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Blood and cerebrospinal fluid biomarkers in neuro-oncology

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Abstract

Purpose of review: The purpose of this review is to discuss the value of blood and CSF biomarkers in primary CNS tumors.

Recent findings: Several analytes can be assessed with liquid biopsy techniques, including circulating tumor cells, circulating cell-free tumor DNA, circulating cell-free RNA, circulating proteins and metabolites, extracellular vesicles and tumor-educated platelets. Among diffuse gliomas of the adult, ctDNA in blood or CSF has represented the most used analyte, with the detection of molecular alterations such as MGMT promoter, PTEN, EGFRVIII, TERT promoter mutation and IDH R132H mutation. In general, CSF is enriched for ctDNA as compared with plasma. The use of MRI-guided focused ultrasounds to disrupt the blood-brain barrier could enhance the level of biomarkers in both blood and CSF. The detection of MYD88 L265P mutation with digital droplet PCR and the detection of ctDNA with next generation sequencing represent the best tools to diagnose and monitoring CNS lymphomas under treatment. In meningiomas, the low concentration of ctDNA is a limiting factor for the detection of driver mutations, such as NF2, AKTs, SMO, KLF4, TRAF7, SMARCB1, SMARCE1, PTEN, and TERT; an alternative approach could be the isolation of ctDNA through circulating extracellular vesicles. Liquid biopsies are being used extensively for diagnosis and surveillance of diffuse midline gliomas, in particular with the detection of the driver mutation H3K27M. Last, specific methylome patterns in CSF may allow the distinction of glioblastomas from CNS lymphomas or meningiomas.

Summary: This review summarizes the current knowledge and future perspectives of liquid biopsy of blood and CSF for diagnosis and monitoring of primary CNS tumors.