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Titel des Beitrags:
Mutations in genes encoding PI3K-AKT and MAPK signaling define anogenital papillary hidradenoma.

Abstract:
Papillary hidradenoma (a.k.a. hidradenoma papilliferum) is a benign tumor of the anogenital region that almost exclusively arises in middle-aged Caucasian women. These tumors may recur and rare cases of malignant development have been reported. The genetic basis of papillary hidradenoma is currently unknown. Hence, we employed targeted high-coverage next generation sequencing interrogating 50 cancer-related genes and conventional Sanger sequencing to investigate the mutational landscape in a cohort of 15 cases. Additionally, we analyzed the HPV status of these tumors. Thirteen cases (87%) harbored mutations in cancer-related genes. Recurrent mutations in PIK3CA and AKT1 were present in 10 of the cases (67%). One PIK3CA mutated case had a concomitant STK11 mutation. Three cases harbored mutually exclusive mutations in BRAF, APC and ERBB4. The remaining two cases showed no mutations. None of the cases harbored DNA of human papilloma virus. Our results also provide evidence that -just as BRAF V600E mutations in hyperplastic polyps and benign nevi- a mutated driver gene does not imply malignant behavior per
se but may set the basis for malignant transformation. The latter point may explain why rare cases of papillary hidradenoma have been reported to take a malignant course. Lastly, our genetic data may suggest treatment avenues beyond conventional surgery for some of these tumors. © 2015 Wiley Periodicals, Inc.