Reporting Title: Whole Exome Sequencing Reanalysis
Performing Location: Rochester

Ordering Guidance:
This test is only appropriate for patients who have previously had one of the following whole exome sequencing tests performed by Mayo Clinic Laboratories:
WES / Whole Exome Sequencing, Varies
WESDX / Whole Exome Sequencing for Hereditary Disorders, Varies
WESPP / Whole Exome Sequencing Plus Pharmacogenomics
WESPM / Whole Exome Sequencing plus Whole Mitochondrial Genome Sequencing, Varies
WESMT / Whole Exome and Mitochondrial Genome Sequencing, Varies

If the patient has not had one of the above tests performed previously, consider either WESDX / Whole Exome Sequencing for Hereditary Disorders, Varies or WESMT / Whole Exome and Mitochondrial Genome Sequencing, Varies.

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

Additional Testing Requirements:
DNA specimens from the patient (proband) and all family member comparators included in the original whole exome 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 exome sequencing test should be available for this testing.

To use stored DNA for this test:
Order WESR / Whole Exome Sequencing Reanalysis, Varies by calling Mayo Clinic Laboratories at 800-533-1710 and requesting 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 WESR 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 WESR 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 exome sequencing test but do not have sufficient stored DNA, collect new comparator specimens and order CMPRE / Family Member Comparator Specimen for Exome 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:
From the Whole Exome Sequencing: Ordering Checklist, Patient Information is required for all patients.
Complete the following sections on pages 2 through 4:
Patient (Proband) Information
Provide reason for reanalysis request in Reason for Testing
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: WES 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 exome 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. 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: 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

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 fibroblasts
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 (Filtration 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. Whole Exome Sequencing: Ordering Checklist, Patient Information 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®
616885	Interpretation	Alphanumeric		69047-9
616886	Specimen	Alphanumeric		31208-2
616887	Source	Alphanumeric		31208-2
616888	Released By	Alphanumeric		18771-6

LOINC and CPT codes are provided by the performing laboratory.

Supplemental Report:
Supplemental

CPT Code Information:

The first reanalysis: No charge
For all subsequent reanalysis requests: 81417

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

Test Definition: WESR

Whole Exome Sequencing Reanalysis, Varies

Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
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.