

Family Member Comparator Specimen for Exome Sequencing, Varies

Test ID: CMPRE

Useful for:

Submitting a biological family member's specimen to be used as a comparator for affected patients (probands) undergoing whole exome sequencing

Reflex Tests:

Test ID	Reporting Name	Available Separately	Always Performed
FIBR	Fibroblast Culture	Yes	No
CRYOB	Cryopreserve for Biochem Studies	No	No

Methods:

Sequence Capture and Targeted Next-Generation Sequencing followed by Sanger Sequencing or Quantitative Polymerase Chain Reaction (qPCR), as needed

Reference Values:

An interpretive report will be provided.

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:	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
Minimum Volume:	1 mL

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 FIBR / Fibroblast Culture for Biochemical and Molecular Testing, Tissue . An additional 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 FIBR / Fibroblast Culture for Biochemical and Molecular Testing, Tissue. An additional 4 weeks is required to culture fibroblasts before genetic testing can occur.

Specimen Type: Blood spot

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

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 fingerstick. For infants younger than 1 year, a heel stick should be used. See [Dried Blood Spot Collection Tutorial](#) 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. For collection instructions, see [Blood Spot Collection Instructions](#)
2. For collection instructions in Spanish, see [Blood Spot Collection Card-Spanish Instructions](#) (T777)
3. For collection instructions in Chinese, see [Blood Spot Collection Card-Chinese Instructions](#) (T800)
4. Due to lower concentration of DNA yielded from blood spot, it is possible that additional specimen may be required to complete testing.

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.

Note:

1. [Whole Exome Sequencing: Ordering Checklist](#) is required.
2. New York Clients-Informed consent is required, included in the above form. Document on the request form or electronic order that a copy is on file.

Additional Testing Requirements:

To order whole exome testing for the patient and the family member comparator specimens, see the following steps:

1. Order this test on the patient (proband).
2. Order CMPRE / Family Member Comparator Specimen for Exome Sequencing, Varies on all family members' specimens being submitted as comparators.
 - a. When available, the patient's biological mother and biological father are the preferred family member comparators.
 - b. If one or both of the patient's biological parents are not available for testing, specimens from other first-degree relatives (siblings or children) can be used as comparators. Contact the laboratory at 800-533-1710 for approval to send specimens from other relatives.
 - c. The cost of analysis for family member comparator specimens is applied to the patient's (proband's) test. Family members will not be charged separately.
3. Collect patient (proband) and family member specimens. Label specimens with full name and birthdate. Do not label family members' specimens with the proband's name.
4. Complete the signature sections of the Informed Consent (required for New York State clients) portion of [Whole Exome Sequencing: Ordering Checklist](#).
5. If the patient wishes to opt-out of receiving secondary findings or change the DNA storage selection, select the appropriate boxes in the Informed Consent section.
6. Attach clinic notes from specialists relevant to patient's clinical features, if available.
7. Attach pedigree, if available.
8. Send paperwork to the laboratory along with the specimens. If not sent with the specimen, fax a copy of the paperwork to 507-284-1759, Attn: WES Genetic Counselors.

Specimen Stability Information:

Specimen Type	Temperature	Time	Special Container
Varies	Ambient (preferred)		
	Frozen		
	Refrigerated		

Cautions:

This testing is intended to be used for biological family members whose specimens are being submitted as comparators for affected patients (probands) undergoing whole exome sequencing. Although test results will only be provided in the context of the proband, it is possible for family members serving as comparators to learn unexpected genetic information about themselves, for example, if biological relationships are not as described. It is also possible for individuals to learn that they carry certain genetic variants that are being reported in the proband.

If the patient has had an allogeneic hematopoietic stem cell transplant or a recent heterologous blood transfusion, results may be inaccurate due to the presence of donor DNA. Call Mayo Clinic Laboratories for instructions for testing patients who have received a bone marrow transplant.

A genetic consultation is recommended for patients undergoing this test, both prior to testing and after results are available.

Day(s) Performed: Varies **Report Available:** 12 weeks

Questions

Contact Michelle Rath, Laboratory Technologist Resource Coordinator at 800-533-1710.