Response to “BAP1 Germline Mutation Associated with Bilateral Primary Uveal Melanoma”

Martina C. Herwig-Carl^{a, b} Amit Sharma^a Charlotte Melzer^a Frank G. Holz^a
Karin U. Loeffler^{a, b}

^aDepartment of Ophthalmology, University Hospital Bonn, Bonn, Germany; ^bDivision of Ophthalmic Pathology, Department of Ophthalmology, University Hospital Bonn, Bonn, Germany

Dear Editor,

With great interest, we read the article by Yu et al. [1] “BAP1 Germline Mutation Associated with Bilateral Primary Uveal Melanoma.” We agree that identification of patients with BAP1 germline mutations represents a major challenge for ophthalmologists. However, we would like to draw your attention to a case reported by our group in 2019 of a woman with bilateral uveal melanoma (choroidal melanoma OD and iris melanoma OS) bearing a BAP1 germline mutation [2]. Our case is – to the best of our knowledge – the first case of a patient suffering from bilateral uveal melanoma in whom a BAP1 tumor predisposition syndrome (BAP1-TPDS) was identified. The patient was genetically tested after she had developed additional basal cell carcinomas, and the genetic analysis revealed a BAP1-TPDS. Since the patient had before been treated at different hospitals, the possible connection between the neoplasias had not been suspected earlier. While the iris melanoma immunohistochemically showed lack of BAP1 expression (supporting its development based on the BAP1-TPDS), the basal cell carcinomas exhibited unexpectedly a strong BAP1 immunoreaction. This finding indicates that the basal cell carcinomas may have developed independently from the BAP1-TPDS.

Conflict of Interest Statement

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References

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- 2 Melzer C, Sharma A, Peters S, Aretz S, Biswas A, Holz FG, et al. Basal cell carcinoma developing independently from BAP1-tumor predisposition syndrome in a patient with bilateral uveal melanoma: diagnostic challenges to identify patients with BAP1-TPDS. [Genes Chromosomes Cancer](#). 2019; 58(6):357–64.