

		NO molecular testing		Proband <i>RB1</i> mutation found (96%) (15% for unilateral)				Proband <i>RB1</i> mutation NOT found (4%*)		
proband	relative	risk for <i>RB1</i> mutation	Surveillance plan	positive blood test		negative blood test		(nothing to test for relative)		
				risk for <i>RB1</i> mutation	Surveillance plan	risk for <i>RB1</i> mutation	Surveillance plan	risk for <i>RB1</i> mutation	Surveillance plan	
Conventional clinical care	proband	100%	EUAs	100%	EUAs	na	na	100%	EUAs	
	offspring	50% (0.5 x 1)	EUAs	100%	early delivery cancer surveillance	0.007%	clinic	50% (0.5 x 1)	EUAs	
	unaffected parent	4% (0.04* x 1)	retinal exam for retinoma	100%	retinal exam for retinoma cancer surveillance	4% (0.04* x 1)	retinal exam for retinoma	4% (0.04* x 1)	retinal exam for retinoma	
Personalized clinical care based on genetics	sibs	2% (0.04* x 1 x 0.5)	EUAs	100%	early delivery clinic visits EUAs cancer surveillance	0.007%	clinic	2% (0.04* x 1 x 0.5)	EUAs	
	bilateral									
	proband	15%	EUAs	100%	EUAs	0.6% (0.04 x 0.15)	clinic exams	15%	EUAs	
unilateral NO tumor available	offspring	7.5% (0.5 x 0.5)	EUAs	100%	early delivery clinic visits EUAs cancer surveillance	0.3% (0.04 x 0.15 x 0.5)	clinic exams	7.5% (0.5 x 0.5)	EUAs	
	unaffected parent	0.6% (0.04* x 0.15)	retinal exam for retinoma	100%	retinal exam for retinoma cancer surveillance	0.24% (0.04* x 0.04* x 0.15)	retinal exam for retinoma	0.6% (0.04* x 0.15)	retinal exam for retinoma	
	sibs	0.3% (0.04* x 0.15 x 0.5)	EUAs	100%	early delivery clinic visits EUAs cancer surveillance	0.012% (0.04* x 0.15 x 0.04* x 0.5)	clinic exams	0.3% (0.04* x 0.15 x 0.5)	EUAs	
	proband	15%	EUAs	100%	EUAs cancer surveillance	0.024% (0.04* x 0.15 x 0.04*)	clinic exams	15%	EUAs	
unilateral tumor <i>RB1</i> mutations found (96%)	offspring	7.5% (0.15 x 0.5)	clinic exams	100%	early delivery clinic visits EUAs cancer surveillance	0.012% (0.04* x 0.15 x 0.04* x 0.5)	clinic exams	7.5% (0.15 x 0.5)	clinic exams	
	unaffected parent	0.6% (0.04 x 0.15)	retinal exam for retinoma	100%	retinal exam for retinoma cancer surveillance	0.001% (0.04* x 0.15 x 0.04* x 0.04*)	population risk	0.6% (0.04* x 0.15)	retinal exam for retinoma	
	sibs	0.3% (0.04 x 0.15 x 0.5)	EUAs	100%	early delivery clinic visits EUAs cancer surveillance	0.0005% (0.04* x 0.15 x 0.04* x 0.04* x 0.5)	population risk	0.3% (0.04 x 0.15 x 0.5)	EUAs	
	proband	15%	EUAs	na	na	0.007%	population risk**	15%	EUAs	
unilateral tumor <i>RB1</i> normal, <i>MYCN</i> amplified (2%)**	offspring	7.5% (0.15 x 0.5)	clinic exams	na	na	0.007%	population risk**	7.5% (0.15 x 0.5)	clinic exams	
	unaffected parent	0.6% (0.04 x 0.15)	retinal exam for retinoma	na	na	0.007%	population risk**	0.6% (0.04 x 0.15)	retinal exam for retinoma	
	sibs	0.3% (0.04 x 0.15 x 0.5)	EUAs	na	na	0.007%	population risk**	0.3% (0.04 x 0.15 x 0.5)	EUAs	
	proband	15%	EUAs	na	na	0.007%	population risk**	15%	EUAs	

2 tumor mutations not detected in blood

Population risk 1:15000 per live birth 0.007% Risk for offspring of *RB1* mutant parent 50%
 **Risk to miss *RB1* mutant allele or that parent is unaffected carrier (most undetectable mosaic) 4% **Based on Rushlow et al 2013, unconfirmed na; not applicable