Brain, CNS, and PNS Cancer: Sequencing Panel

Test Code: MM204
Turnaround time: 4 weeks
CPT Codes: 81201 x1, 81292 x1, 81295 x1, 81298 x1, 81404 x1, 81405 x1, 81406 x1

Condition Description

Approximately 5% of primary brain cancers have known hereditary factors. Specifically, Li-Fraumeni syndrome, p53 defects, neurofibromatosis, tuberous sclerosis, von Hippel-Lindau disease, Turcot's syndrome, and familial polyposis increase the risk of brain tumors.

In 2013, an estimated 23,130 people in the United States will be diagnosed with primary malignant brain and other central nervous system (CNS) neoplasms.

References:

Genes

ALK, APC, ATM, MEN1, MLH1, MSH2, MSH6, NBN, NF2, PALB2, PHOX2B, PMS2, PTCH1, SUFU, TP53, VHL

Indications

The test is indicated for:
- Individuals with a clinical or suspected diagnosis of brain, CNS, or PNS cancer.

Methodology

Next Generation Sequencing: In-solution hybridization of all coding exons is performed on the patient's genomic DNA. Although some deep intronic regions may also be analyzed, this assay is not meant to interrogate most promoter regions, deep intronic regions, or other regulatory elements, and does not detect single or multi-exon deletions or duplications. Direct sequencing of the captured regions is performed using next generation sequencing. The patient's gene sequences are then compared to a standard reference sequence. Potentially causative variants and areas of low coverage are Sanger-sequenced. Sequence variations are classified as pathogenic, likely pathogenic, benign, likely benign, or variants of unknown significance. Variants of unknown significance may require further studies of the patient and/or family members.

Detection

Next Generation Sequencing: Clinical Sensitivity: Unknown. Mutations in the promoter region, some mutations in the introns and other regulatory element mutations cannot be detected by this analysis. Large deletions/duplications will not be detected by this analysis. Results of molecular analysis should be interpreted in the context of the patient's clinical/biochemical phenotype.

Analytical Sensitivity: ~99%.

Specimen Requirements

Submit only 1 of the following specimen types

Type: Whole Blood

Specimen Requirements:
In EDTA (purple top) tube:
- Infants (2 years): 3-5 ml
- Older Children & Adults: 5-10 ml.

Specimen Collection and Shipping: Ship sample at room temperature with overnight delivery.

Type: Isolated DNA

Specimen Requirements:
In microtainer: 60 ug
Isolation using the Qiagen™ Puregene kit for DNA extraction is recommended.

Specimen Collection and Shipping: Refrigerate until time of shipment in 100 ng/ul of TE buffer. Ship sample at room temperature with overnight delivery.

Disclaimer: This information is confidential and subject to change without notice. It may not be reproduced in whole or part unless authorized in writing by an authorized EGL representative.
Special Instructions

This test is for germline mutation analysis. DNA isolated from FFPE tumor samples is not suitable for this test.

Related Tests

- Hereditary Cancer Syndrome: Sequencing Panel.
- Brain, CNS, and PNS Cancer: Deletion/Duplication Panel.