
Molecular Diagnostic Report

Patient Name:	MARTZ, PAUL	Accession #:	CMD-16-00116
Medical Record #:	07058790	Received:	1/15/2016
Date of Birth:	1/7/1963	Reference Lab No.:	
		Reference Lab:	COLORADO RETINA ASSOCIATES
		Physician(s):	Alan Kimura, MD

TEST: *RPGR* ORF15 sequence analysis

RESULTS: POSITIVE

INTERPRETATION: Paul Martz possesses a hemizygous reported mutation in *RPGR* ORF15, namely c.2763_2764delGG (Ref: Vervoort (2000) Nat Genet 25, 462).

Sequence variations are reported according to nomenclature by den Dunnen and Antonarakis (Hum Genet 109(1):121-124, 2001).

TECHNIQUES: Direct testing for mutations in *RPGR* ORF15 was performed by long range PCR amplification followed by Nextera XT DNA library preparation and MiSeq NGS sequencing. The reference sequence for *RPGR* ORF15 was NM_001034853.1.

LIMITATIONS: The sensitivity of this laboratory developed test is unknown at this time.

RECOMMENDATIONS: If interested, additional family members can be tested for the identified mutation.

If you have any questions concerning the information in this report, please feel free to contact Dr. John (Pei-Wen) Chiang (chiangj@ohsu.edu; 503-494-5838).

My electronic signature indicates that I have personally reviewed all diagnostic materials and formulated the final diagnosis

Signature completed by
John Chiang Ph.D.
Director of CMD Laboratory
Date Completed 02/26/2016