MNG LABORATORIES
A LabCorp Company

# MNG Healthy Exome Test Request Form

5424 Glenridge Drive NE | Atlanta, GA 30342 USA | phone: 844.644.8378 | fax: 678.225.0212 | mnglabs.com

## MNG Healthy Exome Sequencing (WES007)

**Note: Clinical Information and Consent Form are required for MNG Healthy Exome Sequencing**

Includes Copy Number Analysis

## Patient and Specimen Information

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

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

***Please complete and include a clinical information form, or attach clinical notes***

## Referring Physician Information

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

## Billing Information *(REQUIRED)*

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

| | |
|---|---|
| Authorized Recipient Name | Authorized Recipient Name |
| Facility Phone | Facility Phone |
| ☐ Fax | ☐ Fax |
| ☐ Email | ☐ Email |

**All of the following are required before we will process Healthy Whole Exome Sequencing orders (please check the following):**

☐ Healthy Whole Exome Sequencing Consent form completed and signed
☐ Clinical Information form completed

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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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# Healthy Whole Exome Sequencing Consent Form

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Patient Name _____ DOB _____

## Notice to Health Care Practitioner:

This document is a consent form for Healthy Exome Sequencing (HWES). Currently, the laboratory will only accept test requests after the individual to be tested has received genetic counseling from a Healthcare Provider with experience in counseling patients. 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. Please be aware of any applicable State laws and applicable terms of any active insurance policies in regard to consent and the release of these results to insurance companies. Please explain the information below to the individual to be tested and obtain an informed consent/signature so the testing can be performed.

## Informed Consent for Patients:

### *What is Healthy Whole Exome sequencing (HWES)?*

Healthy Whole Exome sequencing (HWES) is a genetic test. It is performed on DNA extracted from 3ml of blood or other acceptable tissue type. Its purpose is to identify carrier status for severe inherited disorders in healthy individuals. HWES is performed to detect known pathogenic and predicted pathogenic variants in genes that are causative of severe autosomal dominant and recessive disorders. Rather than focusing solely on known pathogenic variants common in well characterized populations, the MNG Healthy Exome test provides a wider range of coverage for those seeking a more thorough review versus standard screening tests. This test is intended for individuals who are healthy, but have interest in becoming aware of pathogenic or predicted pathogenic variants that can affect future offspring or themselves in a later stage of their life.

Because HWES results have potential consequences for the individual and the individual's potential offspring, we require that ordering of the testing be performed by a licensed medical professional.

### *What kind of results are reported?*

1. Positive: Variant(s) have been identified that are known pathogenic or likely pathogenic, based on well-established scientific evidence or criteria defined by the American College of Medical Genetics (ACMG), and may cause a severe medical condition either in potential offspring or the individual tested at a later time in life.
2. Negative: An absence of pathogenic or likely pathogenic variants that are known or predicted to cause a severe medical condition in offspring or in the individual tested at a later time in life.

### *What implications do positive and negative results have?*

When HWES detects known disease causing variants, the test result is highly accurate. A positive result will help your clinician to better predict the risk of having a severely disruptive disorder later in life, or having a severely affected child. A negative result does not guarantee that you nor your child will not have any inherited health problems in the future, since the cause of many inherited genetic conditions are not well understood at this point of time.

### *Are there limitations to HWES testing?*

HWES is a screening test. A genetic variant that can cause a severe health condition may not be identified as such by the test either because of technical limitations, or incomplete understanding of the significance of variants detected.

1. HWES is not currently guaranteed to detect intermediate-scale alterations in the DNA content. These include the loss or duplication of regions extending less than 10 exons.
2. HWES may not be able to detect genetic disorders that are caused by expansion of repetitive regions of the genome, such as Fragile X Syndrome. If one of these types of conditions is suspected, your physician should order the appropriate diagnostic test.

### *Are there results that will not be reported?*

Variations in genes that affect susceptibility to a condition, but do not cause the person to develop the condition, will not be reported. Variants of Uncertain Significance and Benign/Likely Benign variants are also not 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 health care 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 we adhere to HIPAA privacy and security requirements.

### *How long are HWES results kept in the testing lab?*

The laboratory may keep the identified HWES raw data in the lab indefinitely. This helps us improve our diagnostic capabilities. 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.

_____ (initials) I agree that my HWES data may be stored indefinitely.

_____ (initials) I agree that my HWES data may be used for scientific publication in a de-identified manner.

**NEXT>>>**

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Patient Name _____ DOB _____

### *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 below, the sample will be destroyed.

_____ (initials) Please keep my DNA used for HWES testing indefinitely should I desire additional 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.

### *What are the risks of testing?*

1. 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. 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.
2. HWES may identify carrier status for 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 health care needs of your siblings, children, or other close relatives
3. Although HWES is highly accurate, the interpretation of the report is based on current medical knowledge, which may change as better understanding of genetic conditions is obtained.

## Consent for HWES 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.

**HWES Sequencing Individual**

Print Name: _____ Date: _____

Signature: _____ Date of Birth: _____

I authorize my healthcare provider and insurance company to have access to my results. I understand that these parties may only use this information in accordance with applicable law.

I also authorize the following party/parties to have access to my results (optional):

Print Name: _____ Print Name: _____

Institution: _____ Institution: _____

Fax Number / Email: _____ Fax Number / Email: _____

I have provided genetic counseling and have explained the risks, benefits, and limitations of HWES testing to the patient.

**Health Care Provider Obtaining Consent:**

Print Name: _____ Date: _____

Signature: _____ NPI#: _____