Abstract

Introduction. Retinoblastoma is a childhood cancer of the retina originated by altered or null retinoblastoma protein (pRb) expression. Genetic alterations in both RB1 alleles in the retinal cells are required for the development of retinoblastoma. In the sporadic form, non-hereditary RB1 gene mutations take place in a single retinoblast cell, and are therefore only present in tumor DNA (somatic mutations). Sporadic retinoblastoma is primarily unilateral, lacks family history and has no risk of transmission to descendants. Genetic tests for detection of RB1 mutation has improved the identification of carriers and facilitated accurate genetic counseling. Objective. To identify mutations in the RB1 gene in Colombian patients with sporadic retinoblastoma by PCR-SSCP followed by sequence. Materials and methods. Four patients with sporadic retinoblastoma were analyzed by PCR-SSCP, followed by DNA sequencing to identify variations in the RB1 gene. Results. We identified five variations in RB1 gene: three new mutations (one germline and two somatic mutations), one new polymorphism and one already reported somatic mutation. Four mutations were found in three patients with unilateral retinoblastoma and one mutation was found in a patient with bilateral retinoblastoma. One of these was a germline mutation in a sporadic unilateral retinoblastoma that was not present in the parents or three siblings analyzed. Conclusions. Our results emphasize the importance of identifying mutations for genetic counseling and clinical management of sporadic retinoblastoma patients. Description of a new RB1 gene variant is interesting since there have been a small number of polymorphisms reported for this gene.

Keywords

Retinoblastoma/genetics, genes, retinoblastoma, mutations, polymorphism, genetic, Colombia.