

Reporting Title: Emery-Dreifuss Gene Panel
Performing Location: Rochester

Ordering Guidance:
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. To obtain more information about this testing option, call 800-533-1710.

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

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

Collection Instructions:
1. Invert several times to mix blood.
2. Send whole blood specimen in original tube. **Do not aliquot.**
Specimen Stability Information: Ambient (preferred)/Refrigerated

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:
[-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 [Neurology Specialty Testing Client Test Request](#) (T732) with the specimen.

| Specimen Type | Temperature | Time | Special Container |
|---------------|-------------|------|-------------------|
| Varies | Varies | | |

Result Codes:

| Result ID | Reporting Name | Type | Unit | LOINC® |
|-----------|------------------|--------------|------|---------|
| 617559 | Test Description | Alphanumeric | | 62364-5 |
| 617560 | Specimen | Alphanumeric | | 31208-2 |
| 617561 | Source | Alphanumeric | | 31208-2 |

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|--------|------------------------|--------------|--|---------|
| 617562 | Result Summary | Alphanumeric | | 50397-9 |
| 617563 | Result | Alphanumeric | | 82939-0 |
| 617564 | Interpretation | Alphanumeric | | 69047-9 |
| 618179 | Additional Results | Alphanumeric | | 82939-0 |
| 617565 | Resources | Alphanumeric | | 99622-3 |
| 617566 | Additional Information | Alphanumeric | | 48767-8 |
| 617567 | Method | Alphanumeric | | 85069-3 |
| 617568 | Genes Analyzed | Alphanumeric | | 48018-6 |
| 617569 | Disclaimer | Alphanumeric | | 62364-5 |
| 617570 | Released By | Alphanumeric | | 18771-6 |

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

Supplemental Report:

Supplemental

CPT Code Information:

- 81405
- 81404
- 81406 x 2
- 81479
- 81479 (if appropriate for government payers)

Reference Values:

An interpretive report will be provided.