
Reporting Title: Family Member Comparator for Genome**Performing Location:** Rochester**Ordering Guidance:**

This test is not appropriate for affected patients (probands) undergoing whole genome sequencing (WGS). This test is intended to be ordered for biological family member comparator specimens only. For WGS testing for the proband, order WGS DX / Whole Genome Sequencing for Hereditary Disorders, Varies. If this test is ordered on a proband, the test will be canceled and WGS DX will be performed as the appropriate test.

If this test is ordered on a family member comparator of a proband having WES DX / Whole Exome Sequencing for Hereditary Disorders, Varies; or WES MT / Whole Exome and Mitochondrial Genome Sequencing, Varies, this test will be canceled and CMPRE / Family Member Comparator Specimen for Exome Sequencing, Varies performed as the appropriate test.

Each specimen must be on a separate order.

Additional Testing Requirements:

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

1. Order WGS DX / Whole Genome Sequencing for Hereditary Disorders, Varies on the patient (proband).
2. Order this test 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. Testing typically includes up to 2 family member comparators. Contact the laboratory at 800-533-1710 for approval to send specimens from other relatives or to send the patient and 3 first-degree relatives (quad).
 - 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. For each family, complete the following portions of the Whole Genome Sequencing: Ordering Checklist. A separate form is not needed for each family member.
 - a. Patient Information is required for all clients.
 - b. Informed Consent is required for New York State clients.
 - c. 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.
5. Attach clinic notes from specialists relevant to patient's clinical features, if available.
6. Attach pedigree, if available.
7. Send paperwork to the laboratory along with the specimens. If not sent with the specimens, fax a copy of the paperwork to 507-284-1759, Attn: WGS Genetic Counselors.

For more information see Whole Exome and Genome Sequencing Information and Test Ordering Guide.

Shipping Instructions:

Specimen preferred to arrive within 96 hours of collection.



Necessary Information:

Whole Genome Sequencing: Ordering Checklist is required for all clients, and Informed Consent is required for New York clients. Fill out one form for the family and send with the specimens.

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 for each family member.

Specimen Type: Whole blood

Container/Tube:

Preferred: 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

Additional Information: To ensure minimum volume and concentration of DNA is met, the preferred volume of blood must be submitted. Testing may be canceled if DNA requirements are inadequate.

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: 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 quantity/quality of DNA yielded from saliva, some aspects of the test may not perform as well as DNA extracted from a whole blood sample. When applicable, specific gene regions that were unable to be interrogated will be noted in the report. Alternatively, additional specimen may be required to complete testing.

Specimen Type: Muscle tissue biopsy

Supplies: Muscle Biopsy Kit (T541)

Collection Instructions: Prepare and transport specimen per instructions in Muscle Biopsy Specimen Preparation Instructions.

Specimen Volume: 10 to 80 mg

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

Specimen Minimum Volume:

Whole blood: 1 mL; Other specimen types: See Specimen Required

Forms:

1. Whole Genome 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.

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

Ask at Order Entry (AOE) Questions:

Test ID	Question ID	Description	Type	Reportable
CULFB	CG770	Reason for Referral	Plain Text	No
CULFB	CG899	Specimen	Plain Text	No

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
614930	Interpretation	Alphanumeric		69047-9
614931	Specimen	Alphanumeric		31208-2

Result ID	Reporting Name	Type	Unit	LOINC®
614932	Source	Alphanumeric		31208-2
614941	Released By	Alphanumeric		18771-6

LOINC and CPT codes are provided by the performing laboratory.

Supplemental Report:
Supplemental**Reflex Tests:**

Test ID	Reporting Name	CPT Units	CPT Code	Always Performed	Orderable Separately
CULFB	Fibroblast Culture for Genetic Test			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.