Mohammad Yahya Vahidi Mehrjard et.al., J Mol Genet Med 2018, Volume 12
DOI: 10.4172/1747-0862-C1-024

A Novel Missense Mutation in the ALDH13 Gene Causes Anophthalmia in Two Unrelated Iranian Consanguineous Families

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Anophthalmia or microphthalmia (A/M) describe a rare group of congenital/developmental ocular malformations, characterized by absent or small eye within the orbit affecting one or both eyes. They have complex etiology with chromosomal, monogenic and environmental causes. From the genetic point of view, it is a highly heterogeneous disease and can be caused by mutations in several different genes. We performed genome SNP-array analysis followed by autozygosity mapping and sequencing in three patients with severe bilateral anophthalmia from two unrelated consanguineous families. The genetic analysis revealed a homozygous missense mutation, causing a substitution of glycine (Gly) to arginine (Arg) at residue 237 of Aldehyde Dehydrogenase 1 (ALDH1A3). This study consolidates the importance of ALDH1A3 gene screening in autosomal recessive anophthalmia.

Biography

Mohammad Yahya Vahidi Mehrjardi has completed his PhD in molecular genetics at the age of 29 years from Science and Research Branch Islamic Azad University. He is the Head of Genetics Unit, Yazd Diabetes Research Center and Manager of the Medical Genetics Research Center Shahid Sadoughi University of Medical Sciences. He has published more than 13 papers in reputed journals and he has written a book about diabetes and genetic factors. he has been awarded a science prize of 7thYazd International Student Award in Reproductive Medicine.

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