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High-Grade Astrocytoma with Piloid Features: A Dual Institutional Review of Imaging Findings of a Novel Entity

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Abstract

High-grade astrocytoma with piloid features (HGAP) is a recently identified brain tumor characterized by a distinct DNA methylation profile. Predominantly located in the posterior fossa of adults, HGAP is notably prevalent in individuals with neurofibromatosis type 1. We present an image-centric review of HGAP and explore the association between HGAP and neurofibromatosis type 1. Data were collected from 8 HGAP patients treated at two tertiary care institutions between January 2020 and October 2023. Demographic details, clinical records, management, and tumor molecular profiles were analyzed. Tumor characteristics, including location and imaging features on MR imaging, were reviewed. Clinical or imaging features suggestive of neurofibromatosis 1 or the presence of *NF1* gene alteration were documented. The mean age at presentation was 45.5 years (male/female = 5:3). Tumors were midline, localized in the posterior fossa (*n* = 4), diencephalic/thalamic (*n* = 2), and spinal cord (*n* = 2). HGAP lesions were T1 hypointense, T2-hyperintense, mostly without diffusion restriction, predominantly peripheral irregular enhancement with central necrosis (*n* = 3) followed by mixed heterogeneous enhancement (*n* = 2). Two *NF1* mutation carriers showed signs of neurofibromatosis type 1 before HGAP diagnosis, with one diagnosed during HGAP evaluation, strengthening the HGAP-*NF1* link, particularly in patients with posterior fossa masses. All tumors were *IDH1* wild-type, often with *ATRX*, *CDKN2A/B, and NF1* gene alteration. Six patients underwent surgical resection followed by adjuvant chemoradiation. Six patients were alive, and two died during the last follow-up. Histone H3 mutations were not detected in our cohort, such as the common H3K27M typically seen in diffuse midline gliomas, linked to aggressive clinical behavior and poor prognosis. HGAP lesions may involve the brain or spine and tend to be midline or paramedian in location. Underlying neurofibromatosis type 1 diagnosis or imaging findings are important diagnostic cues.