UNIVERSITY of IOWA HEALTH CARE

JOHN AND MARCIA CARVER
Nonprofit Genetic Testing Laboratory
CLIA# 16D0985634

Laboratory Results

CARVER FAMILY CENTER for MACULAR DEGENERATION

Patient Name: **Martz, Paul**
Accession Number: RPXL-13-004
Birth Date: 01/07/1963
Sex: Male
Report Date: 05/13/13

Ordering Physician
Kimura, Alan, MD
Colorado Retina Associates
400 N Indiana St Suite 310
Golden CO 80401
Phone: 303-261-1600
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X-LINKED RETINITIS PIGMENTOSA

MOLECULAR DIAGNOSIS: No disease-causing variations were identified in this patient in the RPGR and RP2 genes.

COMMENT: This test detected 2 genetic variations in this patient's DNA that are unlikely to be disease-causing. For reporting purposes, we use a system that combines known functional and association information about genetic variations to estimate their pathogenic probability (EPP). EPP divides variations into four categories with increasing pathogenic potential: 0) very unlikely to be disease-causing 1) unlikely to be disease-causing 2) possible disease-causing 3) probable disease-causing. Details about the calculation of these values can be obtained at www.carverlab.org or Stone, Am J Ophthalmol. 2007 Dec;144(6):791-811. It is important to note that over 50% of patients with XLRP will harbor mutations that would not be detected by this test. Therefore, a negative test does not significantly reduce the likelihood of this diagnosis when classic clinical findings are present.

TEST PROCEDURE: Genomic DNA was obtained from the patient's white blood cells. The patient's DNA was directly sequenced through a portion of the RP2 gene (codons 1-323) and through the subset of the RPGR gene that is most amenable to automated DNA sequencing (codons 1-800 & 1071-1152).

RESULTS: Automated DNA sequencing of this patient's DNA revealed 2 non-disease-causing variations (see list below).

RPGR - IVS7+84 del1aT hemizygous - EPP=0
RPGR - IVS9+67 G>A hemizygous - EPP=0

Signed electronically by Edwin M. Stone, MD, PhD, Date: 05/15/2013 14:41:13

The performance characteristics of this test were determined by the John and Marcia Carver Nonprofit Genetic Testing Laboratory. It has not been cleared or approved by the U.S. Food and Drug Administration (FDA). The FDA

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has determined that such clearance is not necessary. This test is for clinical purposes. It should not be regarded as investigational or for research. The laboratory is certified under the Clinical Laboratory Improvements Amendments of 1988 (CLIA) as qualified to perform high complexity clinical laboratory testing.

This test is performed under a license agreement with Roche Molecular Systems, Inc.

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