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A Study of the DNA Mutation Causing Leber Congenital Amaurosis (LCA) Using Gene Editing Program and Molecular Geometry Analysis JUSTIN NAMJOO KIM, ANDREW KYUNG, YOONJEONG KWON, Choice Research Group — Retinitis pigmentosa (RP) is a genetic eye disorder that causes visual impairment related to retinal degeneration. Symptoms of RP include a gradual functional decrease in night vision and peripheral vision, often leading to tunnel vision. It is rare, however, for a patient to experience complete blindness as a result of RP. Leber Congenital Amaurosis (LCA) is another genetic eye disorder that causes visual impairment in the form of light sensitivity. RP and LCA Type 2 (LCA2) genetic eye disorders have both been linked to a mutation in gene RPE65. RPE65 gene mutation has been commonly identified in LCA patients across a number of countries. This paper identifies and analyzes abnormalities in the genetic sequence that lead to mutations in patients with genetic eye disorders. First, a computational study was performed with genetic mutations identified in RP patients, then a computational study was also performed with pathogenic mutations identified in LCA patients, examining sequence abnormalities in their DNA. Finally, sterochemical analysis was performed to examine the thermodynamic stability of the previously proposed RPE65 O-alkyl cleavage mechanism.

Richard Kyung
Choice Research Group

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