

The prevalence of ocular albinism has been reported to be one male in 20,000 births. The diagnosis of ocular albinism is based on the characteristic eye findings. Female relatives who carry the gene for ocular albinism will have some retinal pigment abnormalities (seen as mild iris transillumination) but usually will not have the visual changes observed in affected males. Very rarely females can be affected with the hallmarks of OA1 including nystagmus and foveal hypoplasia with reduced visual acuity. Molecular genetic testing for GPR143 gene detects mutations in approximately 90% of affected males and is available to confirm the diagnosis.