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Title:	Exome sequencing identifies procollagen-lysine 2-oxoglutarate 5-dioxygenase 2 mutations in primary congenital and juvenile glaucoma
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Keywords:	Exome sequencing procollagen-lysine 2-oxoglutarate 5-dioxygenase 2 mutations
Issue Date:	2021
Publisher:	IJO
Citation:	Indian Journal of Ophthalmology, 69(10), 2710-2716.
Abstract:	To report the association of procollagen-lysine 2-oxoglutarate 5-dioxygenase 2 (PLOD2) mutations with bilateral primary congenital glaucoma (PCG) in monozygotic twins and with nondominant juvenile-onset primary open-angle glaucoma (JOAG). Methods: We utilized family-based whole-exome sequencing to detect disease-causing mutations in a pair of monozygotic twins with de-novo PCG and compared its existence in 50 nonfamilial cases of JOAG and 30 healthy controls. To validate the identified mutations, direct Sanger sequencing was performed. For further evaluation of gene expression in the ocular tissues, we performed whole-mount in situ hybridization in zebrafish embryos. Results: We identified a novel missense mutation (c.1925A>G, p.Tyr642Cys) in the PLOD2 gene in the monozygotic twin pair with PCG and another missense mutation (c.1880G>A, p.Arg627Gln) in one JOAG patient. Both mutations identified were heterozygous. Neither the parents of the twins nor the parents of the JOAG patient harbored the mutation and it was probably a de-novo change. The zebrafish in situ hybridization revealed expression of the PLOD2 gene during embryogenesis of the eye. Conclusion: We observed an association of PLOD2 mutations with PCG and with nonfamilial JOAG. This new gene needs to be further investigated for its role in pathways associated with glaucoma pathogenesis.
Description:	Only IISERM authors are available in the record.
URI:	https://doi.org/10.4103/ijo.ijo_1750_21 (https://doi.org/10.4103/ijo.ijo_1750_21) http://hdl.handle.net/123456789/4653 (http://hdl.handle.net/123456789/4653)
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