| rsID        | Allele frequency | Ensmbol   | Location                          | Regulation | Comment                           | Consequence  |
|-------------|------------------|-----------|-----------------------------------|------------|-----------------------------------|--------------|
| NM_201253.3:c.2688T>A | 98.7%            | CRB1      | exon 6 of 12 position 12 of 957 (coding, NMD) | Likely Pathogenic | 58.6%                                |
| NM_201253.3:c.2290C>T | 100%             | CRB1      | exon 6 of 12 position 12 of 957 (coding) | Likely Pathogenic | 51%                                |
| c.1673T>G   | 99.99%           | CRB1      | exon 9 of 12 position 232 of 907 (coding) | Likely Pathogenic | 67.1%                                |
| c.1612_1613insCTTA | UNCERTAIN SIGNIFICANCE | CRB1 | exon 6 of 12 position 496 of 957 (coding) | Likely Pathogenic | 90.1%                                |
| NM_201253.3:c.2533_2539del | 99.99%           | CRB1      | exon 12 of 12 position 89 of 792 (coding) | Likely Pathogenic | 97.7%                                |
| NM_201253.3:c.2017A>G | 99.99%           | CRB1      | exon 12 of 12 position 89 of 792 (coding) | Likely Pathogenic | 99.99%                               |
| c.1844G>T   | 99.99%           | CRB1      | exon 9 of 12 position 813 of 907 (coding, NMD) | Likely Pathogenic | 99.99%                               |
| c.2401A>T   | 74.3%            | CRB1      | exon 9 of 12 position 1 of 907 (coding) | Likely Pathogenic | 76%                                |
| NM_201253.3:c.2533_2539del | 99.99%           | CRB1      | exon 12 of 12 position 89 of 792 (coding) | Likely Pathogenic | 99.99%                               |
| NM_201253.3:c.2017A>G | 99.99%           | CRB1      | exon 12 of 12 position 89 of 792 (coding) | Likely Pathogenic | 99.99%                               |
| c.4142C>G   | 99.99%           | CRB1      | exon 9 of 12 position 813 of 907 (coding, NMD) | Likely Pathogenic | 99.99%                               |
| c.254G>A    | 99.99%           | CRB1      | exon 9 of 12 position 1 of 907 (coding) | Likely Pathogenic | 99.99%                               |
| c.717_718insG | 99.99%           | CRB1      | exon 9 of 12 position 1 of 907 (coding) | Likely Pathogenic | 99.99%                               |
| c.253T>C    | 66.5%            | CRB1      | exon 3 of 12 position 98 of 196 (coding) | Likely Pathogenic | 69%                                |
| NM_201253.3:c.3307G>A | 77.9%            | CRB1      | exon 9 of 12 position 166 of 907 (coding) | Likely Pathogenic | 100%                                |
| c.254G>A    | 99.99%           | CRB1      | exon 9 of 12 position 1 of 907 (coding) | Likely Pathogenic | 99.99%                               |
| c.2308G>C   | 77.9%            | CRB1      | exon 9 of 12 position 1 of 907 (coding) | Likely Pathogenic | 100%                                |
| c.1429G>A   | 48.7%            | CRB1      | exon 9 of 12 position 1 of 907 (coding) | Likely Pathogenic | 43%                                |
| p.Cys195Phe | 63.4%            | CRB1      | exon 6 of 12 position 673 of 957 (coding) | Likely Pathogenic | 62%                                |
| p.Glu1330SerfsTer11 | 96.4%            | CRB1      | exon 6 of 12 position 141 of 957 (coding) | Likely Pathogenic | 97.7%                                |
| p.Leu1012Ser | 77.9%            | CRB1      | exon 6 of 12 position 141 of 957 (coding) | Likely Pathogenic | 97.7%                                |
| p.Phe786LeufsTer10 | 77.9%            | CRB1      | exon 9 of 12 position 783 of 907 (coding) | Likely Pathogenic | 97.7%                                |
| p.Arg744Ter  | 77.9%            | CRB1      | exon 9 of 12 position 783 of 907 (coding) | Likely Pathogenic | 97.7%                                |
| p.Ser487Pro  | 77.9%            | CRB1      | exon 9 of 12 position 783 of 907 (coding) | Likely Pathogenic | 97.7%                                |
| p.Glu524Asp  | 77.9%            | CRB1      | exon 9 of 12 position 783 of 907 (coding) | Likely Pathogenic | 97.7%                                |
| p.Cys336Arg  | 77.9%            | CRB1      | exon 9 of 12 position 783 of 907 (coding) | Likely Pathogenic | 97.7%                                |
| p.Asn986ArgfsTer3 | 77.9%            | CRB1      | exon 9 of 12 position 783 of 907 (coding) | Likely Pathogenic | 97.7%                                |
| p.Pro987AlafsTer11 | 77.9%            | CRB1      | exon 9 of 12 position 783 of 907 (coding) | Likely Pathogenic | 97.7%                                |
| p.Ser611Pro  | 77.9%            | CRB1      | exon 9 of 12 position 783 of 907 (coding) | Likely Pathogenic | 97.7%                                |
| p.Leu556Pro  | 77.9%            | CRB1      | exon 9 of 12 position 783 of 907 (coding) | Likely Pathogenic | 97.7%                                |
| p.Leu538ProfsTer14 | 77.9%            | CRB1      | exon 9 of 12 position 783 of 907 (coding) | Likely Pathogenic | 97.7%                                |
| p.Ser487Pro  | 77.9%            | CRB1      | exon 9 of 12 position 783 of 907 (coding) | Likely Pathogenic | 97.7%                                |
| p.Cys336Arg  | 77.9%            | CRB1      | exon 9 of 12 position 783 of 907 (coding) | Likely Pathogenic | 97.7%                                |
| p.Asn986ArgfsTer3 | 77.9%            | CRB1      | exon 9 of 12 position 783 of 907 (coding) | Likely Pathogenic | 97.7%                                |
| p.Pro987AlafsTer11 | 77.9%            | CRB1      | exon 9 of 12 position 783 of 907 (coding) | Likely Pathogenic | 97.7%                                |
| p.Ser611Pro  | 77.9%            | CRB1      | exon 9 of 12 position 783 of 907 (coding) | Likely Pathogenic | 97.7%                                |
| p.Leu556Pro  | 77.9%            | CRB1      | exon 9 of 12 position 783 of 907 (coding) | Likely Pathogenic | 97.7%                                |
| p.Leu538ProfsTer14 | 77.9%            | CRB1      | exon 9 of 12 position 783 of 907 (coding) | Likely Pathogenic | 97.7%                                |
