Aims:

To highlight a Hot Topic in ocular oncology from recent literature

Methods:

Papers discussed: van Hoefen Wijsard M, et al. Benign tumors in long-term survivors of retinoblastoma. Cancers 2021;13:1773, and Libbrecht S, et al. The rapidly expanding group of RB1-deleted soft tissue tumors: an updated review. Diagnostics 2021;11:430.

Results:

A subset of patients with hereditary retinoblastoma, especially males, are known to develop multiple benign lipomatous tumors during adolescence. A survey in the United States found the cumulative incidence of lipomatous tumors to be as high as 14% (95% CI, 8-22) by 60 years after diagnosis of RB in males. In females, however, the most common subsequent benign tumor was leiomyoma in 9 %. The lipomas are typically of spindle cell type, composed of mature adipocytes, bland spindle cells, and ropey collagen with or without atypical features, or of pleomorphic type, which also contain pleomorphic multinucleated floret-like giant cells. When occurring in non-retinoblastoma patients, molecular genetic studies show heterozygous deletions of 13q14, including RB1. Especially atypical or pleomorphic lipomas can undergo sarcomatous transformation. The RB1-deleted spectrum of benign tumors also includes myofibroblastoma, cellular angiofibroma, and acral fibromyxoma. The occurrence of any such benign neoplasm subsequent to hereditary retinoblastoma predicts a higher risk of also developing a second cancer, with hazard rate 1.8 (95% CI, 1.1-2.9).

Conclusions:

The general ophthalmologist should be aware, in particular, of later occurrence of multiple lipomas in survivors of hereditary retinoblastoma, especially because they may undergo malignant change and predict occurrence of second cancers.