



# Neuromuscular Disorders Exome Requisition Form

## The University of Chicago Genetic Services Laboratories

5841 South Maryland Avenue, Room G701/MC0077, Chicago, IL 60637

Toll Free: 888.824.3637 | Local: 773.834.0555 | Fax: 773.702.9130

[ucgslabs@genetics.uchicago.edu](mailto:ucgslabs@genetics.uchicago.edu) | [dnatesting.uchicago.edu](http://dnatesting.uchicago.edu) | CLIA#: 14D0917593 | CAP#: 18827-49

### Patient Information

Name: Last \_\_\_\_\_ First \_\_\_\_\_ Date of Birth: \_\_\_\_\_

Gender:  Male  Female MRN: \_\_\_\_\_

Ethnicity:  Caucasian  African-American  Hispanic  Asian  Ashkenazi Jewish  Other \_\_\_\_\_

### Ordering Physician Information

REPORTING RESULTS: Reports will only be faxed out. Please check the boxes below for those who should receive by fax.

Referring Physician: \_\_\_\_\_  Genetic Counselor: \_\_\_\_\_

Phone: \_\_\_\_\_ Fax: \_\_\_\_\_ Phone: \_\_\_\_\_ Fax: \_\_\_\_\_

Email: \_\_\_\_\_ Email: \_\_\_\_\_

Referring Lab: \_\_\_\_\_

Phone: \_\_\_\_\_ Fax: \_\_\_\_\_

Email: \_\_\_\_\_

### Indication for Testing

REQUIRED INFORMATION. NECESSARY FOR TESTING

Symptomatic: \_\_\_\_\_ ICD-10: \_\_\_\_\_

Results of previous genetic testing (including information on whether patient has been screened for SCA trinucleotide repeats): \_\_\_\_\_

Testing for known mutation/variant\*: Gene Name: \_\_\_\_\_ Mutation/Variant: \_\_\_\_\_

Symptomatic  Asymptomatic Name of Proband/UofC Lab Number: \_\_\_\_\_ Relationship to Proband: \_\_\_\_\_

In addition, the following are strongly recommended to be sent with samples for the Neuromuscular Disorders Exome: detailed clinic notes, pedigree, results of prior genetic testing, and muscle biopsy reports.

### Sample Information

Date Sample Drawn: \_\_\_\_\_

Specimen Type:  Peripheral Blood  Saliva  DNA  Other: \_\_\_\_\_

Specimen Requirements: 3-10cc blood in an EDTA (purple top) tube, T-25 of cultured cells. DNA samples are only accepted if the DNA extraction or isolation was performed at a CLIA-certified laboratory. All samples should be shipped via overnight delivery at room temperature to the address at the top of this page. No weekend or holiday deliveries. Label each specimen with the patient's name, date of birth and date sample collected. Please see our website for other specimen requirements.

### Ordering Checklist

- Test Requisition Form (required)
- Completed Indication for Testing/ICD-10 study code (required)
- Completed Clinical Checklist (required)
- Completed Billing Information (required)
- Completed Neuromuscular Disorders Consent Form (required)
- Completed Research Consent Form (recommended)

### For Office Use Only

# NEUROMUSCULAR DISORDERS CLINICAL CHECKLIST

## REQUIRED

Please check all clinical features that apply, and use the additional space provided at the bottom of the form if needed

### Neuromuscular disorder(s) present?

- YES (note age of onset below)  NO  
 UNKNOWN

Age of onset: \_\_\_\_\_

### Progressive neuromuscular disorder?

- YES  NO  UNKNOWN

### Type of neuromuscular disorder

- Myopathy  
 With prominent contractures

Details: \_\_\_\_\_

- Muscular dystrophy

Details: \_\_\_\_\_

- Congenital myasthenic syndrome

Details: \_\_\_\_\_

- Peripheral neuropathy

Details: \_\_\_\_\_

- Other: \_\_\_\_\_

### Muscle biopsy performed?

- YES (see below)  NO  UNKNOWN

Please specify results and attach copy of report:

\_\_\_\_\_

### Mitochondrial abnormality present?

- YES (see below)  NO  UNKNOWN

Please specify mitochondrial testing/results and attach copy of report: \_\_\_\_\_

\_\_\_\_\_

### Electromyography/ Nerve Conduction Velocity/ (EMG/NCV) performed?

- YES (see below)  NO  UNKNOWN

### Results of EMG/NCV testing:

#### EMG

- No abnormalities detected  
 Abnormal – please specify:

\_\_\_\_\_

#### NCV

- No abnormalities detected  
 Abnormal – please specify:

\_\_\_\_\_

### Family history of neuromuscular disorder or other relevant findings?

- YES (see below)  NO  UNKNOWN

Please specify: \_\_\_\_\_

\_\_\_\_\_

### Other neurological findings present?

- YES (see below)  NO  UNKNOWN

- Ataxia

- Dementia

- Dysarthria

- Hyperreflexia

- Hypotonia

- Seizures

- Spasticity

- Other: \_\_\_\_\_

### Cognitive/developmental delays or behavioral issues present?

- YES (see below)  NO  UNKNOWN

- Cognitive impairment

- Developmental regression

- Global developmental delay

- Mild  Moderate  Severe

- Autism spectrum disorder

- Behavioral / Psychiatric abnormality - please specify: \_\_\_\_\_

- Other: \_\_\_\_\_

### Brain MRI performed?

- YES (see below)  NO  UNKNOWN

### Results of brain MRI:

- No abnormalities identified

- Cerebellar hypoplasia

- Migration defect (please specify)

\_\_\_\_\_

- Lactate peak

- Other: \_\_\_\_\_

### Craniofacial findings present?

- YES (see below)  NO  UNKNOWN

- Dysmorphic facies

Please specify: \_\_\_\_\_

\_\_\_\_\_

- Macrocephaly, HC: \_\_\_\_\_

- Microcephaly, HC: \_\_\_\_\_

- Other: \_\_\_\_\_

### Cutaneous findings present?

- YES (see below)  NO  UNKNOWN

- Please specify: \_\_\_\_\_

\_\_\_\_\_

### Biochemical testing performed?

- YES (see below)  NO  UNKNOWN

### Results of biochemical testing:

- Elevated lactate: \_\_\_\_\_

- Elevated CK: \_\_\_\_\_

- Normal results for the following tests:

\_\_\_\_\_

- Other: \_\_\_\_\_

### Cardiac findings present?

- YES (see below)  NO  UNKNOWN

- Cardiomyopathy

- Hypertrophic cardiomyopathy

- Dilated cardiomyopathy

- Other – please specify:

\_\_\_\_\_

### Gastrointestinal (GI) issues present?

- YES (see below)  NO  UNKNOWN

Please specify: \_\_\_\_\_

### Growth abnormality present?

- YES (see below)  NO  UNKNOWN

Please specify: \_\_\_\_\_

### Skeletal findings present?

- YES (see below)  NO  UNKNOWN

Please specify: \_\_\_\_\_

### Neoplasms or immunodeficiency?

- YES (see below)  NO  UNKNOWN

Please specify: \_\_\_\_\_

### Hearing/Vision

- YES (see below)  NO  UNKNOWN

- Vision abnormality – please specify

\_\_\_\_\_

- Hearing impairment – please specify

\_\_\_\_\_

- Other: \_\_\_\_\_

Please include any additional relevant clinical information here:

In addition to this checklist, we strongly recommend sending the following documentation with the sample: detailed clinic notes, pedigree, results of prior genetic and metabolic testing.

## BILLING OPTIONS

All samples received with incomplete billing information will delay processing time.  
 Test cancelled while "in progress" will be billed for the amount of work completed up to that point.  
 Please note, we do not currently offer direct insurance billing for our Ataxia Exome.  
 Please forward all billing questions to: [youtlaw@bsd.uchicago.edu](mailto:youtlaw@bsd.uchicago.edu) or call (773-834-8220).

Patient Name: Last \_\_\_\_\_ First \_\_\_\_\_ (MI): \_\_\_\_\_ Date of Birth: \_\_\_\_\_

### 1.) Institutional Billing *(Pre-payment is required for all samples referred from outside the US or Canada.)*

Billing Institution: \_\_\_\_\_ PO#: \_\_\_\_\_

Financial Contact: \_\_\_\_\_ Phone: \_\_\_\_\_ Fax: \_\_\_\_\_

Address: \_\_\_\_\_ City: \_\_\_\_\_ State: \_\_\_\_\_ Zip: \_\_\_\_\_

Email (required): \_\_\_\_\_

### 2.) Self-Pay

We accept all major credit cards. Please call our office (773-834-8220) for credit card processing.



**Important notice:** We will not be responsible for refunding any "cost differential" that may occur as a result of a patient seeking any type of reimbursement.

Wire Transfer *(Please include 'Genetics Services Laboratories' and invoice numbers to ensure proper receipt.)*

**Electronic funding information, as follows:** The Northern Trust Bank – (Physical Address) 50 S. LaSalle Street, Chicago, IL 60675  
 ABA/Routing No.: 071000152, International SWIFT Code: CNORUS44, University of Chicago Wire Account No.: 28509

Amount \$ \_\_\_\_\_ (USD) Date of Transfer: \_\_\_\_\_ Name of Institution: \_\_\_\_\_

Check/Money Order **(Make check/money order payable to: The University of Chicago Genetic Services)** Amount Enclosed \$ \_\_\_\_\_  
 (Please note: All bank fees for returned checks will be added to the original charge of patient invoice)

We currently only offer institutional billing and self-pay for our Ataxia Exome. Insurance prior authorization is not absolutely mandatory before sending a sample to our laboratory. Insurance prior authorization services are offered as a courtesy and can be requested PRIOR to sending a sample to our laboratory (please see website for prior authorization request form). Samples received with appropriate billing information (institutional billing or self-pay) will be processed accordingly

## NEUROMUSCULAR DISORDER EXOME CONSENT FORM

### University of Chicago Genetic Services Laboratories (UCGSL)

**REQUIRED FOR EXOME TESTS**

**Patient Name:** \_\_\_\_\_

**Date of Birth:** \_\_\_\_\_

### Overview

Exome sequencing attempts to evaluate the coding regions of approximately 20,000 genes in the genome. This is called the 'exome'. The exome represents only 1.5% of the genome and comprises the majority of DNA variations that cause human disease. The UCGSL Neuromuscular Disorders Exome limits analysis of the exome sequencing data to a predefined set of 422 genes that have been associated with neuromuscular disease. For cases without a clearly pathogenic variant identified in the pre-defined list of genes, an additional analysis of previously reported pathogenic variants and truncating variants in known disease genes (present in the HGMD database) will be performed. The purpose of this test is to identify the underlying molecular basis of the specific neuromuscular disorder in the patient's family.

### Accuracy

The analysis performed is specific to genes associated with neuromuscular disorders. Accurate interpretation of test results requires accurate and complete information regarding the patient's medical and family history. There is always a small possibility of an error or failure in sample analysis; this is always a possibility with complex testing in any laboratory. Extensive measures are taken to avoid these errors. The accuracy of genetic testing is limited by the methods employed, and sometimes by the nature of the condition for which testing is requested.

### Limitations

Not all the exons in the genes being analyzed are targeted and captured due to certain inherent characteristics of the genome. Approximately 97-99% of exons are targeted in the diagnostic Exome tests. In addition, there is limited or no coverage in regions outside of the exome. Certain types of mutations are not detectable by this test, including trinucleotide repeat expansions, which are a common genetic cause of ataxia. This methodology will also not detect low level mosaicism and copy number variation mutations (i.e such as the deletion or duplication of an exon). Other types of rare genetic variation can interfere with this analysis. Pathogenic variants may be present in a region of a gene not covered by this test. Absence of findings for any particular gene does not mean that there are no pathogenic variants present in that gene. It is the responsibility of the referring physician, or a health care professional designated by the physician, to understand the limitations of the testing ordered, and to educate the patient regarding these limitations.

### Testing & Analysis Pipeline

Of the thousands of variants identified by exome sequencing, a list of variants that are located within the predefined set of genes is generated. Most variants that are identified as part of exome sequencing will NOT undergo interpretation by a laboratory staff member. Only those variants identified that fall within a predefined gene and are considered to be potentially relevant to the patient's condition are reviewed by a team of Board-Certified PhD geneticists, MD geneticists, and genetic counselors who will determine the likelihood of the variant being related to the patient's disorder.

### What is Reported?

UCGSL will report on genetic variants that have been reported to be pathogenic or predicted to be pathogenic or possibly pathogenic as well as unclassified variants in established genes on the pre-defined gene list.

### What is Not Reported?

- Variants that occur in genes outside of the pre-defined gene list, unless they are clearly the cause of the patient's phenotype.
- Variants that occur in genes defined as medically actionable by the American College of Medical Genetics and Genomics (ACMG), unless those genes are also included on the pre-defined gene list.
- Carrier status for recessive disorders, with the exception of carrier status for genes that are included on the pre-defined gene list.
- Benign sequence changes not associated with disease, which are commonly identified in healthy people.
- Synonymous (silent) sequence changes not associated with a change in the amino acid.
- Variations associated with increased or decreased risk to develop common disorders (like high blood pressure) or involved in drug metabolism.
- Variations that have been associated with an increased risk for diseases that might present at an advanced age (like Alzheimer's Disease) in which there is no treatment or preventative measures.
- Pathogenic mutations and variants in genes with no current known association with disease.

### Implication of Results

Because the implications of genetic testing results can be complex, involving both medical and emotional and social issues, results will only be reported through the referring physician or a professional designated by the physician, such as a genetic counselor. The issues associated with some types of genetic testing are particularly sensitive. Therefore, the laboratory reserves the right to provide testing only if genetic counseling can be provided.

### Confidentiality

Results and patient information are confidential and will only be released to the referring physician, unless written consent for further distribution is provided or the laboratory directors are required by law to release this information. For patients within The University of Chicago affiliated centers, policy may require that reports are provided to the medical records department.

### Consent for Neuromuscular Disorders Exome

**I consent to the Neuromuscular Disorders Exome being performed on my sample, as requested by my healthcare provider. I understand that the genes and variants reviewed as part of exome analysis will be dependent on the test selected by my healthcare provider.**

**Signature of Patient/Parent /Legally Authorized Representative:**

\_\_\_\_\_

**Date:** \_\_\_\_\_

If Legally Authorized Representative please describe relationship to individual:

\_\_\_\_\_

### Referring Clinician

By signing this consent form, the referring clinician 1) indicates that this consent form has been reviewed with the patient and/or the patient's parent or guardian, and 2) accepts responsibility for pre- and post- test genetic counseling.

**Signature of Referring Clinician:**

\_\_\_\_\_

**Date:** \_\_\_\_\_

## RESEARCH CONSENT FORM – The University of Chicago

### The Division of Biological Sciences | University of Chicago

**RECOMMENDED BUT OPTIONAL**

#### CONSENT/AUTHORIZATION BY SUBJECT FOR PARTICIPATION IN A RESEARCH PROTOCOL FOR THE BETTER UNDERSTANDING OF THEIR GENETIC CONDITION

Protocol Number: 11-0151

Name of Subject : \_\_\_\_\_

Date of Birth: \_\_\_\_\_

#### STUDY TITLE: Molecular Genetic Studies of Rare Orphan Genetic Disease

**Research Team:** Soma Das, Ph.D.  
5841 S. Maryland Ave. Room L-155 MC 0077, Chicago, IL 60637  
773-834-0555

You are being asked to allow your child to participate in a research study that may help us learn more about the genetic condition for which you are being tested. This consent form describes the study, the risks and benefits of participation, as well as how your confidentiality will be maintained. Please take your time to contact us with questions and feel comfortable making a decision whether to participate or not. If you decide to participate in this study, please sign this form. **Throughout this consent form, “you” will refer to you or your child, as appropriate.**

#### WHY IS THIS STUDY BEING DONE?

You have already consented to clinical genetic testing. We are asking you to also participate in further studies. The purpose of these studies is to learn more about the genetic cause of diseases tested for in our lab, gather more information about these disorders, and experiment with new methods that may be better for testing.

#### WHAT IS INVOLVED IN THE STUDY?

During this study, Dr. Das and her team will collect information about you for this research. We may contact your doctor to request additional Protected Health Information (PHI), which consists of any health information related to your diagnosis (such as date of birth, medical record number, primary diagnosis, clinical features, relevant and family history, outcome). The data collected will be used to develop a database of patients being tested for genetic diseases and will be kept for the duration of the database. This study will look at how often different genetic mutations happen and clinical information related to the mutation.

When our lab is researching new genes or testing methods that are related to your diagnosis, we may include your sample, with others from similar patients in a small study before offering this new test. This data will help in directing doctors about the likelihood of a positive or negative test result in their patient. We may also use your sample to set up new methods that will improve the clinical testing in our

laboratory. Your clinical information and sample, without any identifiers, may also be shared with other researchers that are interested in this specific condition.

#### HOW LONG WILL I BE IN THE STUDY?

Once enrolled, you will likely remain in this study as long as your DNA sample remains in our laboratory. If you want your sample, to be removed from the study at any time, please contact us, and the sample will not be used for further studies. Existing results will remain in our database until the study ends.

#### WHAT ARE THE RISKS OF THE STUDY?

There are no known added risks of the research. No additional information will be obtained from you, as all of the information has already been collected as part of clinical genetic testing or evaluation by your doctor.

#### ARE THERE ANY BENEFITS TO TAKING PART IN THE STUDY?

If you agree to take part in this study, there may be direct medical benefit to your family. We may identify a cause for the genetic disease in your family. If a mutation is identified in your DNA, through our testing, your referring doctor will be notified and will receive a clinical report. Our study may also be helpful in finding the genetic causes of disease and will benefit doctors and patients as a group.

#### WHAT OTHER OPTIONS ARE THERE?

You may choose not to participate.

#### WHAT ARE THE COSTS?

There will be no additional costs to you or your insurance company resulting from this research study. However, you or your insurance company will be responsible for costs related to your usual medical care.

#### WILL I BE PAID FOR MY PARTICIPATION?

You and your child will not be paid to participate.

#### WHAT ABOUT PRIVACY?

Study records that identify you will be kept private. All of your personal information will be entered into a password-protected database to prevent access to non-authorized personnel. If your data is shared with other researchers, all patient identifiers will be removed. Data from this study may be used in medical journals or presentations. If results from this study or related studies are made public in a medical journal, individual patients will not be identified. If we wish to use a patient's identity in a medical journal, we will ask for your permission at that time.

As part of the study, Dr. Das and her team will report any positive results of further testing to your referring doctor and/or genetic counselor. Dr. Das may also share these results, without your name or date of birth, with other researchers.

**RESEARCH CONSENT FORM – The University of Chicago**  
**The Division of Biological Sciences | University of Chicago**  
**RECOMMENDED BUT OPTIONAL**

People from the University of Chicago, including the Institutional Review Board (IRB), a committee that oversees research at the University of Chicago, may also view the records of the research. If health information is shared outside the University of Chicago, the same laws that the University of Chicago must obey may not protect your health information. Dr. Das does not have to give you any results that are not important to your health or your family's health at that time.

This consent form will be kept by the research team for at least six years. The study results will be kept in your child's research record and be used by the research team indefinitely. When the study ends, your personal information will be removed from all results. Any information shared with your doctor may be included in your medical record and kept forever.

The Genetic Information Nondiscrimination Act (GINA) is a federal law that may help protect you from health insurance or employment discrimination based on genetic information. GINA is a federal law that will protect you in the following ways:

- Health insurance companies and group plans may not request genetic information from this research;
- Health insurance companies and group plans may not use your genetic information when making decisions regarding your eligibility or premiums;
- Employers with 15 or more employees may not use your genetic information when making a decision to hire, promote, or fire you or when setting the terms of your employment.

GINA does not protect you against genetic discrimination by companies that sell life insurance, disability insurance, or long-term care insurance. GINA also does not protect you against discrimination based on an already-diagnosed genetic condition or disease.

**WHAT ARE MY RIGHTS AS A PARTICIPANT?**

Taking part in this study is optional. You may choose not to participate at any time during the study. Choosing not to participate or leaving the study will not affect your child's testing at the University of Chicago.

If you choose to leave the study and you do not want any of your child's future health information to be used, you must inform Dr. Das in writing at the address on the first page. Dr. Das may still use your child's information that was collected before to your written notice. You will be given a signed copy of this form. This consent form does not have an expiration date.

**WHO DO I CALL IF I HAVE QUESTIONS OR PROBLEMS?**

If you have further questions about the study, please call 773-834-0555.

*If you have any questions about your rights in this research study you may contact the IRB, which protects participants in research projects. You may reach the Committee office between 8:30 am and 5:00 pm, Monday through Friday, by calling (773) 702-6505 or by writing: University of Chicago, Institutional Review Board, 5841 S. Maryland Ave., MC7132, I-625, Chicago, IL 60637.*

**Consent**

I have received information about this research project and the procedures. No guarantee has been given about possible results. I will receive a signed copy of this consent form for my records.

I give my permission to participate in the above research project.

**Signature of Subject:** \_\_\_\_\_

**Date:** \_\_\_\_\_

I give my permission for my child/relative/the person I represent to participate in the above research project.

**Signature of Parent / Legal Guardian / Legally Authorized Representative:**

\_\_\_\_\_

**Date:** \_\_\_\_\_