


# Tumours of the Central Nervous System Molecular Information Reporting Guide

Family/Last name  Date of birth

Given name(s)

Patient identifiers  Date of request  Accession/Laboratory number

Elements in **black text** are CORE. Elements in **grey text** are NON-CORE. 


indicates multi-select values  indicates single select values

SCOPE OF THIS DATASET SECTION 

## REFER TO Tables 1-3 FOR CORE ELEMENTS REQUIRED FOR CENTRAL NERVOUS SYSTEM TUMOUR CLASSIFICATION (Elements 2-38 are only required for some tumours)

(Based on the World Health Organization Classification of Tumours of the Central Nervous System (2021))

### ADEQUACY OF SPECIMEN FOR MOLECULAR ASSESSMENT

- Specimen is adequate for analysis 
- Specimen is inadequate for analysis (select all that apply)

- Crush
- Autolysis
- Cautery
- Necrosis
- Decalcification
- Tumour cell quantity
- Fixation issues, *specify*

Other, *specify*

**Representative blocks for ancillary studies, specify those blocks best representing tumour and/or normal tissue for further study**

### ALK/ROS1/MET/NTRK FAMILY ALTERATIONS<sup>a</sup>

- Indeterminate
- Absent
- Present, *describe*

TESTING METHOD<sup>b</sup> (select all that apply)

- Immunohistochemistry (IHC)
- In situ hybridisation (ISH)
- Next generation sequencing (NGS)
- Other, *specify*

<sup>a</sup> Only core for some tumours - refer to [Tables 1-3](#).

<sup>b</sup> Repeat for each alteration.

### ATRX ALTERATIONS<sup>a</sup>

- Indeterminate
- Absent
- Present, *describe*

TESTING METHOD (select all that apply)

- Sanger sequencing
- NGS
- PCR-based method
- IHC

- Indeterminate
- Intact nuclear expression
- Loss of nuclear expression

Other, *specify*

### BCOR INTERNAL TANDEM DUPLICATION<sup>a</sup>

- Indeterminate
- Absent
- Present, *describe*

TESTING METHOD (select all that apply)

- IHC
- ISH
- NGS
- Other, *specify*

### BRAF ALTERATIONS<sup>a</sup>

#### BRAF variant

- Indeterminate
- Absent
- BRAF p.V600E (c.1799T>A) variant present
- Other BRAF sequence variant present, *specify*

**BRAF ALTERATIONS<sup>a</sup>** 

VARIANTS ASSESSED (select all that apply)

- p.V600E
- Any variant in exon 15
- Other *BRAF* variant, *specify*

▼

**BRAF variant continued**

TESTING METHOD (select all that apply)

- Sanger sequencing
- NGS
- PCR-based method
- IHC

▼ **BRAF p.V600E expression**

- Indeterminate
- Negative
- Positive

Other, *specify*

▼

**BRAF rearrangement/duplication**

- Indeterminate
- Absent
- Present, *describe*

▼

VARIANTS ASSESSED (select all that apply)

- KIAA1549::BRAF* fusion
- BRAF::RAF1* fusion
- Other, *specify*

▼

TESTING METHOD (select all that apply)

- ISH
- RT-PCR
- Array-based method
- RNA-sequencing
- Other, *specify*

▼

**C19MC ALTERATIONS<sup>a</sup>** 

- Indeterminate
- Absent
- Present with low level gain
- Present, *describe including copy number*

▼

TESTING METHOD (select all that apply)

- ISH
- Array-based method
- NGS
- Other, *specify*

▼

**LIN28A expression (IHC)<sup>a</sup>**

- Indeterminate
- Negative
- Positive

<sup>a</sup> Only core for some tumours - refer to [Tables 1-3](#).

**CDKN2A/B DELETION<sup>a</sup>** 

- Indeterminate
- Absent
- Homozygous deletion
- Hemizygous/heterozygous deletion

TESTING METHOD (select all that apply)

- ISH
- Array-based method
- NGS
- Other, *specify*

▼

**CHROMOSOMAL ARM 1p/19q CODELETION<sup>a</sup>** 

- Indeterminate
- None detected
- 1p/19q codeletion
- 1p only deletion
- 19q only deletion
- Polysomy, *specify*

▼

TESTING METHOD (select all that apply)

- ISH
- Array-based method (including methylation arrays<sup>c</sup>)
- PCR/Loss of heterozygosity assay
- NGS
- Other, *specify*

▼

**CHROMOSOME 7 GAIN (COMBINED WITH CHROMOSOME 10 LOSS)<sup>a</sup>** 

- Indeterminate
- Absent
- Present, *describe*

▼

TESTING METHOD (select all that apply)

- ISH
- Array-based method
- NGS
- Other, *specify*

▼

**C1C ALTERATIONS<sup>a</sup>** 

- Indeterminate
- Absent
- Present, *describe*

▼

TESTING METHOD (select all that apply)

- IHC
- ISH
- NGS
- Other, *specify*

▼

<sup>c</sup> Methylation array-based methods may provide strong but circumstantial evidence.

**DICER1 ALTERATIONS<sup>a</sup>** 

- Indeterminate
- Absent
- Present, *describe*

▼

TESTING METHOD (select all that apply)

- IHC
- ISH
- NGS
- Other, *specify*

▼

**EGFR ALTERATIONS<sup>a</sup>** 

**EGFR amplification**

- Indeterminate
- Absent
- Absent with low level gain
- Present, *describe including copy number*

▼

TESTING METHOD (select all that apply)

- ISH
- Array-based method
- NGS
- Other, *specify*

▼

**EGFR variants (e.g., EGFRvIII or EGFR single nucleotide variants)**

- Indeterminate
- Absent
- Present, *describe*

▼

TESTING METHOD (select all that apply)

- NGS
- PCR-based method
- IHC
- Other, *specify*

▼

**EZH1P EXPRESSION (IHC)<sup>a</sup>** 

- Indeterminate
- Negative
- Positive

**FET ALTERATIONS<sup>a</sup>** 

- Indeterminate
- Absent
- Present, *describe*

▼

TESTING METHOD (select all that apply)

- IHC
- ISH
- NGS
- Other, *specify*

▼

<sup>a</sup> Only core for some tumours - refer to [Tables 1-3](#).

**FGFR FAMILY ALTERATIONS<sup>a</sup>** 

- Indeterminate
- Absent
- Present, *describe*

▼

TESTING METHOD<sup>b</sup> (select all that apply)

- IHC
- ISH
- NGS
- Other, *specify*

▼

**FOXR2 ALTERATIONS<sup>a</sup>** 

- Indeterminate
- Absent
- Present, *describe*

▼

TESTING METHOD (select all that apply)

- IHC
- ISH
- NGS
- Other, *specify*

▼

**HISTONE H3 VARIANTS AND LOSS OF H3 p.K27me3 (K27me3)<sup>a</sup>** 

**Histone H3 gene family variants**

- Indeterminate
- Negative
- Positive for K27M
- Positive for G34R or G34V
- Positive, for other H3 variants, *specify*

▼

TESTING METHOD (select all that apply)

- Sanger sequencing
- NGS
- PCR-based method
- IHC

**Histone H3 K27M expression**

- Indeterminate
- Negative
- Positive

**Histone H3 G34R expression**

- Indeterminate
- Negative
- Positive

**Histone H3 K27me3 expression**

- Indeterminate
- Intact expression
- Loss of expression

- Other, *specify*

▼

<sup>b</sup> Repeat for each alteration.

**IDH1/IDH2 ALTERATIONS<sup>a</sup>** 

- Indeterminate
- Absent
- Present, *describe*

TESTING METHOD (select all that apply)

- Sanger sequencing
- NGS
- PCR-based method
- IHC

**IDH1 R132H expression**


- Indeterminate
- Negative
- Positive

Other, *specify*

**MAPK PATHWAY ALTERATIONS** 

- Indeterminate
- Absent
- NF1 loss, *describe*

For *BRAF-KIAA1549* alterations refer to **BRAF ALTERATIONS** 

For *FGRFR* alterations refer to **FGFR FAMILY ALTERATIONS** 

Positive for other MAPK alteration, *describe*

TESTING METHOD<sup>b</sup> (select all that apply)

- IHC
- ISH
- NGS
- Other, *specify*

**METHYLOME PROFILING<sup>a</sup>** 

Classifier (e.g., Heidelberg Brain Tumour Classifier)	Version (e.g., 12.5)	Methylation class	Score

**MGMT promoter status**

- Indeterminate
- Unmethylated
- Methylated

**Most informative copy number variations, specify**

<sup>a</sup> Only core for some tumours - refer to [Tables 1-3](#).

<sup>b</sup> Repeat for each alteration.

**MN1 ALTERATIONS<sup>a</sup>** 

- Indeterminate
- Absent
- Present, *describe*

TESTING METHOD (select all that apply)

- IHC
- ISH
- NGS
- Other, *specify*

**MYB, MYBL1 ALTERATIONS<sup>a</sup>** 

- Indeterminate
- Absent
- Present, *describe*

TESTING METHOD<sup>b</sup> (select all that apply)

- IHC
- ISH
- NGS
- Other, *specify*

**MYC GENE FAMILY AMPLIFICATION (MYC and/or MYCN)<sup>a</sup>** 

- Indeterminate
- Absent
- Absent with low level gain
- Present, *describe including copy number*

TESTING METHOD<sup>b</sup> (select all that apply)

- ISH
- Array-based method
- NGS
- Other, *specify*

**PDGFRA ALTERATIONS<sup>a</sup>** 

- Indeterminate
- Absent
- Present, *describe including copy number*

TESTING METHOD (select all that apply)

- IHC
- ISH
- NGS
- Other, *specify*

**PITUITARY HORMONES AND TRANSCRIPTION FACTORS IMMUNOHISTOCHEMISTRY<sup>a</sup>** 

**Tumour cells are reactive for** (select all that apply)

- Indeterminate
- Prolactin
- Human growth hormone
- β-TSH
- β-FSH
- β-LH
- Alpha subunit
- ACTH
- PIT1
- TPIT
- SF1
- Other, *specify*

**PRC2 ALTERATION<sup>a</sup>** 

- Indeterminate
- Absent
- Present, *describe*

TESTING METHOD (select all that apply)

- IHC
- ISH
- NGS
- Other, *specify*

**PRKAR1A ALTERATION<sup>a</sup>** 

- Indeterminate
- Absent
- Present, *describe*

TESTING METHOD (select all that apply)

- IHC
- ISH
- NGS
- Other, *specify*

**PRKCA ALTERATION<sup>a</sup>** 

- Indeterminate
- Absent
- Present, *describe*

TESTING METHOD (select all that apply)

- IHC
- ISH
- NGS
- Other, *specify*

<sup>a</sup> Only core for some tumours - refer to [Tables 1-3](#).

**SHH PATHWAY ALTERATIONS<sup>a</sup>** 

- Indeterminate
- Absent
- Present, *describe*

TESTING METHOD<sup>b</sup> (select all that apply)

- IHC
- ISH
- NGS
- Other, *specify*

**SMARC FAMILY ALTERATIONS<sup>a</sup>** 

**SMARCA4/BRG1 alteration**

- Indeterminate
- Absent
- Present, *describe sequence variant(s)*

TESTING METHOD (select all that apply)

- Sanger sequencing
- NGS
- PCR-based method
- Other, *specify*

**BRG1 loss of expression (IHC)**

- Indeterminate
- Intact nuclear expression
- Loss of nuclear expression

**SMARCB1/INI1/SNF5 alteration**

- Indeterminate
- Absent
- Present, *describe sequence variant(s)*

TESTING METHOD (select all that apply)

- Sanger sequencing
- NGS
- PCR-based method
- Other, *specify*

**INI1 (BAF47) loss of expression (IHC)**

- Indeterminate
- Intact nuclear expression
- Loss of nuclear expression

<sup>b</sup> Repeat for each alteration.

**STAT6 ALTERATIONS<sup>a</sup>** 

**STAT6 expression (IHC)**

- Indeterminate
- Absence of nuclear expression
- Positive nuclear expression

**STAT6 rearrangement**

- Indeterminate
- Absent
- Present, *describe*

TESTING METHOD (select all that apply)

- ISH
- NGS
- Other, *specify*

**TERT PROMOTER ALTERATIONS<sup>a</sup>** 

- Indeterminate
- Absent
- Hotspot variant (C228T or C250T)
- Other sequence variant, *specify*

TESTING METHOD (select all that apply)

- Sanger sequencing
- NGS
- PCR-based method
- Other, *specify*

**TP53 ALTERATIONS<sup>a</sup>** 

**TP53 variant**

- Indeterminate
- Absent
- Present, *describe*

EXONS ANALYSED

- Exons 5-8
- All exons
- Other, *specify*

TESTING METHOD (select all that apply)

- Sanger sequencing
- NGS
- PCR-based method
- IHC

**p53 expression**

- Negative or rare, lightly positive cells
- Intermediate (intermediate numbers of predominantly lightly positive cells)
- Positive (diffuse and strong nuclear positivity)

- Other, *specify*

<sup>a</sup> Only core for some tumours - refer to [Tables 1-3](#).

**TTF1 EXPRESSION (IHC)<sup>a</sup>** 

- Indeterminate
- Negative
- Positive

**WNT PATHWAY ALTERATIONS<sup>a</sup>** 

- Indeterminate
- Absent
- Present, *describe*

TESTING METHOD<sup>b</sup> (select all that apply)

- IHC
- ISH
- NGS
- Other, *specify*

**YAP1 REARRANGEMENT<sup>a</sup>** 

- Indeterminate
- Absent
- Present, *describe*

TESTING METHOD (select all that apply)

- ISH
- NGS
- Other, *specify*

**ZFTA REARRANGEMENT<sup>a</sup>** 

- Indeterminate
- Absent
- Present, *describe*

TESTING METHOD (select all that apply)

- IHC
- ISH
- NGS
- Other, *specify*

**L1CAM expression (IHC)**

- Indeterminate
- Negative
- Positive

**RELA rearrangement**

- Indeterminate
- Absent
- Present, *describe*

TESTING METHOD (select all that apply)

- ISH
- NGS
- Other, *specify*

<sup>b</sup> Repeat for each alteration.

**OTHER IMMUNOHISTOCHEMISTRY FINDINGS<sup>a</sup>** 

- None identified
- Present, *record test(s), methodology and results*


<sup>a</sup> Only core for some tumours - refer to [Tables 1-3](#).

**OTHER MOLECULAR FINDINGS** 

- None identified
- Present, *record test(s), methodology and results*
