
Reporting Title: Whole Genome Sequencing Reanalysis**Performing Location:** Rochester**Ordering Guidance:**

This test is only appropriate for patients who have previously had WGSDX / Whole Genome Sequencing for Hereditary Disorders, Varies performed by Mayo Clinic Laboratories.

This test is for affected patients (probands) only. This test does not need to be ordered for family member comparators (CMPRG / Family Member Comparator Specimen for Genome Sequencing, Varies).

Additional Testing Requirements:

DNA specimens from the patient (proband) and all family member comparators included in the original whole genome sequencing test are required to allow for confirmation of any new reportable variants, based on internal laboratory criteria. For most patients, stored DNA from the original whole genome sequencing test should be available for this testing.

To use stored DNA for this test:

Order WGSR / Whole Genome Sequencing Reanalysis, Varies. By calling Mayo Clinic Laboratories at 800-533-1710 to request that this test be added on to the remaining DNA specimen for the patient (proband). The laboratory will determine if there is sufficient DNA remaining for the proband and all comparators to perform confirmation of any new results. If there is sufficient DNA, the order will proceed.

If the patient and/or family member comparators are found to have an insufficient quantity of stored DNA, follow the instructions below:

1. If there is not sufficient DNA remaining for the patient (proband): If an order for WGSR was already placed in the steps above, the order will be canceled, and the client notified of the test cancellation. Collect a new proband specimen and order WGSR for the new specimen.
2. If there is not sufficient DNA remaining for one or more family member comparators: For the family members who were included as comparators in the original whole genome sequencing test but do not have sufficient stored DNA, collect new comparator specimens and order CMPRG / Family Member Comparator Specimen for Genome Sequencing, Varies for the new specimens.

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

Shipping Instructions:

Specimens are preferred to arrive within 96 hours of collection.

Necessary Information:

Whole Genome Sequencing: Ordering Checklist is required for all patients. Complete the following sections on pages 2 through 4:

Patient (Proband) Information

Reason for Testing: provide reason for reanalysis request

Provide new information in:

-Patient (Proband) Suspected Diagnoses

- Patient (Proband) Clinical Evaluations
- Patient (Proband) Clinical Features

Attach clinic notes and pedigree with any relevant new clinical or family history information.

Fax the paperwork, clinic notes, and pedigree to 507-284-1759, Attn: WGS Genetic Counselors.

Specimen Requirements:

For most patients, a new specimen submission will not be required. Testing can be performed using stored DNA from the original whole genome sequencing test. To order testing on the stored specimen, see Additional Testing 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.

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. 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	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®
618564	Interpretation	Alphanumeric		69047-9
618565	Specimen	Alphanumeric		31208-2

Result ID	Reporting Name	Type	Unit	LOINC®
618566	Source	Alphanumeric		31208-2
618567	Released By	Alphanumeric		18771-6

LOINC and CPT codes are provided by the performing laboratory.

Supplemental Report:

Supplemental

CPT Code Information:

First reanalysis: No charge
81427-For all subsequent reanalysis requests

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.