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Supplemental Data

Mutations in the Spliceosome Component *CWC27* Cause Retinal Degeneration with or without Additional Developmental Anomalies

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SUPPLEMENTAL DATA

Table S1. Detailed information of *CWC27* variants identified in this study

Hg19 coordinate (chromosome 5)	Ref	Alt	cDNA changes	Protein changes	ExAC frequency	gnomAD frequency
64,181,274	G	T	c.943G>T	p.Glu315*	Absent	Absent
64,081,406	G	A	c.495G>A	p.Leu167Glyfs*3	Absent	1 in 30,240
64,181,325	C	CA	c.1002dupA	p.Val335Serfs*13	Absent	Absent
64,082,455	G	A	c.599+1G>A	p.[Val166Lysfs*3; Val191Lysfs*3]	1 in 77,894	1 in 195,976
64,064,992	C	T	c.19C>T	p.Gln7*	1 in 121,230	1 in 252,340
64,081,338	C	T	c.427C>T	p.Arg143*	1 in 111,918	Absent
64,084,795	C	A	c.617C>A	p.Ser206*	1 in 120,770	Absent

Ref, reference allele; Alt, alternative allele; cDNA and proteins annotations were based on GenBank: NM_005869.3 and GenPept: NP_005860.2.

Table S2. Statistical evaluation of genotype ratio for *Cwc27* tm1b allele

Genotype	Expected proportion	# individuals	Observed proportion	z-value	H_0 rejected*	Population size
E12.5 *Cwc27*						
+/+	0.25	9	0.45	-2.066	No	20
+/-	0.50	8	0.40	0.894	No	
-/-	0.25	3	0.15	1.033	No	
E14.5 *Cwc27*						
+/+	0.25	4	0.33	-0.667	No	12
+/-	0.50	6	0.50	0.000	No	
-/-	0.25	2	0.17	0.667	No	
E16.5 *Cwc27*						
+/+	0.25	1	0.14	0.655	No	7
+/-	0.50	3	0.43	0.378	No	
-/-	0.25	3	0.43	-1.091	No	
E18.5 *Cwc27*						
+/+	0.25	5	0.28	-0.272	No	18
+/-	0.50	10	0.56	-0.471	No	
-/-	0.25	3	0.17	0.816	No	
***Cwc27* embryos from E12.5 to E18.5**						
+/+	0.25	19	0.33	-1.453	No	57
+/-	0.50	27	0.47	0.397	No	
-/-	0.25	11	0.19	0.994	No	
***Cwc27* Adult (56 litters)**						
+/+	0.25	111	0.62	-3.130	Yes	180
+/-	0.50	66	0.37	-3.614	Yes	
-/-	0.25	3	0.02	7.229	Yes	

+: *Cwc27*, wild-type allele; -, $Cwc27^{tm1b}$ mutant allele.

* Statistics: two-tailed Z-test of population proportion, H_0: observed proportion = Mendelian proportion, alpha level of 0.01, Z-score value |z| =2.576.