

HistioTrak™ Qualitative Droplet Digital PCR



Langerhans cell histiocytosis (LCH), disseminated juvenile xanthogranuloma (JXG) family neoplasms, and pediatric Erdheim-Chester disease (ECD) are myeloid-derived clonal inflammatory disorders driven by recurrent kinase-activating mutations, most frequently in the MAPK signaling pathway. BRAF p.V600E is a hotspot mutation that strongly activates the MAPK pathway and is commonly harbored by multiple tumors, including histiocytic, hematopoietic, brain, papillary thyroid, and melanocytic neoplasms. It is the most prevalent mutation in LCH and ECD, carries prognostic significance, especially in multisystem LCH, and is a stable marker of histiocytoses that can be leveraged as an indicator of minimal residual disease (MRD). Targeted therapy with MAPK pathway inhibitors has been highly effective in the treatment of histiocytosis with mutations in pathway genes, including the BRAF p.V600E mutation¹⁻⁵. The HistioTrak™ test is a highly sensitive droplet digital PCR (ddPCR)-based molecular assay designed to identify BRAF V600E-driver mutation in diagnostic specimens of histiocytosis and other neoplasms on formalin-fixed, paraffin-embedded (FFPE) tissue specimens. For more information, call the lab at 513-636-9820.

Mutation Target:

BRAF c.1799T>A p.V600E

Reporting Units:

Positive / Suspicious / Negative

Acceptable Specimens:

- FFPE tissue: 1 H&E slide and 6 unstained slides at 5 microns each
- Paraffin block (block will be returned upon completion of test)

Unacceptable Specimens:

- Decalcified tissue
- Whole blood or bone marrow

Shipping Conditions:

- Slides or blocks at ambient temperature.
- **For all samples, ship for delivery on Monday through Friday. Receiving docks are CLOSED on weekends and holidays; do NOT ship if delivery will occur on those days.**

Testing Schedule:

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

TAT: 7 business days.

EPIC Test Code:

LAB00421 (1232400010)

CPT Codes:

81210 – Testing

G0452 – Physician interpretation

Clinical Lab Index:

HistioTrak™ Qualitative:

<https://www.testmenu.com/cincinnatichildrens/Tests/1130008>

Contact Information:

Cincinnati Children's Division of Pathology
Molecular and Genomic Pathology Services (MGPS)

Phone: 513-636-9820

Fax: 513-517-7099

Email: pathology@cchmc.org

Website: cincinnatichildrens.org/pathology

For pricing or billing questions, call 513-636-4261.

Shipping Address:

Cincinnati Children's Hospital Medical Center

Attn: Molecular and Genomic Pathology Services (MGPS)

3333 Burnet Ave, R2.001

Cincinnati, OH 45229

References:

1. Allen C, et al. *N Engl J Med*. 2018; 379: 856-868.
2. Heritier S, et al. *J Clin Oncol*. 2016; 34: 3023-3030.
3. Picarsic J, et al. *Acta Neuropathol Commun*. 2019; 7:168.
4. Goyal G, et al. *Blood*. 2020; 135: 1929-1945.
5. Lee L, et al. *Blood Adv* 2020; 4: 717-727.