

Hemiplegic Migraine With or Without Epilepsy Gene Panel, Varies

Reporting Title: Hemiplegic Migraine Gene Panel **Performing Location:** Rochester

Ordering Guidance:

Customization of this panel and single gene analysis for any gene present on this panel is available. For more information see CGPH / Custom Gene Panel, Hereditary, Next-Generation Sequencing, Varies.

Targeted testing for familial variants (also called site-specific or known variants testing) is available for the genes on this panel. See FMTT / Familial Variant, Targeted Testing, Varies.

Shipping Instructions:

Specimen preferred to arrive within 96 hours of collection.

Specimen Requirements:

Patient Preparation: A previous bone marrow transplant from an allogenic donor will interfere with testing. Call 800-533-1710 for instructions for testing patients who have received a bone marrow transplant.

Submit only 1 of the following specimens:

Specimen Type: Whole blood
Container/Tube:
Preferred: Lavender top (EDTA) or yellow top (ACD)
Acceptable: Any anticoagulant
Specimen Volume: 3 mL
Collection Instructions:

Invert several times to mix blood.
Send whole blood specimen in original tube. Do not aliquot.

Specimen Stability Information: Ambient (preferred)/Refrigerated

Specimen Type: Skin biopsy
Supplies: Fibroblast Biopsy Transport Media (T115)
Container/Tube: Sterile container with any standard cell culture media (eg, minimal essential media, RPMI 1640). The solution should be supplemented with 1% penicillin and streptomycin.
Specimen Volume: 4-mm punch
Specimen Stability Information: Refrigerated (preferred)/Ambient
Additional Information: A separate culture charge will be assessed under CULFB / Fibroblast Culture for Biochemical or Molecular Testing . An additional 3 to 4 weeks is required to culture fibroblasts before genetic testing can occur.

Specimen Type: Cultured fibroblast Container/Tube: T-25 flask Specimen Volume: 2 Flasks Collection Instructions: Submit confluent cultured fibroblast cells from a skin biopsy from another laboratory. Cultured cells from a prenatal specimen will not be accepted.

Specimen Stability Information: Ambient (preferred)/Refrigerated (<24 hours)



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Additional Information: A separate culture charge will be assessed under CULFB / Fibroblast Culture for Biochemical or Molecular Testing. An additional 3 to 4 weeks is required to culture fibroblasts before genetic testing can occur.

Specimen Type: Blood spot

Supplies: Card-Blood Spot Collection (Filtration Paper) (T493)

Container/Tube:

Preferred: Collection card (Whatman Protein Saver 903 Paper)

Acceptable: PerkinElmer 226 (formerly Ahlstrom 226) filter paper or blood spot collection card

Specimen Volume: 5 Blood spots

Collection Instructions:

1. An alternative blood collection option for a patient 1 year of age or older is a fingerstick. For infants younger than 1 year, a heel stick should be used. See <u>Dried Blood Spot Collection Tutorial</u> for how to collect blood spots via fingerstick.

- 2. Let blood dry on the filter paper at ambient temperature in a horizontal position for a minimum of 3 hours.
- 3. Do not expose specimen to heat or direct sunlight.
- 4. Do not stack wet specimens.
- 5. Keep specimen dry.
- Specimen Stability Information: Ambient (preferred)/Refrigerated

Additional Information:

1. Due to lower concentration of DNA yielded from blood spot, it is possible that additional specimen may be required to complete testing.

- 2. For collection instructions, see <u>Blood Spot Collection Instructions</u>
- 3. For collection instructions in Spanish, see <u>Blood Spot Collection Card-Spanish Instructions</u> (T777)
- 4. For collection instructions in Chinese, see <u>Blood Spot Collection Card-Chinese Instructions</u> (T800)

Specimen Type: Saliva

Patient Preparation: Patient should not eat, drink, smoke, or chew gum 30 minutes prior to collection.

Supplies: Saliva Swab Collection Kit (T786)

Specimen Volume: 1 Swab

Collection Instructions: Collect and send specimen per kit instructions.

Additional Information: Due to lower concentration of DNA yielded from saliva, it is possible that additional specimen may be required to complete testing.

Specimen Stability Information: Ambient 30 days

Forms:

1. New York Clients-Informed consent is required.

Document on the request form or electronic order that a copy is on file.

The following documents are available in Special Instructions:

-Informed Consent for Genetic Testing (T576)

-Informed Consent for Genetic Testing (Spanish) (T826)

2. Molecular Genetics: Neurology Patient Information

3. If not ordering electronically, complete, print, and send a <u>Neurology Specialty Testing Client Test Request</u> (T732) with the specimen.

Specimen Type Temperature	Time	Special Container
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Varies	Varies	

Result Codes:

Result ID	Reporting Name	Туре	Unit	LOINC®
616525	Test Description	Alphanumeric		62364-5
616526	Specimen	Alphanumeric		31208-2
616527	Source	Alphanumeric		31208-2
616528	Result Summary	Alphanumeric		50397-9
616529	Result	Alphanumeric		82939-0
616530	Interpretation	Alphanumeric		69047-9
616531	Resources	Alphanumeric		In Process
616532	Additional Information	Alphanumeric		48767-8
616533	Method	Alphanumeric		85069-3
616534	Genes Analyzed	Alphanumeric		82939-0
616535	Disclaimer	Alphanumeric		62364-5
616536	Released By	Alphanumeric		18771-6

LOINC[®] and CPT codes are provided by the performing laboratory.

Supplemental Report:

Supplemental

CPT Code Information:

81185
81405
81406 x 2
81407
81408
81479 (if appropriate for government payers)
88233-Tissue culture, skin, solid tissue biopsy (if appropriate)
88240-Cryopreservation (if appropriate)

Reflex Tests:

Test Id	Reporting Name	CPT Units	CPT Code	Always Performed	Available Separately
CULFB	Fibroblast Culture for Genetic Test	1	88233	No	Yes

Result Codes for Reflex Tests:

Test ID	Result ID	Reporting Name	Туре	Unit	LOINC®
CULFB	52327	Result Summary	Alphanumeric		50397-9



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CULFB	52329	Interpretation	Alphanumeric	69965-2
CULFB	52328	Result	Alphanumeric	82939-0
CULFB	CG770	Reason for Referral	Alphanumeric	42349-1
CULFB	CG899	Specimen	Alphanumeric	31208-2
CULFB	52331	Source	Alphanumeric	31208-2
CULFB	52332	Method	Alphanumeric	85069-3
CULFB	54625	Additional Information	Alphanumeric	48767-8
CULFB	52333	Released By	Alphanumeric	18771-6

Reference Values:

An interpretive report will be provided.