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DOWN-EXPRESSION OF RHO GENE IN EGYPTIAN PATIENT WITH THREE REGULATIVE REGION VARIANTS COULD LEAD TO RETINITIS PUNCTATA ALBESCENS PHENOTYPE

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Retinitis punctata albescens is a rare form of pigmentary retinopathy, generally inheredited as an autosomal recessive trait. It exhibits the same clinical phenotype of a classic pigmentary retinopathy and it is characterized by diffusely scattered white, dot-like lesions localized deep to the retinal vessels and associated with night vision decrease. Electroretinogram exams show scotopic system involvement with total extinction at advanced stages. In most cases, retinitis punctata albescens is caused by mutation in RLBP1, but several evidences have shown that variants in RHO, PRPH2 or RDH5 genes could also determine the onset of disease. Our work investigated the role of three regulative variants in the RHO gene. Two are promoter region variants, c. -51 G>A (rs2269736) and c. -26 A>G (rs7984), in heterozygosity and homozygosity respectively. The third is the 3'-UTR variant c.*140delT (rs796098464), present in heterozygosity. All variants were detected in a 4-year- old Egyptian patient with retinitis punctate albescence clinically confirmed diagnosis. The effects of previously cited variants on RHO expression were firstly predicted by several bioinformatics platforms (Genomatix Software Suite v.3.10 and geneXplain web edition 4.11, supported by TRANSFAC database), then experimentally validated by Dual-Luciferase Reporter assay. Obtained results showed that rs2269736 and rs7984 variants caused a significant expression reduction in mutated RHO promoter, compared to wild-type one. A strong decreased gene expression was hypothesized by coexistence of both variants, with a major effect exerted by the homozygous rs7984, which deleted binding sites for 47 transcription factors. Summarizing, a high downregulation of RHO was evaluated due to combination of three variants. Decrease of RHO enzymatic activity could lead to poor post- Golgi trafficking, dysregulative activation, rod outer segment instability and arresting binding, probably determining rod apoptosis and retinitis punctate albescence etiopathogene.

BIOGRAPHY

Luigi Donato was born in Catania on 9th July 1986, graduated in Biology from University of Messina, Italy. He has completed his PhD in "Applied Biology and Experimental Medicine" at the age of 32 years from University of Messina, and he frequents the Laboratories of Molecular Genetics, Department of Biomedical and Dental Sciences and Morpho-functional Imaging, of the same University. He published more than 25 papers in reputed journals and participated in more than 20 national and international congresses. Moreover, he is a member of "Association of Research in Vision and Ophthalmology" (ARVO) and of "Associazione Italiana di Biologia e Genetica" (AIBG). His main research fields are focused on retinal dystrophies and omics approaches.

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