PANEL GENETIC TESTING FOR PRIMARY BRAIN TUMORS

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Background. Currently, there are no genetic testing or genetic counseling referral guidelines for patients with a primary brain tumor (PBT). This population is largely understudied in terms of the family history, tumor grade, pathology, and genetic contribution. Our aim was to describe patient-specific characteristics and family histories across mutation positive, negative, and VUS cohorts based on cancer-panel genetic test results among patients with a PBT.

Methods. Subjects were referred for multi-gene panel testing between March 2012 and June 2016. Clinical data was ascertained from requisition forms. The incidence of pathogenic mutations (including likely pathogenic), and variant of unknown significance were then calculated for each gene and/or patient cohort.

Results. Almost all tumors were glial (n=293, 53%) or meningeal pathology (n=222, 40%). Age of diagnosis differed significantly between glial and meningeal tumors (p<0.001). Glioblastoma grade was not predictive of a positive genetic testing result; however, trends showed more, high grade glioblastomas yielding a positive result (46/116) than low grade tumors (5/22). Of 654 subjects, panel testing identified 104 individuals (16%) with mutations. Genes most frequently yielding a positive result were: CHEK2 (20/104), BRCA2 (13/104), PMS2 (10/104), TP53 (8/104), and APC (8/104). Of 165 patients with family history information provided, nearly all (n=157, 97%) reported a family history of some cancer.

Conclusions. Further research is critical to establish testing criteria for PBTs, allowing appropriate identification of high-risk patients. Harboring a mutation can alter management and screening as individuals may be at risk for additional cancers.