MNG LABORATORIES
A LabCorp Company

MNG Exome Test Request Form

5424 Glenridge Drive NE | Atlanta, GA 30342 USA | phone: 844.664.8378 | fax: 678.225.0212 | mnglabs.labcorp.com

Proband and Specimen Information

Patient Last Name	Patient First Name	☐ Male ☐ Female
Patient ID #	Date of Birth [MM/DD/YYYY]	**Specimen Type** ☐ Whole Blood ☐ Buccal Swab
Diagnosis/ICD-10	Collection Date [MM/DD/YYYY]	☐ DNA: _____ ☐ Tissue: _____

Referring Physician Information

Physician Name	NPI # *(Required)*
Facility/ Organization	Signature
Facility Address City, State, Zip Code ☐ Same as billing	
Report Delivery ☐ Fax ☐ Email	Phone

Billing Information

Self-Pay? ☐ Yes If yes, **MUST** include payer contact name & details below. Payment must be received in full prior to testing.

Facility	Contact Name	
Billing Address		
City, State, Zip Code		
Phone	Fax	Email

Results

Recipient Name	
Facility	Phone
☐ Fax	
☐ Email	
Recipient Name	
Facility	Phone
☐ Fax	
☐ Email	

MNG Exome Sequencing

- Includes detection of copy number variants (CNVs) and uniparental disomy
- **Testing will not begin until all samples to be analyzed are received**
- Consent forms and clinical information **must** be included

Select a test:

- ☐ Whole Exome - Trio Sequencing
- ☐ Whole Exome - Duo Sequencing
- ☐ Whole Exome - Proband Only Sequencing
- ☐ Whole Exome - Additional Comparator

Optional Add-On Testing

Mitochondrial DNA *(no cost)*

☐ mtDNA Sequencing + Deletion

The following assays are available as add-on tests. To add any of these to your order, please select from the following menu. List or contracted pricing will apply.

Repeat Expansions *(select one or more)*

- ☐ Huntington's disease (HTT)
- ☐ Friedreich's Ataxia (FRDA)
- ☐ Spinocerebellar Ataxia (SCAs)
- ☐ C9orf72
- ☐ Myotonic Dystrophy 1
- ☐ Myotonic Dystrophy 2

Family Member 1 Information

Last Name	First Name	☐ Male ☐ Female
Relationship to Proband	Date of Birth [MM/DD/YYYY]	**Specimen Type** ☐ Whole Blood ☐ Buccal Swab
Affected? *Include Clinical Info* ☐ Yes ☐ No ☐ Unsure	Collection Date [MM/DD/YYYY]	☐ DNA: _____ ☐ Tissue: _____

Family Member 2 Information

Last Name	First Name	☐ Male ☐ Female
Relationship to Proband	Date of Birth [MM/DD/YYYY]	**Specimen Type** ☐ Whole Blood ☐ Buccal Swab
Affected? *Include Clinical Info* ☐ Yes ☐ No ☐ Unsure	Collection Date [MM/DD/YYYY]	☐ DNA: _____ ☐ Tissue: _____

Additional Comparator Information

Last Name	First Name	☐ Male ☐ Female
Relationship to Proband	Date of Birth [MM/DD/YYYY]	**Specimen Type** ☐ Whole Blood ☐ Buccal Swab
Affected? *Include Clinical Info* ☐ Yes ☐ No ☐ Unsure	Collection Date [MM/DD/YYYY]	☐ DNA: _____ ☐ Tissue: _____

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Clinical Information Form

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Patient Name _____ DOB _____

Clinical (Check All That Apply)

Eye
- ☐ Retinitis Pigmentosa
- ☐ Optic Atrophy
- ☐ Other

Hearing
- ☐ Sensorineural
- ☐ Stickler
- ☐ Usher

Neuronal Migration
- ☐ Meckel
- ☐ Joubert
- ☐ Other

☐ Stroke

Cognitive/Neurobehavioral
☐ Intellectual Disability (ID) ☐ Syndromic ID ☐ Nonsyndromic ID ☐ Autism ☐ Dementia

Movement Disorders
☐ Ataxia ☐ Episodic Ataxia ☐ Dystonia ☐ Chorea/Athetosis ☐ Parkinson Disease ☐ L-Dopa Response

Epilepsy ☐ Myoclonic ☐ Other
☐ Absence ☐ Tonic Clonic
☐ Epileptic Encephalopathy

Spasticity
☐ Spastic Paraplegia ☐ Other
☐ Spastic Quadriplegia

Connective Tissue & Bone
☐ Ehlers Danlos ☐ Marfan ☐ Aneurysms
☐ Other

Neuromuscular
☐ Distal ☐ Proximal ☐ Muscle Atrophy ☐ Contractures
☐ Malignant Hyperthermia ☐ Arthrogryposis ☐ Rhabdomyolysis
☐ Periodic Paralysis ☐ Statin Use ☐ Myasthenia

Nerve/Anterior Horn Cell
☐ Neurofibromas ☐ Charcot-Marie-Tooth ☐ Sensory
☐ Autonomic ☐ Pain ☐ Motor ☐ Nerve Conduction
☐ Other

Cardiomyopathy
☐ Dilated ☐ Hypertrophic
☐ Noncompaction

Arrhythmias
☐ Ventricular Tachycardia ☐ Brugada
☐ Long or Short QT ☐ Conduction Defect

Congenital Heart Defects
☐ Heterotaxy
☐ Other

Endocrine
☐ Hypothyroidism ☐ Other
☐ Diabetes Mellitus

Imaging (Check All That Apply)

Brain MRI
- ☐ Leigh Disease
- ☐ Basal Ganglia Calcification
- ☐ Stroke
- ☐ Cerebellar Atrophy
- ☐ Abnormal Myelin (describe)

EEG (Describe Findings)

EMG/NVC (Describe Findings)

Laboratory

Metabolic (Describe Findings)

CPK Maximum _____
Minimum _____

Genetic (Describe Findings)
- ☐ Chromosomal Microarray
- ☐ Deletion/Insertion Testing
- ☐ Other (comment)

Family History

Ethnicity (please check)

☐ Caucasian ☐ Sephardic Jewish ☐ African American (or Black) ☐ Asian
☐ Hispanic ☐ Ashkenazi Jewish ☐ Native American (or American Indian) ☐ Other: _____

Affected Maternal Lineage	Affected Paternal Lineage	Siblings
Relationship to Proband	Relationship to Proband	Number (specify gender)
Symptoms	Symptoms	Healthy/Affected

Additional Comments

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Informed Consent
Whole Exome & Whole Genome Sequencing

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Notice to Health Care Practitioner:

This document is a consent form for clinical whole exome or whole genome sequencing. Currently, the laboratory will only accept whole exome and whole genome test requests after the patient/parent or legal guardian/next of kin has received genetic counseling from a Healthcare Provider with experience in counseling patients for such a test. Please be aware of any applicable state laws in regards to counseling needs related to the current condition, the possibilities of detecting unsuspected conditions as well as other issues related to health insurance, and possible effects on life insurance. Please explain this consent to the patient, or authorized representative/guardian, and obtain an informed consent. Please explain the list of potential incidental findings that may be reported to the patient.

Consent for WES and WGS Testing

All of the above has been explained to me, to my satisfaction, and my signature below attests to the same. I understand that this is a voluntary test, and I have had the opportunity to ask questions about alternative testing.

Whole Exome or Whole Genome Sequencing Participant:

Proband Name: _____

Proband DOB: _____ Date: _____

Signature: _____

(Parent/Guardian signature if person being tested is a minor)

Health Care Provider Obtaining Consent:

Print Name: _____

Signature: _____

NPI#: _____ Date: _____

I have provided genetic counseling and have explained the risks, benefits, and limitations of WES testing to the patient/parent/guardian.

Consent of family members submitting a sample for evaluation of patient's results:

I understand that I am submitting my sample to help evaluate the results obtained on the person being tested, and that results obtained from my sample will be used solely for this purpose. I will NOT be informed of any test results on my sample. If I request any test results, I will have to be tested separately.

Name of Family Member: _____

Signature: _____

Relationship to Proband: _____ Date: _____

Name of Family Member: _____

Signature: _____

Relationship to Proband: _____ Date: _____

Name of Family Member: _____

Signature: _____

Relationship to Proband: _____ Date: _____

Additional Consent

What are incidental findings?

During testing, disease causing variants can be identified that are not related to the patient's condition for which the testing was done. These are referred to as "Incidental Findings" and indicate the presence of previously undiagnosed, potentially serious conditions that can be prevented or treated if diagnosed. A list of such conditions based on the recommendation of the American College of Medical Genetics (ACMG) is provided on the ACMG website. Please state whether you want to be informed about incidental findings in relation to the conditions listed.

How long are WES and WGS results kept in the testing lab?

The laboratory may keep the identified WES/WGS raw data in the lab indefinitely. This helps us improve our diagnostic capabilities and will help others with similar conditions. To advance the understanding of genetic disorders, your results might be analyzed and published in scientific articles in a de-identified manner consistent with HIPAA guidelines.

What will happen to my DNA sample?

No additional tests will be performed on these samples, without specific, signed authorization by the individual(s). After 60 days, unless consent is given, the sample will be destroyed.

1. Incidental Findings Consent (initial)

_____ (Proband Only) I would like to learn of incidental findings to the conditions listed

_____ (Proband + Parents/Family Members) I would like to learn of incidental findings to the conditions listed

_____ I would **NOT** like to learn of incidental findings to the conditions listed

2. Raw Data Storage Consent (initial)

We agree that our WES/WGS data may be stored indefinitely

Proband _____
Mother _____
Father _____
Other _____

We agree that our WES/WGS data may be used for scientific publication in a de-identified manner

Proband _____
Mother _____
Father _____
Other _____

3. DNA Sample Retention Consent

Please keep my DNA used for WGS/WES for future testing should I desire such testing or if I want to participate in research in the future. I understand no additional genetic tests will be performed without my specific consent/ instructions, but my DNA may be used for quality control purposes. I understand that there is no guarantee of availability of my DNA after 60 days.

Proband _____ Mother _____ Father _____ Other _____

Informed Consent for Patients and/or Legal Guardians:

What is whole exome sequencing (WES) and whole genome sequencing (WGS)?

Whole exome sequencing (WES) and whole genome sequencing (WGS) are genetic tests. They are performed on DNA extracted from 3 ml of blood or other acceptable tissue type. Their purpose is to identify a heritable cause of a disorder. WES examines the "exome" or coding regions of DNA, the most important information containing segment of the genome, and will often identify the cause of disease. WGS examines all of the DNA in the human genetic code including coding and non-coding regions.

The sensitivity of both tests is improved if blood is submitted from biological parents or siblings of the patient. The goal of both tests is to identify the genetic cause of the disease for which the patient is presenting. For this reason, it is crucial that a detailed description of the clinical symptoms of the patient and other affected family members is provided. Results will only be reported on the patient. Because WES and WGS results have potential consequences for the patient's family, we recommend that the consenting and ordering process be performed with the assistance of a genetic counselor and/or the ordering physician.

Continued >>

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What kind of results are reported?

1. Positive: Variant(s) have been identified that are known to cause the disease symptoms based on the available scientific evidence at the time of testing.

2. Indeterminate: Variant(s) have been identified that are likely to cause the disease symptoms based on the available scientific evidence at the time of testing, but there is a lack of definitive scientific evidence available to prove it.

3. Negative: No variant has been identified that is known or likely to cause the disease symptoms based on the available scientific evidence at the time of testing.

What implications do positive and negative results have?

When WES/WGS detects known disease causing variants, the test result is highly accurate. A positive result will help your clinician to better predict the course of the condition and provide you with treatment options, if they exist. The results will also help determine the risk of recurrence of the condition in other children. An indeterminate result will point to a probable cause of a condition, but you may wish to consult a genetic counselor or your physician and undergo further independent testing to confirm or rule out the proposed role. A negative result does not indicate the absence of a genetic cause and will not change the clinical diagnosis.

Are there limitations to WES and WGS testing?

WES and WGS are screening tests. There is a possibility a genetic variant caused a condition that is not identified by the WES/WGS tests either because of the technical limitations of the assays, or because of incomplete understanding of the significance of variants detected. Although WES/WGS testing is highly accurate, the interpretation of the report is based on current medical knowledge, which is not complete.

1. WES may not be able to detect genetic disorders that are caused by expansion of repetitive regions of the genome. One example is Fragile X Syndrome. If one of these types of conditions is suspected, your physician should order the appropriate test.

2. Not all regions in the human genome can be sequenced due to limitations in technology, so some variants in such regions might go undetected with WGS methodologies.

Are there results that will not be reported?

1. Samples from the patient's relatives may be used to help diagnose the patient's condition, but results for these relatives will not be reported independently. They will only be referred to in the report for the patient if they are directly relevant to the patient's condition. However, the patient's genetic results may have implications for their relatives, and it is important that these implications are discussed with a genetic counselor.

2. Variations in genes that affect susceptibility to a condition, but do not cause the person to develop the condition, will not be reported.

3. Carrier status for recessive disorders: Most people carry variants that are not disease causing but could become disease causing if that person had children with someone who was healthy but had the same variant. This is referred to as being a "carrier" for a disease. This test is not intended for determining carrier status. If you are concerned about carrier status for conditions that might run in your family, you should get tested separately for carrier status. You should discuss these implications with your genetic counselor.

- Single heterozygous pathogenic/likely pathogenic variants in genes associated with recessive disorders that have potential overlap with a patient's clinical presentation (as provided to our laboratory) will be reported since we cannot definitively exclude that an undetected second variant in trans may be present.
- Single heterozygous variants of uncertain significance in genes associated with recessive disorders that have potential overlap with a patient's clinical presentation (as provided to our laboratory) will be reported at the discretion of the laboratory director.
- Single heterozygous variants in genes associated with recessive disorders that do not have overlap with the patient's clinical presentation (as provided to our laboratory) will not be reported.

Who will have access to the results?

Test results are maintained electronically by the laboratory. The results are provided to the ordering physician and/or healthcare facility that ordered the test. Results may also be made available to individuals/organizations with a legal right of access under applicable Federal and/or State law, or as authorized by the patient or the patient's representative. Patient privacy is of utmost concern to us, and as a CAP certified facility we are prepared to safeguard Protected Health Information.

What are the risks of testing?

1. Non-paternity (when the reported father of the child is not the biological father) or half sibling-ships (when siblings do not share the same father AND mother) would be detected. We do not report these findings unless they have direct clinical significance.

2. Genetic non-discrimination law prevents insurance companies from using your genetic information to deny health insurance coverage, but the law does not cover life insurance, disability insurance or long-term care insurance. The detection of an incidental condition may affect your future ability to buy these forms of insurance or get the best insurance rates. Please be aware of any applicable State laws and applicable terms of any active insurance policies in regards to consent and the release of these results to insurance companies.

3. WES/WGS may identify serious and/or untreatable genetic conditions. It can result in unexpected psychological trauma, both for you and your family. The detection of such a condition or conditions could also affect the health or healthcare needs of your siblings, children, or other close relatives.

4. Although WES/WGS is highly accurate, the interpretation of the report is based on current medical knowledge, which is not complete. We do not report out changes in interpretation of variants automatically, but we do have mechanisms to issue an updated report if requested by the patient's physician.