Search for Hidden Germline Mutations in Bilateral Retinoblastoma

Oluchukwu Okonkwo. University of Pennsylvania, Bronx, NY

Retinoblastoma (RB) is a cancer in the retina that develops in children usually under the age of five and can cause blindness, loss of the affected eye, or death by metastasis. It is known that RB1 gene defects account for the majority of RB cases. However, recent studies have identified RB2/p130 genes in a subset of cases. Therefore, this study aims to determine if there are additional germline mutations in the RB2/p130 gene in the occurrence of bilateral retinoblastoma.