MNG LABORATORIES
A LabCorp Company

Carrier Exome Test Request Form

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

(WES006) MNG Carrier Exome (for couples only)

Note: One sample from both individuals (2) is required for WES006. Carrier testing is not for diagnostic purposes.

Individual 1

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: _____

Individual 2

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: _____

Referring Physician Information

Physician Name	NPI # or equivalent *(Required)*
Facility / Organization	Signature
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 MNG Carrier Exome orders (please check)

☐ Carrier Whole Exome Sequencing Informed Consent signed by both individuals (2)
☐ One sample from both individuals (2)

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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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Patient Name _____ DOB _____

Notice to Health Care Practitioner:

This document is a consent form for Carrier Whole Exome Sequencing (CWES). Currently, the laboratory will only accept carrier whole exome test requests after the individual(s) have 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 testing, 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 individual(s) and obtain an informed consent.

Informed Consent for Patients:

What is carrier whole exome sequencing (CWES)?

CWES is a genetic test. It is performed on DNA extracted from 3 ml of blood or other acceptable tissue type. Its purpose is to identify carrier status in healthy individuals. CWES is performed on both partners in order to detect shared known pathogenic and predicted pathogenic variants in genes that are causative of autosomal recessive disorders. Rather than focusing solely on known pathogenic variants that are common in well characterized populations, the MNG Carrier Exome Screen provides a wider range of coverage for those seeking a more thorough review versus standard carrier screening tests. This test is intended for prospective parents who are interested in a more comprehensive review of their genetic background to better understand their risk of having children with rare autosomal recessive disorders. It is not recommended for diagnostic purposes.

A single report will be generated for the individuals tested highlighting the variants carried by both, which could result in offspring with severe health problems. Because CWES results have potential consequences for the individual's family, we recommend that the consenting and ordering process be performed with the assistance of a genetic counselor and/or the ordering physician.

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 (ACMG), and cause a severe medical condition in the offspring in the homozygous or compound heterozygous form.
2. Negative: An absence of pathogenic or likely pathogenic variants that are known or likely to cause a severe medical condition in the offspring if in the homozygous or compound heterozygous form.

What implications do positive and negative results have?

When CWES 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 an affected child. A negative result does not indicate the absence of carrier status for a recessive disorder, but greatly reduces the risk of having a child affected with health problems.

Are there limitations to CWES testing?

CWES is a screening test. There is a possibility of a genetic variant being shared by both individuals that can cause a condition not identified by the CWES test either because of the technical limitations of the assay or because of incomplete understanding of the significance of variants detected.

1. CWES is not currently validated to detect intermediate-scale alterations in the DNA content. These include the loss or duplication of less than 10 exons.
2. CWES 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 test.

Are there results that will not be reported?

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

2. Dominantly inherited disease causing variants will not be reported for either individual. If a dominant variant is suspected in a family, a TRIO should be submitted for Whole Exome Sequencing, which is a diagnostic test.

3. Carrier status is not reported in one partner unless a pathogenic or likely pathogenic variant is shared by both individuals. The exception to this is x-linked recessive variants found in the female.

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.

NEXT>>>

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Patient Name _____ DOB _____

How long are CWES results kept in the testing lab?

The laboratory may keep the identified CWES 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) We agree that our CWES data may be stored indefinitely.

_____ _____ (initials) We agree that our CWES data may be used for scientific publication in a de-identified manner.

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 our DNA used for CWES testing for future testing should we desire such testing, or if we want to participate in research in the future. We understand no additional genetic tests will be performed without our specific consent/ instructions, but our DNA may be used for quality control purposes. We understand that there is no guarantee of availability of our 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. CWES 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 CWES is highly accurate, the interpretation of the report is based on current medical knowledge.

Consent for CWES 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.

Carrier Whole Exome Sequencing Individual 1
Print Name: _____ Date: _____
Signature: _____ Date of Birth: _____

Carrier Whole Exome Sequencing Individual 2
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.

Carrier Whole Exome Sequencing Individual 1
Print Name: _____ Date: _____
Signature: _____ Date of Birth: _____

Carrier Whole Exome Sequencing Individual 2
Print Name: _____ Date: _____
Signature: _____ Date of Birth: _____

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

Health Care Provider Obtaining Consent:

Print Name: _____ Date: _____
Signature: _____ NPI#: _____