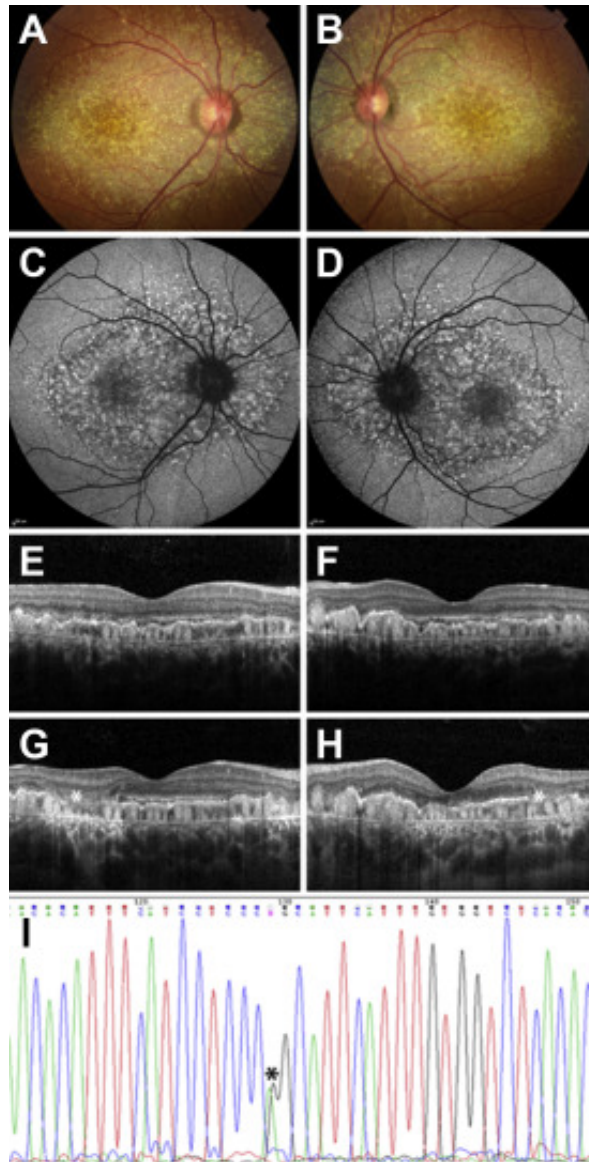


# Malattia Leventinese: EFEMP1 R345W Variant Is a Hot Spot Mutation, Not a Founder Mutation



A 26-year-old woman presented with reading difficulty. Her visual acuity was 0.8 bilaterally and fundus examination showed multiple drusen deposits at posterior pole and nasal to the disc, right eye and left eye (**A**, **B**). Drusen were hyperautofluorescent on autofluorescence imaging (**C**, **D**). OCT showed diffuse hyper reflective material under the retinal pigment epithelium corresponding with the large and small drusen (**E**, **F**). Six years, later OCTs (**G**, **H**) show alteration of the retinal pigment epithelium layer temporal to the fovea (\*), with resorption of some large drusen. Electropherogram showing NM\_001039349.2: c.1033C>T

heterozygous de novo variant EFEMP1 gene (I, \*).

## **Article info**

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