Reviewer's report

Title: Birt-Hogg-Dube Syndrome Presenting as Multiple Oncocytic Parotid Tumors

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Reviewer: Fred H Menko

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Lindor et al. describe a case of Birt-Hogg-Dubé syndrome (BHD) who presented with oncocytic parotid tumors. The aim of this report is to indicate a possible association of BHD with this tumor type. In general, due to the rarity of BHD and the incomplete understanding of its pathogenesis and clinical expression, case reports on possible new associations are important.

However, the association of BHD with parotid tumors has been reported previously, including molecular (loss of heterozygosity) studies. Maffé et al. (Clin Genet 2011; 79: 345-354) stated: “in addition, we provide clinical and molecular evidence that parotid oncocytoma, so far reported in six BHDS cases, is associated with this condition, based on the observation of a patient with bilateral parotid involvement and marked reduction of the wild-type FLCN allele signal in tumor DNA”. Therefore, in my view, the description of an additional case of BHD with a parotid tumor should consist of a detailed clinical and molecular study and a review of previously described cases.

Minor comments: The final pathological diagnosis in this case is not entirely clear (Case report: …favors the diagnosis of Warthin tumors.”…Discussion: “it may be reasonable to include Warthin tumor somewhere in this spectrum”). “No direct link to mitochondrial…”; please clarify. General: Dubé instead of Dube. Add reference in relevant parts of the article (such as the classification of the parotid tumours and the statement that the mutation has been reported previously). Abstract, Introduction, Case report: fibrofolliculoma is considered to be the basic skin lesion, the appearance of trichodiscoma being dependent on sectioning level (Vincent et al., J. Am. Acad. Dermatol. 2003; 49: 698-705). Discussion: The clinical consequence for the patient and the family might be given in somewhat more detail. Is surveillance for dermatological findings needed? Figures: Figures 1 and 2 could be omitted. Instead, a figure showing the histological appearance of the parotid tumor could be added.

Level of interest: An article whose findings are important to those with closely related research interests

Quality of written English: Acceptable