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Tumours of the Central Nervous System Molecular Information Reporting Guide

Family/Last name

Given name(s)

Date of birth DD – MM – YYYY

Patient identifiers

Date of request DD – MM – YYYY

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 from *ALK/ROS1/MET*/NTRK FAMILY ALTERATIONS to OTHER IMMUNOHISTOCHEMISTRY FINDINGS 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[a]

◯ Indeterminate

◯ Absent

◯ Present, *describe*

TESTING METHOD[b] (select all that apply)

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

ATRX ALTERATIONS[a]

◯ 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[a]

◯ Indeterminate

◯ Absent

◯ Present, *describe*

TESTING METHOD (select all that apply)

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

BRAF ALTERATIONS[a]

***BRAF* variant**

◯ Indeterminate

◯ Absent

◯ BRAF p.V600E (c.1799T>A) variant present

◯ Other BRAF sequence variant present, *specify*

[a] *Only core for some tumours - refer to Tables 1-3.*

[b] *Repeat for each alteration.*

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BRAF ALTERATIONS[a] continued

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[a]

- ◯ 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)[a]

- ◯ Indeterminate
- ◯ Negative
- ◯ Positive

[a] *Only core for some tumours - refer to Tables 1-3.*

CDKN2A/B DELETION[a]

- ◯ 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[a]

- ◯ 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[c])
- [ ] PCR/Loss of heterozygosity assay
- [ ] NGS
- [ ] Other, *specify*

CHROMOSOME 7 GAIN (COMBINED WITH CHROMOSOME 10 LOSS)[a]

- ◯ Indeterminate
- ◯ Absent
- ◯ Present, *describe*

TESTING METHOD (select all that apply)

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

CIC ALTERATIONS[a]

- ◯ Indeterminate
- ◯ Absent
- ◯ Present, *describe*

TESTING METHOD (select all that apply)

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

[c] *Methylation array-based methods may provide strong but circumstantial evidence.*

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DICER1 ALTERATIONS[a]

- ◯ Indeterminate
- ◯ Absent
- ◯ Present, *describe*

TESTING METHOD (select all that apply)

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

EGFR ALTERATIONS[a]

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*

EZHIP EXPRESSION (IHC)[a]

- ◯ Indeterminate
- ◯ Negative
- ◯ Positive

FET ALTERATIONS[a]

- ◯ Indeterminate
- ◯ Absent
- ◯ Present, *describe*

TESTING METHOD (select all that apply)

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

FGFR FAMILY ALTERATIONS[a]

- ◯ Indeterminate
- ◯ Absent
- ◯ Present, *describe*

TESTING METHOD[b] (select all that apply)

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

FOXR2 ALTERATIONS[a]

- ◯ Indeterminate
- ◯ Absent
- ◯ Present, *describe*

TESTING METHOD (select all that apply)

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

HISTONE H3 VARIANTS AND LOSS OF H3 p.K28me3 (K27me3)[a]

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*

[a] *Only core for some tumours - refer to Tables 1-3.*

[b] *Repeat for each alteration.*

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IDH1/IDH2 ALTERATIONS[a]

- ◯ 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[a]

- ◯ Indeterminate
- ◯ Absent
- ◯ NF1 loss, *describe*

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

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

- ◯ Positive for other MAPK alteration, *describe*

TESTING METHOD[b] (select all that apply)

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

METHYLOME PROFILING[a]

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*

MN1 ALTERATIONS[a]

- ◯ Indeterminate
- ◯ Absent
- ◯ Present, *describe*

TESTING METHOD (select all that apply)

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

MYB, MYBL1 ALTERATIONS[a]

- ◯ Indeterminate
- ◯ Absent
- ◯ Present, *describe*

TESTING METHOD[b] (select all that apply)

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

MYC GENE FAMILY AMPLIFICATION (*MYC* and/or *MYCN*)[a]

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

TESTING METHOD[b] (select all that apply)

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

PDGFRA ALTERATIONS[a]

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

TESTING METHOD (select all that apply)

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

[a] *Only core for some tumours - refer to Tables 1-3.*

[b] *Repeat for each alteration.*

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PITUITARY HORMONES AND TRANSCRIPTION FACTORS IMMUNOHISTOCHEMISTRY[a]

Tumour cells are reactive for (select all that apply)

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

PRC2 ALTERATION[a]

- ◯ Indeterminate
- ◯ Absent
- ◯ Present, *describe*

TESTING METHOD (select all that apply)

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

***PRKAR1A* ALTERATION**[a]

- ◯ Indeterminate
- ◯ Absent
- ◯ Present, *describe*

TESTING METHOD (select all that apply)

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

***PRKCA* ALTERATION**[a]

- ◯ Indeterminate
- ◯ Absent
- ◯ Present, *describe*

TESTING METHOD (select all that apply)

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

[a] *Only core for some tumours - refer to Tables 1-3.*

SHH PATHWAY ALTERATIONS[a]

- ◯ Indeterminate
- ◯ Absent
- ◯ Present, *describe*

TESTING METHOD[b] (select all that apply)

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

SMARC FAMILY ALTERATIONS[a]

***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

[b] *Repeat for each alteration.*

STAT6 ALTERATIONS[a]

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[a]

- ◯ 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[a]

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*

[a] *Only core for some tumours - refer to Tables 1-3.*

TTF1 EXPRESSION (IHC)[a]

- ◯ Indeterminate
- ◯ Negative
- ◯ Positive

WNT PATHWAY ALTERATIONS[a]

- ◯ Indeterminate
- ◯ Absent
- ◯ Present, *describe*

TESTING METHOD[b] (select all that apply)

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

YAP1 REARRANGEMENT[a]

- ◯ Indeterminate
- ◯ Absent
- ◯ Present, *describe*

TESTING METHOD (select all that apply)

- ☐ ISH
- ☐ NGS
- ☐ Other, *specify*

ZFTA REARRANGEMENT[a]

- ◯ 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*

[b] *Repeat for each alteration.*

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OTHER IMMUNOHISTOCHEMISTRY FINDINGS[a]

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

[a] *Only core for some tumours - refer to Tables 1-3.*

OTHER MOLECULAR FINDINGS

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

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