Molecular In My Pocket…

ONCOLOGY: *Molecular Biomarkers in Tumors of the Central Nervous System*

Molecular Biomarkers in Tumors of the Central Nervous System

Samples to test: Primary or recurrent tumors; formalin fixed paraffin embedded tissue (FFPE).

Biomarker	Specific alterations Alternative terms	Indications	Result interpretation significance	Assays Techniques
IDH1 ***IDH2***	IDH1: Mutations in codon R132 IDH2: Mutations in codons R140, R172	Diagnosis Prognosis	Can be seen in astrocytoma, glioblastoma, oligodendroglioma. Associated with improved prognosis	NGS, pyrosequencing, Sanger sequencing, genotyping, PCR-based assays, IHC
1p/19q co-deletion	Deletion	Diagnosis Prognosis	Co-deletion of 1p/19q is seen in oligodendrogliomas and is associated with improved prognosis.	FISH, array, NGS
BRAF	Mutations in codon V600; fusions	Therapeutic	Seen in tumors such as pilocytic astrocytoma, pleomorphic xanthoastrocytoma, ganglioglioma. Activating mutations or fusions are potentially targetable.	NGS, pyrosequencing, Sanger sequencing, genotyping, PCR-based assays, IHC, rt-PCR, AMP
H3F3A ***HIST1H3B***	Mutations in codon K27	Diagnosis	Seen in diffuse midline glioma, H3 K27M-mutant	NGS, pyrosequencing, Sanger sequencing, genotyping, PCR-based assays, IHC
TERT	Promoter mutation	Prognosis	Associated with less favorable prognosis in IDH-wild type astrocytomas and associated with more favorable prognosis in oligodendroglioma.	NGS, pyrosequencing, Sanger sequencing, genotyping, PCR-based assays
MGMT	Promoter methylation	Therapeutic	Associated with response to temozolomide	Methylation-specific PCR, bisulfite real-time PCR, bisulfite sequencing

Abbreviations:

NGS next generation sequencing
IHC immunohistochemistry
FISH fluorescence in situ hybridization
AMP anchored multiplex PCR

Where to test: Testing should be performed in the laboratories that are certified under clinical laboratory improvement amendments of 1988 (CLIA-88) as qualified to perform high complexity (molecular pathology) testing.

***References*:** Louis DN, Ohgaki H, Wiestler OD, Cavenee WK (Eds). WHO Classification of Tumours of the Central Nervous System. Vol 1. 4th ed. Geneva, Switzerland: World Health Organization; 2016.