Choroid plexus carcinomas are characterized by complex chromosomal alterations related to patient age and prognosis.

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Abstract

Choroid plexus carcinoma is a malignant brain tumor predominantly occurring in young children. Only limited data are available regarding the underlying molecular genetic alterations. Therefore, molecular inversion probe single nucleotide polymorphism (MIP SNP) arrays were performed on a series of 26 neuropathologically well-characterized choroid plexus carcinomas. Recurrent copy number losses of chromosomes 5, 6, 16, 18, 19, and 22 as well as gains of chromosomes 1, 2, 4, 12, and 20 were identified. Furthermore, GISTIC analysis identified significant recurrent gains of 17 genes in 9 regions, and recurrent losses of 96 genes in 14 regions. Clustering analysis separated choroid plexus carcinomas into two groups: one characterized by marked losses and the other characterized by gains across the chromosomes. Chromosomal losses of 9, 19p, and 22q were significantly more frequent in younger children (<36 months), whereas gains on chromosomes 7 and 19, and chromosome arms 8q, 14q, and 21q prevailed in older patients. Multivariate analysis revealed that loss of 12q was associated with shorter survival \(12 \pm 5\) months vs. \(86 \pm 8\) months; \(\text{mean} \pm \text{SD}; P = 0.001\)] and, in addition, 45 smaller chromosomal regions showing genetic alterations significantly associated with survival could be identified. The MIP SNP array profiles also contributed to the diagnosis of two difficult SMARCB1-negative tumors as choroid plexus carcinoma and cribriform neuroepithelial tumor (CRINET), respectively. In conclusion, choroid plexus carcinomas are characterized by complex genetic alterations, which are related to patient age and may have prognostic and diagnostic value. © 2014 Wiley Periodicals, Inc.

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