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MASSACHUSETTS GENERAL HOSPITAL PATHOLOGY

Tumours of the Central Nervous System (CNS) 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

All molecular elements are NON-CORE. DATASET SCOPE OVERVIEW OF SELECTED MOLECULAR MARKERS

ADEQUACY OF SPECIMEN FOR MOLECULAR ASSESSMENT

- ◯ Specimen is adequate for analysis
- ◯ Specimen is inadequate for analysis, *give reason, (select all that apply)*
  - ☐ Crush
  - ☐ Autolysis
  - ☐ Cautery
  - ☐ Necrosis
  - ☐ Decalcification
  - ☐ Tumour cell quantity
  - ☐ Fixation issues, *specify*
  - ☐ Other, *specify*

ATRX MUTATION

ATRX mutation

- ◯ Negative
- ◯ Positive
- ◯ Cannot be determined

TESTING METHOD (select all that apply)

- ☐ Sanger sequencing
- ☐ Next-generation sequencing
- ☐ PCR-based method
- ☐ Other, *specify*

ATRX expression (immunohistochemistry)

- ◯ Intact nuclear expression
- ◯ Loss of nuclear expression
- ◯ Cannot be determined

BRAF ALTERATIONS

BRAF mutation

- ◯ Absent
- ◯ BRAF V600E (c.1799T>A) mutation present
- ◯ Other BRAF mutation present, *specify*
- ◯ Cannot be determined

MUTATIONS ASSESSED (select all that apply)

- ☐ V600E
- ☐ Any mutation in exon 15
- ☐ Other, *specify*

TESTING METHOD (select all that apply)

- ☐ Sanger sequencing
- ☐ Next-generation sequencing
- ☐ PCR-based method
- ☐ Other, *specify*

BRAF V600E expression (immunohistochemistry)

- ◯ Negative
- ◯ Positive
- ◯ Cannot be determined

BRAF rearrangement/duplication

- ◯ Absent
- ◯ Present, *specify*
- ◯ Cannot be determined

MUTATIONS ASSESSED (select all that apply)

- ☐ 7q34 tandem duplication
- ☐ KIAA-BRAF fusion
- ☐ BRAF-RAF1 fusion
- ☐ Other, *specify*

TESTING METHOD (select all that apply)

- ☐ In situ hybridization (FISH)
- ☐ RT-PCR
- ☐ Array-based method
- ☐ RNA-sequencing
- ☐ Other, *specify*

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CDKN2A/B HOMOZYGOUS DELETION

- ◯ Absent
- ◯ Homozygous deletion
- ◯ Heterozygous deletion
- ◯ Cannot be determined

TESTING METHOD (select all that apply)

- ☐ In situ hybridization (FISH, CISH)
- ☐ Array-based method
- ☐ Next-generation sequencing
- ☐ Other, *specify*

C19MC ALTERATION

- ◯ Absent
- ◯ Absent with low level gain
- ◯ Present, *specify, including copy number*
- ◯ Cannot be determined

TESTING METHOD (select all that apply)

- ☐ In situ hybridization (FISH, CISH)
- ☐ Array-based method
- ☐ Next-generation sequencing
- ☐ Other, *specify*

CHROMOSOMAL ARM 1p/19q CODELETION

- ◯ None detected
- ◯ 1p/19q codeletion
- ◯ 1p only deletion
- ◯ 19q only deletion
- ◯ Polysomy, *specify*
- ◯ Cannot be determined

TESTING METHOD (select all that apply)

- ☐ In situ hybridization (FISH, CISH)
- ☐ Array-based method
- ☐ PCR/Loss of heterozygosity assay
- ☐ Next-generation sequencing
- ☐ Other, *specify*

CHROMOSOME 7 GAIN (combined with chromosome 10 loss)

- ◯ Absent
- ◯ Present
- ◯ Cannot be determined

TESTING METHOD (select all that apply)

- ☐ In situ hybridization
- ☐ Array-based method
- ☐ Next-generation sequencing
- ☐ Other, *specify*

CHROMOSOME 10q23 (*PTEN* LOCUS) DELETION AND *PTEN* MUTATION

Chromosome 10q23 (*PTEN* Locus) deletion

- ◯ None detected
- ◯ Interstitial deletion present
- ◯ Monosomy, *specify*
- ◯ Polysomy, *specify*
- ◯ Cannot be determined

TESTING METHOD (select all that apply)

- ☐ In situ hybridization
- ☐ Array-based method
- ☐ PCR/Loss of heterozygosity assay
- ☐ Other, *specify*

PTEN mutation

- ◯ Absent
- ◯ Present, *specify*
- ◯ Cannot be determined

TESTING METHOD (select all that apply)

- ☐ Sanger sequencing
- ☐ Next-generation sequencing
- ☐ PCR-based method
- ☐ Other, *specify*

EGFR AMPLIFICATION AND EGFRvIII MUTATION

EGFR amplification

- ◯ Absent
- ◯ Absent with low level gain
- ◯ Present, *specify, including copy number*
- ◯ Cannot be determined

TESTING METHOD (select all that apply)

- ☐ In situ hybridization (FISH, CISH)
- ☐ Array-based method
- ☐ Next-generation sequencing
- ☐ Other, *specify*

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EGFRvIII mutation

- ◯ Absent
- ◯ Present
- ◯ Cannot be determined

TESTING METHOD (select all that apply)

- ☐ Next-generation sequencing
- ☐ PCR-based method
- ☐ Immunohistochemistry
- ☐ Other, *specify*

HISTONE H3 MUTATION AND H3 K27 TRIMETHYLATION (me3)

Histone H3 gene family mutation

- ◯ Negative
- ◯ Positive for K27M
- ◯ Positive for G34R or G34V
- ◯ Positive, for other H3 mutation, *specify*
- ◯ Cannot be determined

TESTING METHOD (select all that apply)

- ☐ Sanger sequencing
- ☐ Next-generation sequencing
- ☐ PCR-based method
- ☐ Other, *specify*

Histone H3 K27M expression (immunohistochemistry)

- ◯ Negative
- ◯ Positive
- ◯ Cannot be determined

Histone H3 G34R expression (immunohistochemistry)

- ◯ Negative
- ◯ Positive
- ◯ Cannot be determined

Histone H3 K27me3 expression (immunohistochemistry)

- ◯ Intact expression
- ◯ Loss of expression
- ◯ Cannot be determined

IDH1/IDH2 MUTATION

***IDH1/IDH2* mutation**

- ◯ Absent
- ◯ Present, *specify*
- ◯ Cannot be determined

TESTING METHOD (select all that apply)

- ☐ Sanger sequencing
- ☐ Next-generation sequencing
- ☐ PCR-based method
- ☐ Other, *specify*

IDH1 R132H expression (immunohistochemistry)

- ◯ Negative
- ◯ Positive
- ◯ Cannot be determined

Ki-67 IMMUNOHISTOCHEMISTRY

Percentage of positive nuclei ____ %

- ◯ Cannot be determined

L1CAM EXPRESSION (IMMUNOHISTOCHEMISTRY)

- ◯ Negative
- ◯ Positive
- ◯ Cannot be determined

LIN28A EXPRESSION (IMMUNOHISTOCHEMISTRY)

- ◯ Negative
- ◯ Positive
- ◯ Cannot be determined

MEDULLOBLASTOMA IMMUNOHISTOCHEMISTRY

ß-catenin expression (immunohistochemistry)

- ◯ Absence of nuclear expression
- ◯ Positive nuclear expression
- ◯ Cannot be determined

GAB1 expression (immunohistochemistry)

- ◯ Negative
- ◯ Positive
- ◯ Cannot be determined

YAP1 expression (immunohistochemistry)

- ◯ Negative
- ◯ Positive
- ◯ Cannot be determined

MGMT PROMOTER METHYLATION

- ◯ Absent
- ◯ Present
- ◯ Cannot be determined

TESTING METHOD (select all that apply)

- ☐ Methylation-specific PCR
- ☐ Other, *specify*

MONOSOMY 6

- ◯ Absent
- ◯ Present, *specify*
- ◯ Cannot be determined

TESTING METHOD (select all that apply)

- ☐ In situ hybridization
- ☐ Multiplex ligation-dependend probe amplification (MLPA)
- ☐ Array-based method
- ☐ Microsatellite analysis

MYC GENE FAMILY AMPLIFICATION (*MYC* and/or *MYCN*)

- ◯ Absent
- ◯ Absent with low level gain
- ◯ Present, *specify, including copy number*
- ◯ Cannot be determined

TESTING METHOD (select all that apply)

- ☐ In situ hybridization (FISH, CISH)
- ☐ Array-based method
- ☐ Next-generation sequencing
- ☐ Other, *specify*

NAB2-STAT6 FUSION

***NAB2-STAT6* fusion**

- ◯ Negative
- ◯ Positive
- ◯ Cannot be determined

TESTING METHOD (select all that apply)

- ☐ FISH
- ☐ Next generation sequencing
- ☐ Other, *specify*

STAT6 expression (immunohistochemistry)

- ◯ Absence of nuclear expression
- ◯ Positive nuclear expression
- ◯ Cannot be determined

PITUITARY HORMONES AND TRANSCRIPTION FACTORS IMMUNOHISTOCHEMISTRY

Tumour cells are reactive for (select all that apply)

- ☐ Prolactin
- ☐ Human growth hormone
- ☐ ß-TSH
- ☐ ß-FSH
- ☐ ß-LH
- ☐ Alpha subunit
- ☐ ACTH
- ☐ PIT1
- ☐ TPIT
- ☐ SF1
- ☐ Other, *specify*
- ◯ Cannot be determined

RELA FUSION

- ◯ Negative
- ◯ Positive
- ◯ Cannot be determined

TESTING METHOD (select all that apply)

- ☐ FISH
- ☐ Next generation sequencing
- ☐ Other, *specify*

SMARCA4/BRG1 ALTERATION

***SMARCA4/BRG1* mutation**

- ◯ Absent
- ◯ Present, *specify*
- ◯ Cannot be determined

TESTING METHOD (select all that apply)

- ☐ Sanger sequencing
- ☐ Next-generation sequencing
- ☐ PCR-based method
- ☐ Other, *specify*

BRG1 loss of expression (immunohistochemistry)

- ◯ Intact nuclear expression
- ◯ Loss of nuclear expression
- ◯ Cannot be determined

SMARCB1/INI1/HSNF5 ALTERATION

***SMARCB1/INI1/HSNF5* mutation**

- ◯ Absent
- ◯ Present, *specify*
- ◯ Cannot be determined

TESTING METHOD (select all that apply)

- ☐ Sanger sequencing
- ☐ Next-generation sequencing
- ☐ PCR-based method
- ☐ Other, *specify*

INI1 (BAF47) loss of expression (immunohistochemistry)

- ◯ Intact nuclear expression
- ◯ Loss of nuclear expression
- ◯ Cannot be determined

TERT PROMOTER MUTATION

- ◯ Absent
- ◯ Hotspot mutation (C228T or C250T)
- ◯ Other mutation, *specify*
- ◯ Cannot be determined

TESTING METHOD (select all that apply)

- ☐ Sanger sequencing
- ☐ Next-generation sequencing
- ☐ PCR-based method
- ☐ Other, *specify*

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TP53 MUTATION

_TP53_ mutation

- ◯ Absent
- ◯ Cannot be determined
- ◯ Present, *specify*

EXONS ANALYSED

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

TESTING METHOD (select all that apply)

- ☐ Sanger sequencing
- ☐ Next-generation sequencing
- ☐ PCR-based method
- ☐ Other, *specify*

p53 expression (immunohistochemistry)

- ◯ Negative or rare, lightly positive cells
- ◯ Intermediate (intermediate numbers of predominantly lightly positive cells)
- ◯ Positive (diffuse and strong nuclear positivity)
- ◯ Cannot be determined

YAP1 FUSION

- ◯ Negative
- ◯ Positive
- ◯ Cannot be determined

TESTING METHOD (select all that apply)

- ☐ FISH
- ☐ Next generation sequencing
- ☐ Other, *specify*

OTHER FINDINGS

Other immunohistochemical findings, *specify*

Other molecular findings, *specify test, testing method and findings*

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