Supplemental Table 1. Additional Mutations Identified				
Subject #	**Sex**	**Age (yrs)**	***CNGB3* Genotype**	**Additional Mutations**
BPE-022	M	31	c.1148delC:p.Thr383Ilefs*13[*]	*PDE6H* – c.35C>G:p.Ser12Ter[†]
CEI-001	M	18	c.1148delC:p.Thr383Ilefs*13[*]	*GNAT2* – c.370G>A:p.Val124Met[‡,§]
PCI-008	F	40	c.1148delC:p.Thr383Ilefs*13[*]	*PDE6C* – c.1755G>T:p.Lys585Asn[‡,||]
PCI-012	F	8	c.819_826del8:p.Arg274Val fs*12; c.1148delC:p.Thr383Ilefs*13	*CNGA3* – c.1618G>A:p.Val540Ile[‡,#]
PCI-017	M	35	c.1148delC:p.Thr383Ilefs*13[*]	*CNGA3* – c.513G>A:pTrp171Ter[**]

[*]Subject is homozygous for this mutation.

[†]This mutation was previously reported as disease causing by Kohl S, Coppieters F, Meire F, et al. A nonsense mutation in PDE6H causes autosomal-recessive incomplete achromatopsia. *Am J Hum Genet* 2012;91:527-532.

[‡]The likely pathogenicity for this mutation was determined using SIFT, PolyPhen-2, and PROVEAN analysis tools. SIFT (version 1.03 http://sift.jcvi.org/, accessed January 2016) results are reported to be tolerated if tolerance index > 0.05 or damaging if tolerance index ≤ 0.05. PolyPhen-2 (version 2.2.2 http://genetics.bwh.harvard.edu/pph2/, accessed January 2016) appraises mutations qualitatively as Benign, Possibly Damaging or Probably Damaging based on the model's false positive rate. PROVEAN (version 1.1 http://provean.jcvi.org/human_protein_batch_submit.php, accessed January 2016) results are reported as having a neutral or deleterious effect as determined by averaged delta alignment scores.

[§]Predicted pathogenicity is - SIFT: damaging, PolyPhen-2: possibly damaging, PROVEAN: neutral.

[||]Predicted pathogenicity is - SIFT: damaging, PolyPhen-2: probably damaging, PROVEAN: neutral.

[#]Predicted pathogenicity is - SIFT: damaging, PolyPhen-2: benign, PROVEAN: neutral.

[**]To our knowledge this mutation has not been previously reported; the early termination of this protein is expected to be damaging.

F = female; M = male