MAYO CLINIC LABORATORIES

NEW TEST

Notification Date: December 15, 2022
Effective Date: December 20, 2022

Family Member Comparator Specimen for Genome Sequencing, Varies

Test ID: CMPRG

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

Genetics Information:
This test is for the biological family members whose specimens are being submitted as comparators for patients undergoing WGSDX / Whole Genome Sequencing for Hereditary Disorders, Varies.

Methods:
Polymerase Chain Reaction-Free Next-Generation Sequencing followed by Sanger Sequencing, Quantitative Polymerase Chain Reaction (qPCR), or other methods, 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.

Specimen Type:	**Whole blood**
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.**
Minimum Volume:	1 mL

Note:
Specimen preferred to arrive within 96 hours of collection

Specimen Stability Information:

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

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 WGSDX / Whole Genome Sequencing for Hereditary Disorders, Varies. If this test is ordered on a proband, the test will be canceled and WGSDX will be 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 WGSDX / 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 Genome Sequencing (WGS): Questions and Answers for Providers.

Necessary Information:

Whole Genome Sequencing: Ordering Checklist **Patient Information is required for all patients**.

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 genome 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, it is possible for individuals to learn that they carry certain genetic variants that are being reported in the proband, or that biological relationships are not as described.
- It is not a guarantee that patient data will be stored indefinitely.
- If the patient has had an allogeneic hematopoietic stem cell transplant or a recent 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.

CPT Code:

No associated CPT codes

Day(s) Performed: Varies

Report Available: 12 weeks

Questions:

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