

Characteristics of patients with a primary brain tumor undergoing panel genetic testing

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ABSTRACT

Background. Currently, there are no genetic testing guidelines for patients with a primary brain tumor (PBT). This population is largely understudied in terms of the age at diagnosis, tumor pathology, family history, and their relation to genetic contribution. We aimed to describe these patient-specific characteristics and family histories based on hereditary cancer multi-gene panel test (MGPT) results among patients with a PBT.

Methods. A total of 654 consecutive subjects were referred for MGPT at a diagnostic laboratory between March 2012-June 2016. Clinical data were ascertained from requisition forms. Statistical analyses were completed using STATA (v.13, College Station, TX).

Results. In our cohort, 67% (339/506) were diagnosed ≤ 50 years of age. Most PBTs were gliomas (293/558, 53%) or meningiomas (222/558, 40%). Test result positive astrocytomas were diagnosed at a significantly younger age than patients with negative and VUS results ($p=0.021$). Of 165 patients with available family history information, 95% ($n=157$) reported a family history of some cancer and 18% reported a family history of brain tumors. MGPT identified 104/654 (16%) individuals with mutations. Of these, 35 (34%) had an isolated PBT with no additional primary cancers. About half of identified genes predisposed a risk to PBTs, while the other half did not.

Conclusions. While no genetic testing criteria currently exist for PBTs, our data suggest earlier age of diagnosis is being utilized as an indicator for testing. Pathology can be helpful in narrowing down the differential diagnosis; however patients' PBT pathology can be atypical in relation to their hereditary cancer syndrome. Our data suggest PBTs can be the primary presenting cancer in hereditary syndromes. Family history evaluations are a beneficial risk assessment tool, particularly until testing criteria are developed. Further research is necessary for the development of solidified genetic testing criteria in the PBT population and more robust identification of at-risk individuals.

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