Supplementary Table S1. Distribution of OCT Types on *RS1* Variants.

Variants No.	Complementary DNA	Protein	Mutation type	1A	1B	2	3	4	5A	5B
1	c.78+1G>T	Splice_donor_variant & intron_variant	Truncated	1	1	0	0	0	0	0
2	c.97delT	p.Trp33fs	Truncated	2	0	0	0	0	0	0
3	c.130_140del	p.Gly44fs	Truncated	2	0	0	0	0	0	0
4	c.150G>A	p.Trp50* premature stop	Truncated	2	0	0	0	0	0	0
5	Exon 4 deletion		Truncated	2	0	0	0	0	0	0
6	c.306_308dupGCT	p.Leu103dup	Truncated	0	2	0	0	0	0	0
7	c.326+2T>G	Splice_donor_variant & intron_variant	Truncated	0	1	0	0	0	0	1
8	c.522+1G>T	Splice_donor_variant & intron_variant	Truncated	2	0	0	0	0	0	0
9	c.188G>A	p.Cys63Tyr	Missense	0	0	0	0	2	0	0
10	c.214G>A	p.Glu72Lys	Missense	1	0	0	0	5	0	0
11	c.286T>C	p.Trp96Arg	Missense	0	4	3	1	1	1	3
12	c.302C>A	p.Ala101Asp	Missense	2	0	0	0	0	0	0
13	c.305G>A	p.Arg102Gln	Missense	0	0	0	1	0	0	1
14	c.325G>A	p.Gly109Arg	Missense	0	0	0	0	0	2	0
15	c.328T>C	p.Cys110Arg	Missense	2	0	0	0	0	0	0
16	c.331G>A	p.Ala111Thr	Missense	0	0	0	0	0	2	0
17	c.404G>A	p.Gly135Glu	Missense	0	0	1	0	0	0	0
18	c.410T>C	p.Leu137Pro	Missense	2	6	2	0	0	0	0
19	c.421C>T	p.Arg141Cys	Missense	3	1	0	0	0	0	0
20	c.422G>A	p.Arg141His	Missense	6	4	0	0	0	0	0
21	c.426T>G	p.Cys142Trp	Missense	0	1	1	0	0	0	0
22	c.433G>T	p.Asp145Tyr	Missense	0	0	1	0	0	0	1
23	c.499A>G	p.Lys167Glu	Missense	0	0	0	0	0	0	2

24	**c.544C>T**	**p.Arg182Cys**	**Missense**	**1**	**0**	**0**	**0**	**0**	**1**	**2**
25	**c.574C>T**	**p.Pro192Ser**	**Missense**	**0**	**0**	**2**	**0**	**0**	**2**	**0**
26	**c.577C>T**	**p.Pro193Ser**	**Missense**	**2**	**0**	**0**	**0**	**0**	**0**	**0**
27	**c.578C>G**	**p.Pro193Arg**	**Missense**	**0**	**0**	**0**	**0**	**0**	**2**	**0**
28	**c.589C>T**	**p.Arg197Cys**	**Missense**	**6**	**4**	**1**	**2**	**0**	**0**	**0**
29	**c.590G>A**	**p.Arg197His**	**Missense**	**9**	**1**	**1**	**0**	**0**	**1**	**0**
30	**c.608C>T**	**p.Pro203Leu**	**Missense**	**1**	**0**	**0**	**0**	**0**	**0**	**0**
31	**c.617G>C**	**p.Trp206Ser**	**Missense**	**0**	**0**	**0**	**0**	**0**	**2**	**0**
32	**c.625C>T**	**p.Arg209Cys**	**Missense**	**4**	**2**	**1**	**0**	**0**	**1**	**1**
33	**c.638G>A**	**p.Arg213Gln**	**Missense**	**0**	**0**	**2**	**0**	**0**	**0**	**0**
34	**c.647T>C**	**p.Leu216Pro**	**Missense**	**0**	**4**	**0**	**0**	**1**	**0**	**3**
35	**c.668G>A**	**p.Cys223Tyr**	**Missense**	**1**	**0**	**0**	**0**	**0**	**0**	**1**
		Total (139 eyes)		**51**	**31**	**15**	**4**	**9**	**14**	**15**

A = adenine; Ala = Alanine; Arg = Arginine; Asp = Aspartic acid; C = cytosine; Coys = Cysteine; del = deletion; DS domain = discoidin domain; dup = duplication; fs = frameshift; G = guanine; Gln = Glutamine; Glu = Glutamic acid; Gly = Glycine; His = Histidine ; Leu = Leucine; Lys = Lysine; n = number; Pro = Proline; RS domain = Retinoschisin 1 domain; Ser = Serine; T = thymine; Thr = Threonine; Trp = Tryptophan; Tyr = Tyrosine; Val = Valine

Supplementary Table S2. Comparison of BCVA, Age, OCT Types Between the Truncating and Missense Variants in X-linked Retinoschisis Patients.

	Group 1 (Truncating mutation)	Group 2 (Missense mutation)	*P*-value
Mean ± SD in logMAR	**0.64 ± 0.42**	**0.70 ± 0.39**	**0.692**
Mean ± SD in age	**18.43 ± 10.76**	**15.16 ± 9.09**	**0.559**
1A (n=51 eyes)	**11 (21.6%)**	**40 (78.4%)**	**0.042**
Mean ± SD in logMAR	**0.53 ± 0.20**	**0.44 ± 0.18**	**0.233**
Mean ± SD in age	**15.76 ± 8.87**	**11.06 ± 5.27**	**0.119**
1B (n=31 eyes)	**4 (12.9%)**	**27 (87.1%)**	**0.814**
Mean ± SD in logMAR	**0.60 ± 0.27**	**0.71 ± 0.34**	**0.479**
Mean ± SD in age	**24.73 ± 15.21**	**13.59 ± 6.14**	**0.239**
2 (n=15 eyes)	**0 (0.0%)**	**15 (100.0%)**	**<0.001**
Mean ± SD in logMAR	**-**	**0.77 ± 0.27**	**-**
Mean ± SD in age	**-**	**15.06 ± 6.29**	**-**
3 (n=4 eyes)	**0 (0.0%)**	**4 (100.0%)**	**<0.001**
Mean ± SD in logMAR	**-**	**1.35 ± 0.47**	**-**
Mean ± SD in age	**-**	**13.61 ± 4.92**	**-**
4 (n=9 eyes)	**0 (0.0%)**	**9 (100.0%)**	**<0.001**
Mean ± SD in logMAR	**-**	**1.17 ± 0.44**	**-**
Mean ± SD in age	**-**	**13.29 ± 5.32**	**-**
5A (n=14 eyes)	**0 (0.0%)**	**14 (100.0%)**	**<0.001**
Mean ± SD in logMAR	**-**	**0.48 ± 0.27**	**-**
Mean ± SD in age	**-**	**20.64 ± 14.75**	**-**
5B (n=15 eyes)	**1 (6.7%)**	**14 (93.3%)**	**0.535**
Mean ± SD in logMAR	**2.00 ± 0.00**	**0.96 ± 0.40**	**0.159**
Mean ± SD in age	**22.65 ± 0.00**	**26.32 ± 10.52**	**0.908**

BCVA = best corrected visual acuity; logMAR = logarithm of the minimum angle of resolution; n = number; OCT = optical coherence tomography SD = standard deviation; XLRS = X-linked retinoschisis