1p/19q Deletion in Oligodendrogliomas

Malignant gliomas (oligodendrogliomas) are the most common type of primary brain tumors. Identification of the 1p/19q deletion is useful in differentiating oligodendrogliomas from astrocytomas and assists in the diagnosis and prognosis of both low- and high-grade oligodendrogliomas, as well as in predicting response to therapy. The 1p/19q deletion should not be used alone for diagnosis.

Disease Overview

Prevalence

- Second most common glioma in adults
- Accounts for 2% of central nervous system (CNS) tumors

Diagnostic Issues

Malignant gliomas are the most common type of primary brain tumors (>70% of all CNS tumors).

- Subtypes are astrocytoma and oligodendroglioma
  - Differentiating astrocytoma from oligodendroglioma is crucial
- Treatment and prognosis differ between tumors
- Combined loss of chromosomal arms 1p and 19q is diagnostic for oligodendrogliomas
- Gain of chromosome 19 supports diagnosis of high-grade astrocytoma (glioblastoma)
- Loss of 1p may identify treatment-sensitive high-grade oligodendroglioma (for both chemotherapy and radiotherapy)
  - Prognostic relevance in low-grade tumors is less well characterized

Genetics

Gene

Chromosomes 1 and 19 involved

Structure/Function

Deletion of short arm of 1p and long arm of 19q results in loss of mediators of resistance to therapy

Variants

- 1p/19q codeletion is mutually exclusive for *TP53* and *ATRX* mutations and *EGFR* amplification
- 1p/19q codeletion is frequently associated with *IDH1* or *IDH2* variants

Test Interpretation

Results

- Positive
  - Tumors with 1p/1q ratio <0.80 and ≥24% deleted cells are deemed deleted for 1p
  - Tumors with 19q/19p ratio <0.80 and ≥26% deleted cells are deemed deleted for 19q
  - Both deletions are associated with a better prognosis
    - Codeletion has better prognosis than single deletion
  - Presence of codeletion establishes diagnosis of oligodendroglioma

Featured ARUP Testing

1p19q Deletion by FISH and IDH1 R132H Point Mutation by Immunohistochemistry with Reflex to IDH1 and IDH2 Mutation Analysis, Exon 4 3002135

Method: Fluorescence in situ Hybridization (FISH)/Immunohistochemistry/Polymerase Chain Reaction/Sequencing

- Preferred initial test for the diagnosis of oligodendrogliomas
- Detect IDH1 or IDH2 mutation and 1p/19q codeletion, which are both necessary for a firm diagnosis of oligodendrogliomas

IDH1 R132H Point Mutation by Immunohistochemistry with Reflex to IDH1 and IDH2 Mutation Analysis, Exon 4 3002134

Method: Immunohistochemistry

Use to identify the most common IDH mutation in diffuse gliomas

1p/19q Deletion by FISH 3001309

Method: Fluorescence in situ Hybridization (FISH)

Use when oligodendrogliomas are suspected

IDH1 R132H by Immunohistochemistry 2005857

Method: Immunohistochemistry

- Use when morphology indicates tumor may be a glioma
- Use to differentiate tumor from reactive gliosis

IDH1 R132H Point Mutation with Interpretation by Immunohistochemistry 2007357

Method: Immunohistochemistry

Includes pathologist interpretation

IDH1 and IDH2 Mutation Analysis Exon 4, Formalin-Fixed, Paraffin-Embedded (FFPE) Tissue 3004267

Method: Polymerase Chain Reaction/Sequencing

- *IDH1/IDH2* mutational status is a prognostic marker in individuals with low- and high-grade gliomas
- Aid in distinguishing a primary from a secondary glioblastoma

- Negative
  - Effectively rules out diagnosis of oligodendroglioma

Limitations

Test should not be used alone for the diagnosis of malignancy

Related Information

Primary Brain Tumors – Brain Tumor Molecular Markers

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