

**Table 2. *CRB1* mutations identified in Czech probands with early-onset severe retinal dystrophy**

<b>Chromosomal position</b>	<b>DNA level</b>	<b>Protein level</b>	<b>ExAc frequency; mutated/ total alleles tested</b>	<b>EVS frequency; mutated/ total alleles tested</b>	<b>Comment</b>
1:197396763	c.2308G>A	p.(Gly770Ser)	0.00002486; 3/120682	No entry	
1:197403836	c.2843G>A rs140074554	p.(Cys948Tyr)	0.0002086; 25/119820	0.000231; 3/12983	False negativity on genotyping microarray in proband 1
1:197404114	c.3121A>G	p.(Met1041Val)	0.000008254; 1/121156	No entry	

ExAC = Exome Aggregation Consortium, EVS = exome variant server

NM\_201253.2 was used as the reference sequence