

HistioTrak™ Test Information



The p. V600E is the most common oncogenic mutation located in the kinase domain (exon 15) of the *BRAF* gene. This hotspot mutation is commonly harbored by multiple tumors, including histiocytic, hematopoietic, brain, papillary thyroid, and melanocytic neoplasms. Mutation in the codon 600 results in the strong activation of the downstream MAPK cellular pathway.

A subset of tumors harboring the V600E mutation may be amenable to targeted treatment with *BRAF/MEK* inhibitors. The *BRAF V600E* mutation is the most prevalent mutation in Langerhans cell histiocytosis (LCH) and Erdheim-Chester Disease (ECD). In histiocytic neoplasms, minimal residual disease (MRD) in the peripheral blood can cause disease recurrence. In recent studies, droplet digital PCR (ddPCR) has been demonstrated as a sensitive tool to reliably detect MRD in patients with histiocytosis. HistioTrak™ is highly sensitive ddPCR-based molecular test that is designed for measuring MRD in patients with BRAF V600E driven histiocytic and other neoplasms.

Mutation Target:

BRAF c.1799T>A p.V600E

Reporting Units:

Qualitative: Positive / Suspicious / Negative

Acceptable Specimens:

- 3-5 mL Whole blood in EDTA tube
- 3-5 mL Bone marrow in EDTA tube
- FFPE-1 H&E slide and 6 unstained slides at 5 microns each
- Paraffin Block (block will be returned upon completion of test)

Shipping Conditions:

- For Peripheral blood and bone marrow samples, ship at 4°C in insulated container by overnight courier. Do NOT freeze.
- FFPE- 1 H&E slides and 6 unstained slides or paraffin blocks shipped at ambient temperature. For all samples, please ship for delivery on **Monday through Friday**.

Testing Schedule / TAT:

HistioTrak™ test is performed Mon-Fri. For testing outside of this schedule, call the lab at 513-636-9820.

TAT: 3-4 business days.

CPT Code:

81210

Methodology:

Droplet Digital Polymerase Chain Reaction (ddPCR) platform.

Contact Information:

Molecular and Genomic Pathology Services (MGPS)
Division of Pathology and Laboratory Medicine, CCHMC

Tel: 513-636-9820

Fax: 513-803-2941

Email: pathology@cchmc.org

Website: www.cincinnatichildrens.org/pathology

For pricing or billing questions, call 513-636-0120

Shipping Address:

ATTN: Molecular and Genomic Pathology Services (MGPS)

Division of Pathology and Laboratory Medicine
Cincinnati Children's Hospital Medical Center
3333 Burnet Avenue, R2040
Cincinnati, Ohio 45229-3039

References:

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2. Heritier S, et al. J Clin Oncol. 2016; 34: 3023-30
3. Picarsic J, et al, Acta Neuropathol Commun. 2019; 7:168
4. Goyal G, et al. Blood. 2020; 135: 1929-45
5. Lee L, et al. Blood Adv 2020; 4 (4): 717-727