impact genetics inc.
advancing genetic diagnostics

# Retinoblastoma Risks and Surveillance Plans

| proband | | *NO molecular testing* | | Proband *RB1* Mutation Known (96%) *(15% for unilateral )* | | | | Proband *RB1* mutation NOT found (4%*) | |
|---|---|---|---|---|---|---|---|---|---|
| | | | | positive blood test for known mutation | | negative blood test for known mutation | | (nothing to test for relative) | |
| | | risk for *RB1* mutation | Surveillance plan | risk for *RB1* mutation | Surveillance plan | risk for RB1 mutation | Surveillance plan | risk for *RB1* mutation | Surveillance plan |
| bilateral | proband | 100% | EUAs | 100% | EUAs cancer surveillance | na | na | 100% | EUAs |
| | offspring | 50% (0.5 x 1) | EUAs | 100% | early delivery EUAs cancer surveillance | 0.007% | NO clinic exams | 50% (0.5 x 1) | EUAs |
| | unaffected parent | 5% (0.05¶ ) | retinal exam for retinoma | 100% | retinal exams for retinoma cancer surveillance | 0.2% (0.04* x 0.05¶) | retinal exam for retinoma | 5% (0.05¶ ) | retinal exams for retinoma |
| | sibs | 2.5% (0.05¶ x 0.5 ) | EUAs | 100% | early delivery EUAs cancer surveillance | 0.007% | NO clinic exams | 2.5% (0.05¶ x 0.5 ) | EUAs |
| unilateral NO tumor available | proband | 15% | EUAs | 100% | EUAs cancer surveillance | na | na | 0.6% (0.04* x 0.15) | clinic exams |
| | offspring | 7.5% (0.5 x 0.15) | EUAs | 100% | early delivery EUAs cancer surveillance | 0.007% | NO clinic exams | 0.3% (0.5 x 0.006) | clinic exams |
| | unaffected parent | 0.75% (0.05¶ x 0.15) | retinal exam for retinoma | 100% | retinal exams for retinoma cancer surveillance | 0.2% (0.04* x 0.05¶ ) | retinal exam for retinoma | 0.03% (0.05¶ x 0.006) | retinal exams for retinoma |
| | sibs | 0.38% (0.05¶ x 0.15 x 0.5) | EUAs | 100% | early delivery EUAs cancer surveillance | 0.007% | NO clinic exams | 0.015% (0.03* x 0.5) | clinic exams |
| unilateral **tumor** *RB1* mutations known (96%) | proband | 15% | EUAs | 100% | EUAs cancer surveillance | 0.3% (0.02§ x 0.15) | clinic exams | | |
| | offspring | 7.5% (0.5 x 0.5) | EUAs | 100% | early delivery EUAs cancer surveillance | 0.007% if negative for parental tumor mutations | NO clinic exams | | |
| | unaffected parent | 0.75% (0.05¶ x 0.15) | retinal exam for retinoma | 100% | retinal exam for retinoma cancer surveillance | 0.2% (0.04* x 0.05¶) | retinal exam for retinoma | | |
| | sibs | 0.38% (0.05¶ x 0.15 x 0.5) | EUAs | 100% | early delivery EUAs cancer surveillance | 0.007% | NO clinic exams | | |
| unilateral tumor *RB1* normal *MYCN* amplified (2%)∞ | proband | 15% | EUAs | na | na | 0.007% | NO clinic exams∞ | | |
| | offspring | 7.5% (0.5 x 0.5) | EUAs | na | na | 0.007% | NO clinic exams∞ | | |
| | unaffected parent | 0.75% (0.05¶ x 0.15) | retinal exam for retinoma | na | na | 0.007% | NO clinic exams∞ | | |
| | sibs | 0.38% 0.05¶ x 0.15 x 0.5) | EUAs | na | na | 0.007% | NO clinic exams∞ | | |

*Population risk 1:15000 per live birth 0.007%*
*Risk for offspring of RB1 mutant parent 50%*
**Risk to miss RB1 mutant allele 4%*
*¶Unaffected parental carrier rate (6/120 parents of bilaterals, Impact Genetics) 5%*
*§estimated risk to miss mosaicism for a known mutation 2%*
*∞based on Rushlow et al. The lancet oncology. 2013 Apr;14:327-34 (unconfirmed)*
*na, not applicable*

Conventional clinical care

Personalized clinical care based on genetics

*Note: Data based on research by Dr. B Gallie and Impact Genetics. Not to replace best judgement of medical practitioner.*