**Appendix 1: *RP1* alleles found in patients with either dominant *RP1*-associated disease or with an uncertain mode of inheritance**

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| **Patient** | **Allele** | **Affected exona** | **gnomAD Frequency** | **SIFT & PolyPhen-2** | **MutationTaster** | **Additional variant comments** |
| MEE6b | c.1625C>G  p.(Ser542\*) | 4 (BIF) | 12/282190 | Deleterious  n/a | Disease causing | Previously-described in association with arRP[1].  In trans with patient’s other variant by cloning and long-range PCR. |
| MEE6 | c.2041dupA  p.(Ile681Asnfs\*17) | 4 | 4/25076  (no homozygotes) | n/a | Disease causing | Novel mutation that falls within exon 4 hotspot associated with adRP.  Reported in a family member without a reported ocular phenotype in a publicly-available databasec.  In trans with patient’s other variant by cloning and long-range PCR. |
| CEI22989 | c.2028C>T p.(Arg677\*) | 4 | n/a | n/a | n/a | First reported mutation described causing *RP1*-associated adRP[2]. |
| CEI22989 | c.4250T>C p.(Leu1417Pro) | 4 | 383/281894  (no homozygotes) | Benign  Tolerated | Polymorphism | Interpreted here as a variant of unknown significance based upon frequency and pathogenicity predictions. |

a Affected protein domain is indicated as relevant (DCX amino acids 36-118 and 154-233; BIF amino acids 486-635)[3]

b Patient seen for genetic testing only but had a diagnosis of Leber congenital amaurosis and reported symptomatic onset at age 6 with “hand motion” visual acuity in each eye in his mid-40s. Family members were reportedly unaffected but were unavailable for genetic testing or clinical evaluation.

c http://geno2mp.gs.washington.edu/Geno2MP/#/gene/RP1/gene/0/0/0

References:

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3. Siemiatkowska AM, Astuti GDN, Arimadyo K, den Hollander AI, Faradz SMH, Cremers FPM, Collin RWJ. Identification of a novel nonsense mutation in RP1 that causes autosomal recessive retinitis pigmentosa in an Indonesian family. Mol Vis. 2012; 18:2411–9. [PMID 23077400]