CORE DIAGNOSTICS[™]

(GlioSeqv2) that analyzes for an additional 110 types of gene fusions including RELA, YAP1, NTRK1-3, BRAF, FGFR1, MYB, MYBL1, and others. The analytical sensitivity is 3%–5% allele frequency for detection of SNVs and short indels (<40 bp) and 1%–5% for detection of gene fusions.

The minimum required sequencing depth is 300x. This targeted NGS-panel can be performed on FFPE blocks with a fast turn around time of 7-10days and requires only a small amount of DNA/ RNA. The various genes assayed in this panel are as follows:

To conclude, molecular testing in CNS tumors is the need of the hour. With the new WHO classification of CNS tumors around the corner, these tests will be essential for the diagnosis of new molecularly defined entities as well as act as predicitive and prognostic markers for many of the existing tumor entities.

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Mutations (SNVs, indels)	Copy Number Variations	Gene Fusions			
AKT1	ATRX	AGBL4	FGFR2	NBDY	TACC1
ATRX	CDK6	AKAP9	FGFR3	NFASC	TACC3
BRAF	CDKN2A	ALK	FOXR2	NTRK1	TCF4
CDK6	CIC (19q)	ATP2B4	FYCO1	NTRK2	TFG
CDKN2A	CTNNB1	BCAN	GLI2	NTRK3	THAP9
CIC	DDX3X	BCL6B	GNAI1	NUTM1	TMEM40
CTNNB1	EGFR	BEND2	GOPC	NYAP2	TP53
DDX3X	FUBP1 (1p)	BEND5	GTF2I	PAPSS1	TPM3
EGFR	HRAS	BRAF	HTRA1	PCDHGC3	TRIM24
FUBP1	IDH2	BTBD1	JMJD1C	PDGFRA	TTYH1
H3F3A	KRAS	C11orf95	JPX	PIK3CD	VCL
HRAS	MET	C19MC	KDR	PPP1CB	YAP1
IDH1	MYC	CAND1	KIAA1549	PRKCA	ZC3H3
IDH2	MYCN	CCDC88A	KIAA1598	PSPH	
KLF4	NF1	CEP85L	KTN1	PTEN	
KRAS	NF2	CEP89	LINC00964	PTPRZ1	
MET	NRAS	CIC	LOC101927141	PVT1	
MYC	PIK3CA	CLCN6	LSM14A	QKI	
MYCN	PTCH1	CLIP2	MACF1	RAF1	
NF1	PTEN	CTNNA3	MAMLD1	RELA	
NF2	RB1	CXXC5	MARCKSL1	RNF130	
NRAS	SETD2	CYP17A1-AS1	MET	ROCK1	
PIK3CA	SMO	DDX3X	MKRN1	ROS1	
PTCH1	TP53	EGFR, EGFRvIII	MMP16	SEPT14	
PTEN		EPB41L5	MN1	SH3PXD2A	
RB1		ESR1	MYB	SLC44A1	
SETD2		ETV6	MYBL1	SLMAP	
SMO		FAM118B	MYC	SRGAP3	
TERT		FAM131B	NACC2	SUFU	
TP53		FGFR1	NAV1	SULT1C2P1	