

‘Insight’ into Molecular Genetic Testing in Retinoblastoma

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ABSTRACT Retinoblastoma is an intraocular tumor with hereditary and sporadic forms. Tumor develops when both alleles of the tumor suppressor gene, RB1 is inactivated in the embryonal retinal cells. Clinical screening for retinoblastoma is most often done on examination under anesthesia for proband and closely related infants. Genetic testing in retinoblastoma could avoid unwanted ophthalmic surveillance for infants who do not have mutations in the gene. However establishing a single genetic test is not possible due to the multiple RB1 inactivation mechanisms like gene mutations, deletions and also hypermethylation of the gene promoter. The review focuses on the application of molecular screening methods- multiplex PCR, sequencing of 27 exons of RB1 gene, methylation analysis of promoter region and peripheral blood karyotyping in retinoblastoma. The cost benefit analysis of genetic testing compared to conventional clinical management of retinoblastoma is also discussed. Clinical management of retinoblastoma could be aided by coordinated application of the molecular genetic testing.