Insight Medical Publishing

Journal of Clinical Epigenetics

Journal of Clinical Epigenetics
ISSN: 2472-1158

2020
Vol.6 No.4

Molecular diagnosis of retinal disorder with next-generation sequencing masquerading as hereditary optic neuropathy: A report of 2 cases

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Abstract

The initial clinical diagnosis was hereditary optic neuropathy, after next-generation sequencing the final diagnosis are changed on 2 patients with retinal diseases. A 19 years old male visit my clinic for the evaluation of low vision. His cycloplegic refration was – sph 5.50 + cyl 0.25 Ax 105 on right eye, and – sph 5.00 + cyl 0.50 Ax 80. His best corrected vision was 20/100 on both eyes at initail visit. He has the maculo-papillary bundle defect on both eyes. And visual field test showed central scotoma on bothy eyes. The first case was identified the ABCA4 mutant variant carried compound heterozygous variant.

A 19 years old male visit my clinic for the evaluation of low vision and color blindness. His cycloplegic refration was – sph 9.25 + cyl 0.75 Ax 105 on right eye, and –sph 9.75 + cyl 1.00 Ax 75. His best corrected vision was 20/100 on right eye and 20/70 on left eye at initail visit. He had temporal disc pallor and large cupping on both optic discs. And visual field defect showed superotemporal defect on right eye and central scotoma at left eye. The second case was identified the heterozygous R838C mutation in the GUCY2D gene.

Next-generation sequencing (NGS) technology allowed more patients to obtain a accurate molecular diagnosis. Although in small proportion of patients precision care can be provided, these findings are significant as individualized management can be achieved through genetic testing.

Biography:

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13th International Conference on Genomics and Molecular Biology; May 25-26, 2020 Webinar

Abstract Citation:

Hye Won Park, Molecular diagnosis of retinal disorder with next-generation sequencing masquerading as hereditary optic neuropathy: A report of 2cases, Genomics 2020, 13th International Conference on Genomics and Molecular Biology; May 25-26, 2020 Webinar.

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