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**Reporting Title:** Tyrosine Disorders Gene Panel**Performing Location:** Mayo Clinic Laboratories - Rochester Main Campus**Ordering Guidance:**

The recommended first-tier biochemical tests include quantitative plasma amino acids (AAQP / Amino Acids, Quantitative, Plasma), urine organic acids (OAU / Organic Acids Screen, Random, Urine), and measurement of succinylacetone (SUAC / Succinylacetone, Blood Spot).

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 mutations 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.

**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. For instructions for testing patients who have received a bone marrow transplant, call 800-533-1710.

**Submit only 1 of the following specimens:**

**Specimen Type:** Whole blood

**Container/Tube:** Lavender top (EDTA) or yellow top (ACD)

**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) 4 days/Refrigerated 14 days

**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)  
**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 (Filter 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 older than 1 year is a fingerstick. For detailed instructions, see [How to Collect Dried Blood Spot Samples](#).
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 [Blood Spot Collection Instructions](#)
3. For collection instructions in Spanish, see [Blood Spot Collection Card-Spanish Instructions](#) (T777)
4. For collection instructions in Chinese, see [Blood Spot Collection Card-Chinese Instructions](#) (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.  
**Specimen Stability Information:** Ambient 30 days  
**Additional Information:** Due to lower concentration of DNA yielded from saliva, it is possible that additional specimen may be required to complete testing.

- 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: Biochemical Disorders Patient Information](#) (T527)
  3. If not ordering electronically, complete, print, and send a [Biochemical Genetics Test Request](#) (T798) with the specimen.

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

Result Codes:

| Result ID | Reporting Name         | Type         | Unit | LOINC®  |
|-----------|------------------------|--------------|------|---------|
| 608800    | Test Description       | Alphanumeric |      | 62364-5 |
| 608801    | Specimen               | Alphanumeric |      | 31208-2 |
| 608802    | Source                 | Alphanumeric |      | 31208-2 |
| 608803    | Result Summary         | Alphanumeric |      | 50397-9 |
| 608804    | Result                 | Alphanumeric |      | 82939-0 |
| 608805    | Interpretation         | Alphanumeric |      | 69047-9 |
| 608806    | Resources              | Alphanumeric |      | 99622-3 |
| 608807    | Additional Information | Alphanumeric |      | 48767-8 |
| 608808    | Method                 | Alphanumeric |      | 85069-3 |
| 608809    | Genes Analyzed         | Alphanumeric |      | 48018-6 |
| 608810    | Disclaimer             | Alphanumeric |      | 62364-5 |
| 608811    | Released By            | Alphanumeric |      | 18771-6 |

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

Supplemental Report:  
Supplemental

CPT Code Information:  
81406  
81479  
88233-Tissue culture, skin, solid tissue biopsy (if appropriate)  
88240-Cryopreservation (if appropriate)  
81479 (if appropriate for government payers)

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      | Type         | Unit | LOINC®  |
|---------|-----------|---------------------|--------------|------|---------|
| CULFB   | 52327     | Result Summary      | Alphanumeric |      | 50397-9 |
| 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.