Overview

Useful For
Carrier screening for Fanconi anemia in individuals of Ashkenazi Jewish ancestry
Prenatal diagnosis of Fanconi anemia in at-risk pregnancies
Confirmation of suspected clinical diagnosis of Fanconi anemia in individuals of Ashkenazi Jewish ancestry

Reflex Tests

<table>
<thead>
<tr>
<th>Test ID</th>
<th>Reporting Name</th>
<th>Available Separately</th>
<th>Always Performed</th>
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</thead>
<tbody>
<tr>
<td>CULFB</td>
<td>Fibroblast Culture for Genetic Test</td>
<td>Yes</td>
<td>No</td>
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<tr>
<td>CULAF</td>
<td>Amniotic Fluid Culture/Genetic Test</td>
<td>Yes</td>
<td>No</td>
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<tr>
<td>MATCC</td>
<td>Maternal Cell Contamination, B</td>
<td>Yes</td>
<td>No</td>
</tr>
</tbody>
</table>

Testing Algorithm

For prenatal specimens only: If amniotic fluid (nonconfluent cultured cells) is received, amniotic fluid culture will be added and charged separately. If chorionic villus specimen (nonconfluent cultured cells) is received, fibroblast culture will be added and charged separately. For any prenatal specimen that is received, maternal cell contamination studies will be added.

Special Instructions

- [Molecular Genetics: Biochemical Disorders Patient Information](#)
- [Informed Consent for Genetic Testing](#)
- [Informed Consent for Genetic Testing (Spanish)](#)

Method Name
Polymerase Chain Reaction (PCR) analysis is used to test for the following mutations associated with Fanconi anemia: 322delG and IVS4(+4)A->T

NY State Available
Yes

Specimen

Specimen Type
Varies

Additional Testing Requirements
All prenatal specimens must be accompanied by a maternal blood specimen.
- Order MATCC / Maternal Cell Contamination, Molecular Analysis on the maternal specimen.
Shipping Instructions
Specimen preferred to arrive within 96 hours of collection.

Prenatal specimens can be sent Monday through Thursday and must be received by 5 p.m. CST on Friday in order to be processed appropriately.

Specimen Required

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: Yellow top (ACD) or lavender top (EDTA)
Acceptable: Any anticoagulant
Specimen Volume: 2.6 mL

Collection Instructions:
1. Invert several times to mix blood.
2. Send specimen in original tube.

Specimen Stability Information: Ambient (preferred)/Refrigerated/Frozen

Prenatal Specimens

Due to the complexity of prenatal testing, consultation with the laboratory is required for all prenatal testing.

Specimen Type: Amniotic fluid

Container/Tube: Amniotic fluid container
Specimen Volume: 20 mL

Specimen Stability Information: Refrigerated (preferred)/Ambient

Specimen Type: Chorionic villi

Container/Tube: 15 mL tube containing 15 mL of transport media
Specimen Volume: 20 mg

Specimen Stability Information: Refrigerated
Acceptable:

**Specimen Type:** Confluent cultured cells

**Container/Tube:** T-25 flask

**Specimen Volume:** 2 flasks

**Collection Instructions:** Submit confluent cultured cells from another laboratory.

**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 in Special Instructions:
   - Informed Consent for Genetic Testing (T576)
   - Informed Consent for Genetic Testing-Spanish (T826)

2. **Molecular Genetics: Biochemical Disorders Patient Information** (T527) in Special Instructions.

**Specimen Minimum Volume**

- Blood: 0.5 mL; Amniotic Fluid: 10 mL; Chorionic Villi: 5 mg

**Reject Due To**

All specimens will be evaluated by Mayo Clinic Laboratories for test suitability.

**Specimen Stability Information**

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<th>Temperature</th>
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<tr>
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**Clinical and Interpretive**

**Clinical Information**

Fanconi anemia is an aplastic anemia that leads to bone marrow failure and myelodysplasia or acute myelogenous leukemia. Physical findings include short stature; upper limb, lower limb, and skeletal malformations; and abnormalities of the eyes and genitourinary tract. The proteins encoded by the genes associated with Fanconi anemia may work together to repair DNA damage.

Mutations in several genes have been associated with Fanconi anemia, although 1 mutation, IVS4(+4)A->T in the *FANCC* gene has been shown to be common in the Ashkenazi Jewish population. The carrier rate in the Ashkenazi Jewish population is 1 in 89 and the detection rate for this mutation using this assay is greater than 99%. A second *FANCC* mutation, 322delG, is overrepresented in patients of Northern European ancestry.

**Reference Values**

An interpretive report will be provided.
Test Definition: FANCP
Fanconi Anemia, Mutation Analysis

Interpretation
An interpretive report will be provided.

Cautions
This test is not recommended as a first-tier test to diagnose Fanconi anemia in individuals of non-Ashkenazi Jewish descent. In the non-Ashkenazi Jewish population, the recommended test is cytogenetic testing in the presence of mitomycin C or diepoxybutane (DEB), which may provide useful diagnostic information.

This assay will not detect all of the mutations that cause Fanconi anemia. Therefore, the absence of a detectable mutation does not rule out the possibility that an individual is a carrier of or affected with this disease.

Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Errors in our interpretation of results may occur if information given is inaccurate or incomplete.

Rare polymorphisms exist that could lead to false-negative or false-positive results. If results obtained do not match the clinical findings, additional testing should be considered.

In rare cases, DNA alterations of undetermined significance may be identified.

Clinical Reference

Performance

Method Description

PDF Report
No

Day(s) and Time(s) Test Performed
Tuesday; 10 a.m.

Analytic Time
9 days

Maximum Laboratory Time
12 days

Specimen Retention Time
Whole Blood: 2 weeks (if available) Extracted DNA: 3 months

Performing Laboratory Location
Test Definition: FANCP
Fanconi Anemia, Mutation Analysis

Rochester

Fees and Codes

Fees
- Authorized users can sign in to Test Prices for detailed fee information.
- Clients without access to Test Prices can contact Customer Service 24 hours a day, seven days a week.
- Prospective clients should contact their Regional Manager. For assistance, contact Customer Service.

Test Classification
This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

CPT Code Information
81242-FANCC (Fanconi anemia, complementation group C) (eg, Fanconi anemia, type C) gene analysis, common variant (eg, IVS4+4A->T)

Fibroblast Culture for Genetic Test
88233-Tissue culture, skin or solid tissue biopsy (if appropriate)

88240-Cryopreservation (if appropriate)

Amniotic Fluid Culture/Genetic Test
88235-Tissue culture for amniotic fluid (if appropriate)

88240-Cryopreservation (if appropriate)

Maternal Cell Contamination, B

81265-Comparative analysis using Short Tandem Repeat (STR) markers; patient and comparative specimen (eg, pre-transplant recipient and donor germline testing, post-transplant non-hematopoietic recipient germline [eg, buccal swab or other germline tissue sample] and donor testing, twin zygosity testing or maternal cell contamination of fetal cells (if appropriate)

LOINC® Information

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