



THE CARVER FAMILY CENTER
for MACULAR DEGENERATION

AT THE UNIVERSITY OF IOWA

Dear *INSURANCE CARRIER*,

We are writing to ask that your company consider including the costs of genetic testing for Leber congenital amaurosis as part of insurance coverage for your customers.

Leber congenital amaurosis (LCA) is an autosomal recessive eye condition that affects 1 in 80,000 infants and is characterized by profound vision loss.

At present, nine genes that can cause LCA have been discovered including: CEP290, AIPL1, CRB1, CRX, GUCY2D, LCA5, RDH12, RPE65, and RPGRIP1. Genetic testing for abnormalities in these genes is currently available from the Carver Non-profit Genetic Testing Laboratory (CNGTL) at the University of Iowa Hospitals and Clinics (<http://www.carverlab.org/β>). Ninety-three gene segments (out of 177 total segments that make up the complete coding regions of the nine LCA genes) have been previously reported to harbor disease-causing genetic variations. Certain of these regions are known to harbor more variations than others. In the test offered by the CNGTL, sixty-eight of these gene segments are screened in the order of the likelihood of discovering a genetic variation. These gene segments are screened with a combination of techniques including single strand conformation polymorphism analysis; allele-specific PCR; and automated DNA sequencing in a carefully crafted balance to result in the most reliable and cost effective testing. Once disease-causing variations are detected in one of the nine genes, testing of the other genes is halted and patients are only billed for the portion of the test that is completed. As a result, over half of the patients do not reach the upper end of the pricing scale for LCA, which is \$957 (nonprofit). This testing strategy identifies disease-causing variations in about 70% of LCA cases. A more complete description of this strategy is outlined at the CNGTL website (<http://www.carverlab.org/requesting-genetic-testy>). The CNGTL is a CLIA and CAP certified laboratory and has a long history of providing reliable genetic testing for inherited eye conditions such as LCA.

Genetic testing by the CNGTL provides useful information that helps physicians diagnose LCA with increased certainty. Patients with LCA frequently undergo intense work-ups for their vision loss at multiple tertiary care centers that include expensive evaluations and imaging studies. **Genetic testing by the CNGTL may provide firm support for a diagnosis of LCA and eliminate the need for additional costly studies to rule out other possible causes of vision loss.**

Best regards,

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