9. BILATERAL RETINOBLASTOMA IN CHILDREN

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Introduction. Retinoblastoma is the most common intraocular malignancy affecting children. The incidence of this disease varies by country from 3.4 to 42.6 cases per million live births. Retinoblastoma usually affects young children with the highest incidence in patients under 4 years of age. Retinoblastoma has no sex. About 60% of retinoblastoma cases are unilateral and 40% are bilateral. The most common causes are mutations of germinal or somatic origin. Retinoblastoma does not spread from one eye to another. When both eyes are affected and / or several tumors form, each tumor is born from a single retinal cell. All bilateral retinoblastomas are caused by genetic errors in the RB1 gene that can be inherited from a parent or can occur during the early development of the embryo. Bilateral retinoblastoma can develop an average of three tumors or can vary up to 14 tumors in both eyes. The risk of growing a new tumor decreases significantly after the age of three.

Aim of study. Genetic counseling should be offered to every parent with a child with retinoblastoma and patients with a family history of retinoblastoma. An analysis of the child's personal and family history and direct and indirect molecular studies should be performed by the geneticist. The risk of transmission depends on family history and the type of retinoblastoma. In the case of hereditary retinoblastoma, the risk of transmission is 50%. In the case of unilateral, unifocal, non-familial retinoblastoma, the risk of transmission is 5%. Genetic analysis in affected children may include the following molecular tests: Direct search for a constitutional mutation in the RB1 gene performed on constitutional DNA. The rate of mutation detection is very high in hereditary forms. No preferential mutations or "hot spots" have been identified in the RB1 gene. Indirect demonstration of the allele carrying the mutation in cases of family history. This test consists of identifying intragenic or RB1 flanking markers common to all affected family members. Tumor loss assessment of heterozygosity. This technique requires tumor material and allows the determination of the allele that remains and carries the mutation.

Methods and materials. The research was descriptive-observational, which was based on various scientific studies conducted in various countries on bilateral retinoblastoma in children.

Results. Indices such as the incidence and risk factors in the development of bilateral retinoblastoma in children have been studied, as well as methods of prophylaxis of bilateral retinoblastoma in various scientific studies.

Conclusion. Retinoblastoma involving both eyes occurs in 1/3 of patients, and is accompanied by a germline mutation RB1. Genetic testing determines the risk of retinoblastoma, and 50% of babies at genetic risk are born with already formed tumors. Children with bilateral retinoblastoma have a lifetime risk of second cancer, which is increased by radiation exposure. These patients should avoid radiotherapy, computed tomography and x-rays whenever possible and receive genetic counseling as adults so that they understand their risks and potential risks to their children. Children born to a parent with a history of retinoblastoma, especially bilaterally, should be tested for this cancer shortly after birth, as early detection of this cancer greatly improves the chances of treatment.