

Molecular In My Pocket™ ...

ONCOLOGY: Pediatric Brain Tumors

Prepared by the Association for Molecular Pathology Training and Education Committee

Abbreviations:

CGH	Comparative Genomic Hybridization	CMA	Chromosomal Microarray Analysis	ddPCR	Digital Droplet PCR
FISH	Fluorescent in situ Hybridization	IHC	Immunohistochemistry	MLPA	Multiplex Ligation-dependent Probe Amplification
NGS	Next Generation Sequencing	PCR	Polymerase Chain Reaction		

Tumor Type	Gene/Biomarker	Alteration	Significance	Primary Assays
Pleomorphic xanthoastrocytoma	<i>BRAF</i>	p.V600E mutation	diagnosis	NGS (DNA)
Pilocytic astrocytoma	<i>BRAF</i>	KIAA1549-BRAF fusion or p.V600E	diagnosis, prognosis	ddPCR, NGS (DNA or RNA), array CGH for duplication involving BRAF
Low grade glioma	<i>FGFR</i>	mutation, fusion	diagnosis	NGS (DNA), CMA
	<i>NF1</i>	loss or inactivating mutation, loss of heterozygosity (LOH)	diagnosis, familial cancer risk if germline	NGS (DNA), CMA
	<i>RAF1</i>	fusion	diagnosis	NGS (DNA)
	<i>MYB</i>	fusion, amplification	diagnosis, prognosis	NGS (DNA), CMA
	<i>MYBL1</i>	fusion, amplification	diagnosis, prognosis	NGS (DNA), CMA
	<i>BRAF</i>	p.V600E mutation	diagnosis, prognosis	NGS (DNA)
Diffuse midline glioma, H3 K27M-mutant	<i>H3F3A, HIST1H3B, HIST1H3C</i>	K27M (aka K28M)	diagnosis	NGS (DNA), IHC, ddPCR
Glioma, H3 G34-mutant	<i>H3F3A</i>	G34R (aka G35R)	diagnosis, prognosis	NGS (DNA), IHC, ddPCR
Glioma, NTRK alterations	<i>NTRK3, NTRK1, NTRK2</i>	fusion, mutation, amplification, mRNA overexpression	diagnosis, therapeutic (NTRK inhibitors)	NGS (RNA), NGS (DNA)
High grade glioma, IDH wildtype	<i>TERT</i>	mutation	prognosis	NGS (DNA)
	<i>EGFR</i>	amplification	prognosis	NGS (DNA), CMA, FISH
	<i>CDKN2A/B</i>	homozygous deletion	prognosis	CMA, NGS (DNA)
	<i>TP53</i>	Loss-of-function (LOF) and gain-of-function mutations, loss, LOH	prognosis	NGS (DNA), CMA
	<i>PTEN</i>	mutation	prognosis	NGS (DNA)
	<i>NF1</i>	loss or inactivating mutation, loss of heterozygosity (LOH)	prognosis	NGS (DNA), CMA
	<i>PIK3CA</i>	mutation	prognosis	NGS (DNA)
	<i>PDGFRA</i>	amplification	prognosis	CMA, NGS (DNA)
	<i>RB1</i>	loss or inactivating mutation, loss of heterozygosity (LOH)	prognosis	NGS (DNA), CMA
	<i>PIK3R1</i>	mutation	prognosis	NGS (DNA)
	<i>MDM2</i>	mutation	prognosis	NGS (DNA)
	<i>MDM4</i>	amplification	prognosis	NGS (DNA), CMA, FISH
	<i>MET</i>	amplification, fusion, mutation	prognosis	NGS (DNA), CMA, FISH
	<i>Gain of chromosome 7</i>	chromosomal abnormality (gain of 7)	prognosis	CMA
	<i>10q loss</i>	chromosomal abnormality (loss of 10)	prognosis	CMA
	<i>High tumor mutational burden</i>	mutations	diagnosis, therapeutic (immunotherapy), familial cancer risk if germline alterations in mismatch repair pathway	NGS (DNA)



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Oligodendroglioma	IDH1/2	mutation	diagnosis	NGS (DNA)
	<i>1p/19q codeletion</i>	chromosomal abnormality (loss of 1p and 19q)	diagnosis	CMA, FISH
Ependymoma	<i>c11orf95-RELA, YAP1</i>	Fusions	diagnosis, prognosis	NGS (RNA or DNA)
	<i>1q gain</i>	chromosomal abnormality (gain of 1p)	prognosis	CMA, FISH
Medulloblastoma, WNT-activated	<i>CTNNB1</i>	mutation	Subtype- prognosis	NGS (DNA)
	<i>Monosomy 6</i>	chromosomal abnormality (loss)	Subtype-diagnosis	CMA, FISH
	<i>DDX3X</i>	mutation	diagnosis	NGS (DNA)
Medulloblastoma, SHH-activated	<i>PTCH1</i>	inactivating mutation, LOH	Subtype-diagnosis, familial cancer risk if germline	NGS (DNA), CMA
	<i>SUFU</i>	inactivating mutation, LOH	Subtype-diagnosis, familial cancer risk if germline	NGS (DNA), CMA
	<i>TP53</i>	Loss-of-function (LOF) and gain-of-function mutations, loss, LOH, structural alterations (rare)	prognosis	NGS (DNA), CMA
	<i>10q loss or LOH</i>	chromosomal abnormality (loss or LOH)	diagnosis	CMA, FISH
	<i>TERT promoter</i>	mutation	diagnosis	NGS (DNA)
	<i>MYCN</i>	amplification	diagnosis, prognosis	FISH, CMA, NGS (DNA)
Medulloblastoma, Group 3	<i>Isochromosome 17q</i>	chromosomal abnormality (loss of 17p and gain of 17q)	Subtype-diagnosis	CMA, FISH
	<i>MYC</i>	Amplification, fusion (PVT1)	prognosis	CMA, FISH
Medulloblastoma, Group 4	<i>Isochromosome 17q</i>	chromosomal abnormality (loss of 17p and gain of 17q)	Subtype-diagnosis	CMA, FISH
	<i>MYCN</i>	amplification	diagnosis, prognosis	FISH, CMA, NGS
	<i>KDM6A</i>	inactivating mutation	diagnosis	NGS (DNA)
Spindle Cell Sarcoma with rhabdomyosarcoma-like feature, DICER1 mutant	<i>DICER1</i>	sequence variants, deletions	diagnosis, familial cancer risk if germline	NGS (DNA)
Meningioma	<i>NF2</i>	Loss-of-function (LOF) and gain-of-function mutations, loss, LOH	diagnosis, familial cancer risk if germline	NGS, CMA, MLPA
Choroid plexus tumors	<i>TP53</i>	Loss-of-function (LOF) and gain-of-function mutations, loss, LOH, structural alterations (rare)	diagnosis, familial cancer risk if germline	NGS, CMA, MLPA
ETMR	<i>C19MC</i>	amplification and gain	diagnosis, prognosis	CMA, FISH
SEGA	<i>TSC1/TSC2</i>	loss-of-function mutation, loss, LOH	diagnosis, familial cancer risk if germline	NGS (DNA)
ATRT	<i>SMARCB1 (INI1)</i>	loss or inactivating mutation, loss, LOH	diagnosis, familial cancer risk if germline	NGS, MLPA, IHC



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