Complete and print.

Reset Form

MAYO CLINIC
LABORATORIES

Whole Genome Sequencing: Ordering Checklist

Instructions: Select the box for the test requested on the patient (proband) and complete the corresponding ordering checklist.

☐ Whole Genome Sequencing for Hereditary Disorders

- ☐ For the patient (proband), order WGSDX / Whole Genome Sequencing for Hereditary Disorders, Varies.
- ☐ For each family member specimen that will be submitted as a comparator, order CMPRG / Family Member Comparator Specimen for Genome Sequencing, Varies. Separate orders need to be placed for each family member. Biological parents are the preferred family member comparators; see test catalog for additional information.
- ☐ Collect patient (proband) and family member specimens. Label specimens with full name and birth date. Do not label family members' specimens with the proband's name. See test catalog for specimen requirements.
- ☐ Complete the Patient Information form on pages 2–4 (required for all clients).
- ☐ Complete the signature sections of the Informed Consent on page 7 (required for New York State clients).
- ☐ If the patient wishes to opt out of receiving secondary findings or change the DNA storage selection, select the appropriate boxes on page 7.
- ☐ Attach clinic notes from specialists relevant to patient's clinical features.
- ☐ Attach pedigree.
- ☐ Send paperwork, clinic notes, and pedigree along with specimens. If not sent with the specimen, fax a copy of the paperwork to 507-284-1759, Attn: WGS Genetic Counselors.

☐ Whole Genome Sequencing Reanalysis

- ☐ For the patient (proband), order WGSR / Whole Genome Sequencing Reanalysis, Varies.
- ☐ Call Mayo Clinic Laboratories at 800-533-1710 and request that WGSR is added on to remaining DNA specimen from the original whole genome sequencing test. If the laboratory determines that the patient previously opted out of DNA storage or the specimen was depleted, a new specimen will be requested. See test catalog for specimen requirements.
- ☐ Complete the following sections of the Whole Genome Sequencing paperwork:
    - Patient (Proband) Information (page 2)
    - Provide reason for reanalysis request in Reason for Testing (page 2)
    - Provide **new** information in Suspected Diagnoses (page 3), Patient (Proband) Clinical Evaluations (page 3), and Patient (Proband) Clinical Features (page 4)
- ☐ Attach clinic notes and/or a 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.

Questions: Call with any questions and ask to speak to a WGS genetic counselor at 507-293-7299.

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Whole Genome Sequencing: Patient Information

Label Here

Instructions: Provide the requested information below for appropriate interpretation of this test.
In addition, submit relevant clinic notes and pedigree.

Patient (Proband) Information (required)

Patient Name *(Last, First, Middle)*		Birth Date *(mm-dd-yyyy)*
Medical Record No.	Sex Assigned at Birth ☐ Male ☐ Female ☐ Unknown	Legal/Administrative Sex ☐ Male ☐ Female ☐ Nonbinary

Referring Provider Information

Referring Provider Name *(Last, First)*	Phone	Fax*
Other Contact/Geneticist/Genetic Counselor *(Last, First)*	Phone	Fax*

**Fax number given must be from a fax machine that complies with applicable HIPAA regulations.*

Reason for Testing

Biological Family Member Information

Required information for each family member whose specimen is being sent as a comparator sample. The priority should always be to include both parents as comparators, if possible. Contact a genetic counselor at 507-293-7299 to discuss sending more than 2 comparators or comparators that are not first degree relatives.

Family Member 1 Information ☐ Sent with proband specimen ☐ To be sent later—must be received within 3 weeks of proband specimen. Testing will proceed at that time with specimens that have been received.

Name *(Last, First, Middle)*	Medical Record No.	Birth Date *(mm-dd-yyyy)*
Sex Assigned at Birth ☐ Male ☐ Female ☐ Unknown	Legal/Administrative Sex ☐ Male ☐ Female ☐ Nonbinary	
Relationship to Proband ☐ Mother ☐ Father ☐ Full sibling ☐ Maternal half-sibling ☐ Paternal half-sibling ☐ Child ☐ Other relatives are accepted on a case-by-case basis; contact a genetic counselor at 507-293-7299 to discuss before ordering testing:		
REQUIRED: Does this relative share any relevant clinical features or clinical history with the patient? ☐ No ☐ Yes If "Yes," describe:		

Family Member 2 Information ☐ Sent with proband specimen ☐ To be sent later—must be received within 3 weeks of proband specimen. Testing will proceed at that time with specimens that have been received.

Name *(Last, First, Middle)*	Medical Record No.	Birth Date *(mm-dd-yyyy)*
Sex Assigned at Birth ☐ Male ☐ Female ☐ Unknown	Legal/Administrative Sex ☐ Male ☐ Female ☐ Nonbinary	
Relationship to Proband ☐ Mother ☐ Father ☐ Full sibling ☐ Maternal half-sibling ☐ Paternal half-sibling ☐ Child ☐ Other relatives are accepted on a case-by-case basis; contact a genetic counselor at 507-293-7299 to discuss before ordering testing:		
REQUIRED: Does this relative share any relevant clinical features or clinical history with the patient? ☐ No ☐ Yes If "Yes," describe:		

Family Member 3 Information ☐ Sent with proband specimen ☐ To be sent later—must be received within 3 weeks of proband specimen. Testing will proceed at that time with specimens that have been received.
Contact the laboratory to discuss sending a third comparator sample.

Name *(Last, First, Middle)*	Medical Record No.	Birth Date *(mm-dd-yyyy)*
Sex Assigned at Birth ☐ Male ☐ Female ☐ Unknown	Legal/Administrative Sex ☐ Male ☐ Female ☐ Nonbinary	
Relationship to Proband ☐ Mother ☐ Father ☐ Full sibling ☐ Maternal half-sibling ☐ Paternal half-sibling ☐ Child ☐ Other relatives are accepted on a case-by-case basis; contact a genetic counselor at 507-293-7299 to discuss before ordering testing:		
REQUIRED: Does this relative share any relevant clinical features or clinical history with the patient? ☐ No ☐ Yes If "Yes," describe:		

Patient Name *(Last, First, Middle)*	Birth Date *(mm-dd-yyyy)*

Provide information above or place label to the right.

Label Here

Ancestry

☐ African/African American
☐ Ashkenazi Jewish
☐ East Asian
☐ European
☐ Latinx
☐ Middle Eastern
☐ South Asian
☐ None of the above
☐ Choose not to disclose
☐ Unknown

History of Consanguinity

☐ No ☐ Yes; relationship details:

Suspected Diagnoses/Genes of Interest

List suspected diagnoses or specific genes that you would like considered for this evaluation.

Patient (Proband) Clinical Evaluations

Indicate the previous tests and evaluations performed for this proband, and provide details regarding the specific tests and pertinent results below. It will be assumed that any evaluations left blank were not performed or are unknown.

Karyotype	☐ Normal	☐ Abnormal:
Chromosomal Microarray	☐ Normal	☐ Abnormal:
Gene Sequencing/Panel/ Exome**	☐ Normal	☐ Abnormal:
Repeat Expansion	☐ Normal	☐ Abnormal:
Methylation/UPD**	☐ Normal	☐ Abnormal:
Mitochondrial DNA**	☐ Normal	☐ Abnormal:
Metabolic Work-up**	☐ Normal	☐ Abnormal:
Brain MRI	☐ Normal	☐ Abnormal:
Brain Spectroscopy	☐ Normal	☐ Abnormal:
Electroencephalogram (EEG)	☐ Normal	☐ Abnormal:
Echocardiogram	☐ Normal	☐ Abnormal:
Electrocardiogram (ECG/EKG)	☐ Normal	☐ Abnormal:
Skeletal Survey	☐ Normal	☐ Abnormal:
Renal Imaging	☐ Normal	☐ Abnormal:
Muscle Biopsy	☐ Normal	☐ Abnormal:
Electromyogram (EMG)	☐ Normal	☐ Abnormal:
Ophthalmology Exam	☐ Normal	☐ Abnormal:
Audiology Evaluation	☐ Normal	☐ Abnormal:

**Describe details of above evaluations or other evaluations not listed above:

Patient Name *(Last, First, Middle)*

Birth Date *(mm-dd-yyyy)*

Provide information above or place label to the right.

Label Here

Patient (Proband) Clinical Features

Check all that apply to the patient (proband) and provide additional descriptions, if available. This information is required to facilitate interpretation of results.

Perinatal History
- ☐ Intrauterine growth restriction
- ☐ Oligohydramnios
- ☐ Polyhydramnios
- ☐ Premature birth

Craniofacial
- ☐ Abnormality of the outer ear
- ☐ Cleft lip
- ☐ Cleft palate
- ☐ Craniosynostosis
- ☐ Facial dysmorphism; specify below
- ☐ Macrocephaly
- ☐ Microcephaly

Growth
- ☐ Failure to thrive
- ☐ Obesity
- ☐ Overgrowth
- ☐ Short stature
- ☐ Tall stature

Developmental/Cognitive
- ☐ Absent speech
- ☐ Cognitive decline
- ☐ Developmental regression
- ☐ Global developmental delay
- ☐ Intellectual disability
- ☐ Motor delay
- ☐ Speech delay

Behavioral/Psychiatric
- ☐ Attention-deficit/Hyperactivity disorder
- ☐ Autism spectrum disorder
- ☐ Behavioral abnormality; specify below
- ☐ Obsessive-compulsive disorder
- ☐ Sleep disturbance

Neuromuscular
- ☐ Abnormality of brain morphology; specify below
- ☐ Ataxia
- ☐ Cerebral palsy
- ☐ Dystonia
- ☐ Encephalopathy
- ☐ Gait abnormality
- ☐ Hypotonia
- ☐ Muscle weakness
- ☐ Peripheral neuropathy
- ☐ Seizures
- ☐ Spasticity
- ☐ Tremor

Musculoskeletal
- ☐ Arthralgia
- ☐ Contractures
- ☐ Elevated creatine kinase
- ☐ Joint hypermobility
- ☐ Joint laxity
- ☐ Pes planus
- ☐ Scoliosis
- ☐ Skeletal dysplasia
- ☐ Talipes
- ☐ Vertebral anomaly

Hearing
- ☐ Conductive hearing loss
- ☐ Mixed hearing loss
- ☐ Sensorineural hearing loss

Ophthalmologic
- ☐ Esotropia
- ☐ High myopia
- ☐ Nystagmus
- ☐ Ptosis
- ☐ Strabismus

Cardiovascular
- ☐ Aortic dilatation/dissection
- ☐ Arrhythmia
- ☐ Atrial septal defect
- ☐ Cardiomyopathy
- ☐ Patent ductus arteriosus
- ☐ Patent foramen ovale
- ☐ Ventricular septal defect

Gastrointestinal
- ☐ Abnormal GI motility; specify below
- ☐ Abnormality of the liver; specify below
- ☐ Dysphagia
- ☐ Feeding difficulties
- ☐ Gastrointestinal inflammation
- ☐ Nausea and vomiting
- ☐ Splenomegaly

Genitourinary
- ☐ Abnormal external genitalia
- ☐ Cliteromegaly
- ☐ Cryptorchidism
- ☐ Hydronephrosis
- ☐ Renal malformation

Skin/Hair/Dental
- ☐ Abnormal hair; specify below
- ☐ Abnormal skin; specify below
- ☐ Café-au-lait spot; specify below
- ☐ Dental abnormalities; specify below
- ☐ Hemangioma
- ☐ Hyperpigmentation

Endocrine
- ☐ Adrenal abnormality
- ☐ Hypothyroidism
- ☐ Pituitary gland abnormality
- ☐ Thyroid gland abnormality

Hematologic/Immunologic
- ☐ Anemia
- ☐ Bruising susceptibility
- ☐ Immunodeficiency
- ☐ Recurrent infections

Cancer/Neoplastic
- ☐ Specify age of onset and tumor type:

Additional Details/Clinical History

Patient Name *(Last, First, Middle)*	Birth Date *(mm-dd-yyyy)*

Provide information above or place label to the right.

Label Here

MAYO CLINIC
LABORATORIES

Whole Genome Sequencing: Informed Consent

This form is provided to ensure that you are informed about genetic testing. Genetic testing can be complex. Genetic counseling is recommended to help you more fully understand the risks and benefits associated with this test. It is your choice whether or not to have this test.

What is Whole Genome Sequencing?

- Whole genome sequencing is a test that detects changes (variants) in a patient's genetic code (DNA). Humans have approximately 20,000 genes. Whole genome sequencing evaluates these genes for variants that may provide or confirm a specific diagnosis for a patient.
- Another test, whole exome sequencing, can be used to detect variants in a patient's DNA. While whole exome sequencing focuses on the coding regions of genes (exons) that contain the majority of disease-causing variants, whole genome sequencing can detect variants in coding and non-coding regions. Additionally, whole genome sequencing can detect variant types including, but not limited to, mitochondrial genome variants and repeat expansions that are not identifiable by whole exome sequencing.

How is this test performed?

- A blood draw or other procedure will be required to obtain samples from all individuals undergoing testing. DNA is obtained from the samples and sequenced to identify genetic variants.
- The laboratory evaluates certain characteristics of each variant (such as the type of genetic change, whether family members have this change, and how common it is in the general population) in order to determine whether it could cause a genetic disorder in a patient.

What are the potential benefits of this test?

- Genetic variants may be detected that explain a patient's clinical features and provide a diagnosis.
- Establishing a diagnosis may allow for a better prediction of the outcome or course of a disorder. It may also help to determine the best medical management for a patient, such as surveillance, treatment, or preventive measures.
- Identification of a diagnosis may also allow for a more accurate risk estimate and/or testing of at-risk or affected family members.

What are the potential risks of this test?

- If a disease-causing variant is found and a specific diagnosis is made, it may not change the medical management that was previously recommended. There also may not be a treatment available for the disorder.
- In some cases, a health care provider may recommend additional tests to better understand the results from whole genome sequencing.
- Other possible risks, such as those associated with financial/insurance considerations, psychological effects, and implications for family members should be discussed with your health care provider.

What are the limitations of this test?

- This test will not establish a diagnosis for all patients.
- Due to technical limitations, variants may exist in regions of the genome that cannot be analyzed.
- Certain types of variants may not be detected by this test.
- Scientific understanding of the role of genes and variants in human diseases is not complete. Therefore, the significance of some variants that are found may not be known. Patients are encouraged to contact their health care provider for updates regarding their test results, as understanding may change with time.
- The laboratory's interpretation is based upon the accuracy of the clinical information and family history provided by the ordering health care provider. If pertinent information is not provided, this may affect whether certain variants are reported.

What types of test results will the laboratory report?

- *Variants associated with the patient's clinical features:* Variants known to cause conditions that have features which overlap with the patient's clinical features will be reported (including carrier status for recessive conditions). Variants will be reported if they are known or expected to cause the genetic condition (pathogenic or likely pathogenic). Variants of uncertain significance will also be reported.
- *Variants involving genes of uncertain significance:* Variants may be found involving genes that are suspected, but not certain, to play a role in human disease. Variants involving these genes of uncertain clinical significance may be reported if there is suspicion that they are related to a patient's clinical features.

Patient Name *(Last, First, Middle)*	Birth Date *(mm-dd-yyyy)*

Provide information above or place label to the right.

Label Here

Will secondary findings be reported?

- Patients are evaluated for medically actionable secondary findings, and these findings are reported in accordance with the American College of Medical Genetics (ACMG) recommendations (Miller et al., 2021). **Individuals can choose to not receive secondary findings by opting out on the following page.** If an individual opts out of secondary findings, variants in these genes will not be evaluated or reported unless they overlap with the reported clinical features. Note that if the proband opts out, secondary findings will not be reported for any family member.
- Rarely, findings may implicate another predisposition or presence of active disease. These findings will be carefully reviewed to determine whether or not they will be reported. Multigenic CNVs that are reported in association with the patient's clinical features could include a gene associated with secondary findings.
- Knowledge of a person's risk for these conditions can help to determine the medical actions available to maintain that person's health, such as screening for cancer or specific heart conditions.
- These results may lead to increased anxiety or worry. They may also result in additional medical interventions.

Why it is recommended that family members should be tested and what types of test results will they receive?

- Interpretation of genetic variants is more accurate when the laboratory is able to compare the results between the patient and their family members.
- Based on published reports, the chance of finding a diagnosis is highest when samples are submitted from both biological parents. However, the patient alone or in combination with other family members can be submitted.
- Family members will not receive their own full test results. However, if the patient's reported genetic variants are identified in another family member, this will be indicated in the patient's report. Family members may learn about a diagnosis of a genetic condition, increased risk for health concerns, or carrier status for a recessive condition.
- Variants present in family members that are absent from the patient will not be reported.

What else could the test results reveal about family members?

- It is possible to uncover that a parent or other family member is unrelated to the patient, or that relationships are not as described due to mis-attributed paternity, maternity, or adoption. In this situation, the ordering provider will be notified and options will be discussed.
- In some cases, results may suggest that the parents of a patient are biologically related, such as first cousins or another familial relationship.

What types of test results will the laboratory not report?

- Variants that are benign (not disease causing) or likely benign will not be reported.
- Variants in genes associated with conditions that are not related to a patient's reported clinical features will not be reported, with the possible exception of the secondary findings described above.

What does a negative report mean?

- A negative report means that no variants were reported and an explanation for the patient's clinical features was not identified. However, because of the testing limitations noted above, there may still be a genetic explanation for a patient's features that was not identified by this test.

How will the test results become available?

- The laboratory will release a patient's test report directly to the ordering health care provider and it will become part of the patient's medical record.
- Requests for the raw data should be directed to the laboratory. A separate fee may apply. The laboratory is not responsible for providing software or other tools needed to visualize, filter, or interpret this data.

Will my test results be shared with databases or researchers?

- Mayo Clinic is an active participant in the National Institutes of Health-funded Clinical Genome Resource (ClinGen) and shares information about genetic variants identified through clinical genetic testing with publicly available databases, such as ClinVar and Matchmaker Exchange.
- No patient-identifying information (ie, name, birth date) is shared.
- Genomic data sharing enables health care providers, clinical laboratories, and researchers to share experiences. This can lead to improved interpretations of genetic test results.

What will happen to my DNA after testing is complete?

- The laboratory does not guarantee indefinite storage of patient samples and may discard them within 60 days of test completion, in accordance with state-specific regulations.
- Any sample remaining after testing is complete may be used for internal laboratory quality control or research purposes, after the removal of patient identifiers such as name and birth date. **You may request that your DNA sample not be used for these purposes by indicating this preference on the next page.**
- At this time, it is not standard practice for the laboratory to systematically re-review patient results or previous variant classifications. However, due to broadening genetic knowledge, it is possible that the laboratory may discover new information of relevance to the patient. Should that occur, the laboratory will recontact the healthcare provider to discuss the new findings or classification of previously reported variants; the laboratory may issue an amended report.

Patient Name *(Last, First, Middle)*	Birth Date *(mm-dd-yyyy)*	Label Here

Provide information above or place label to the right.

Informed Consent Signature Page

Instructions: Informed consent is required for New York clients. My signature below acknowledges my voluntary participation in this test. I understand that the genetic analysis performed by Mayo Clinic Laboratories in no way guarantees my health, the health of an unborn child, or the health of other family members.

Secondary findings:

- **Patient (proband):** Checking the "Opt out of secondary findings" box below means that the laboratory will not look for variants in the list of secondary findings genes published by ACMG and will not report them unless the variant is in a gene related to the patient's clinical features. If the patient (proband) opts out, secondary findings will not be reported for any family member comparators. If the boxes are not checked or this page is not returned, opt in will be assumed.
- **Family member comparators:** Checking the "Opt out of secondary findings" box below means that if secondary findings are reported in the proband, the presence or absence of these variants in the family member will not be stated. If the boxes are not checked or this page is not returned, opt in will be assumed. Family members will not receive their own separate report; the presence of these variants in the family member will be stated on the proband's report.

Patient (Proband) Signature

My signature below acknowledges my voluntary participation in this test for myself or my child.

Patient/Guardian Signature ▶ Signature Required	Date *(mm-dd-yyyy)*	☐ Opt out of secondary findings
Parent/Guardian Printed Name *(Last, First, Middle)*	Guardian Relationship to Patient	

Family Member Signatures

Only fill out information for family members whose specimens are being sent as comparators.

Family Member 1 Signature ▶ Signature Required	Date *(mm-dd-yyyy)*	☐ Opt out of secondary findings
Family Member 1 Printed Name *(Last, First, Middle)*	Birth Date *(mm-dd-yyyy)*	
Family Member 2 Signature ▶ Signature Required	Date *(mm-dd-yyyy)*	☐ Opt out of secondary findings
Family Member 2 Printed Name *(Last, First, Middle)*	Birth Date *(mm-dd-yyyy)*	
Family Member 3 Signature ▶ Signature Required	Date *(mm-dd-yyyy)*	☐ Opt out of secondary findings
Family Member 3 Printed Name *(Last, First, Middle)*	Birth Date *(mm-dd-yyyy)*	

Provider/Genetic Counselor Signature

I have explained the above information to this individual. I have addressed the limitations outlined above and have answered all questions to the best of my ability.

Provider/Genetic Counselor Signature ▶ Signature Required	Date *(mm-dd-yyyy)*	Provider/Genetic Counselor Printed Name *(Last, First)*

DNA storage:

- **All clients residing outside of New York:** Checking the "Opt out of DNA storage" box below means that samples for the proband and any family member comparators will be destroyed upon completion of this test, and will not be used for research or quality assurance performed in the laboratory. Should reanalysis be requested in the future, new sample(s) will be required. If the box below is not checked or this page is not returned, opt in will be assumed.
  ☐ **Opt out of DNA storage**
- **New York clients:** Checking the "New York clients: permission to retain remaining sample(s)" box below means that permission is given to retain any remaining samples for the proband and any family member comparators longer than 60 days after the completion of testing, and can be used as de-identified samples for research or quality assurance performed in the laboratory. If the box is not checked, all samples from New York clients will be disposed of 60 days after testing is complete and will not be used for research or quality assurance purposes. Should reanalysis be requested in the future, new sample(s) will be required.
  ☐ **New York clients: permission to retain remaining sample(s)**

Print