

Table 1. Clinical features of patients with RB.

Patient	Sex	Minority Nationalities	Age (Months)	Onset Age (Months)		Fundus Appearance	
				LE	RE	LE	RE
1	M	Dai	12	6	6	RB	RB
2	M	Hani	106	14 *	17	-	RB
3	F	Bai	12	9	-	RB	-
4	F	Han	18	13	13	RB	RB
5	M	Hui	28	26	26	RB	RB
6	M	Yi	11	6	-	RB	-
7	M	Hani	60	54	-	RB	-
8	M	Han	6	5	5	RB	RB
9	M	Han	10	3	3	RB	RB

Abbreviations: M: male; LE, left eye; RE, right eye; Retinoblastoma (RB);The children of these families exhibited similar clinical features of RB. Ophthalmic examinations showed that the child in these families are affected by RB. * Removed at 15 months

Table 2 Variants identified by NGS in blood samples of RB children

No.	Locations on the chromosomes	Exon	Mutation		Pathogenic analysis	Allele	Protein	Cosegregation in family
1	chr13-48942673-48942675	11	c.1061_1062del	novel	pathogenic	het	p.R355Nfs*6	De novo
2	chr13-49030485	19	c.1960 G>C	known	Likely pathogenic	het	p.V654L	De novo
3	chr13-49039158	22	c.2236 G>T	novel	pathogenic	het	p.E746X	Heterozygous mother
4	chr13-49047526-49047530	24	c.2520+1_2520+4delGTGA	novel	pathogenic	het	splicing	De novo
5	chr13-48955538	17	c.1654 C>T	known	Pathogenic	het	p.R552X	De novo
6	chr13-49027168	18	c.1735 C>T	known	pathogenic	het	p.R579X	De novo
7	chr13-48881547	2	c.264+5G>A	novel	pathogenic	het	splicing	Heterozygous mother
8	chr13-48942685	11	c.1072C>T	known	pathogenic	het	p.R358X	De novo
9	chr13-48923160	6	c.607+1G>A	known	pathogenic	het	splicing	De novo