AT THE FOREFRONT UChicago Medicine | Medical Laboratories

# Movement Disorders Requisition Form

**Client Account Code:**

If code not known or client account not set up, contact Venessa Gamboa at 312-213-5441.


**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
ucglabs@bsd.uchicago.edu | dnatesting.uchicago.edu | CLIA#: 14D0671659 | CAP#: 18827-01


## Patient Information

**Name:** *Last*_____ *First*_____ **Date of Birth:** _____

**Gender:** ☐ Male ☐ Female **MRN:** _____

**Ethnicity:** ☐ European ☐ African-American ☐ Hispanic ☐ Asian ☐ Ashkenazi Jewish ☐ Other _____

## Ordering Physician Information

☐ **Referring Physician:** _____

Phone:_____ Fax:_____

Email: _____

☐ **Genetic Counselor:** _____

Phone:_____ Fax:_____

Email: _____

☐ **Referring Lab:** _____

Phone:_____ Fax: _____

Email: _____

## Test Requested (REQUIRED)

☐ Comprehensive Ataxia Panel **CONCURRENT** [Ataxia Exome + Repeat Expansion Panel **run concurrently**]
☐ Comprehensive Ataxia Panel **REFLEX** [Repeat Expansion Panel **with reflex to** Ataxia Exome ]
☐ Ataxia Repeat Expansion Panel
☐ Ataxia Exome [proband only]
☐ Hereditary Spastic Paraplegia Exome [proband only]
☐ Dystonia Chorea Parkinson Exome [proband only]
☐ Single Gene Repeat Expansion Testing (list gene here): _____
☐ Single Gene Sequencing (list gene here): _____

## 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: _____

## 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 Movement Disorders Consent Form *(required)*
☐ Completed Research Consent Form *(recommended)*

## For Office Use Only

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# MOVEMENT 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**

**Movement disorder(s) present?**
☐ YES (note age of onset below) ☐ NO
☐ UNKNOWN
**Age of onset:** _____
- ☐ Ataxia
  - ☐ Cerebellar ataxia
  - ☐ Episodic ataxia
  - ☐ Sensory ataxia
  - ☐ Vestibular ataxia
- ☐ Chorea
- ☐ Dystonia
  - ☐ Blepharospasm
  - ☐ Cervical dystonia
  - ☐ Focal dystonia
  - ☐ Generalized dystonia
  - ☐ Hemidystonia
  - ☐ Paroxysmal dystonia
  - ☐ Segmental dystonia
  - ☐ Temporal dystonia
- ☐ Myoclonus
- ☐ Parkinson's
- ☐ Spastic paraplegia
  - ☐ Complicated / complex SP
  - ☐ Uncomplicated / pure SP
- ☐ Other: _____

**Progressive movement disorder?**
☐ YES ☐ NO ☐ UNKNOWN

**Previous testing for trinucleotide repeat expansions (SCA's, Friedreich's ataxia)?**
☐ YES (see below) ☐ NO ☐ UNKNOWN
Please specify results: _____

**Other neurological findings present?**
☐ YES (see below) ☐ NO ☐ UNKNOWN
- ☐ 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 atrophy
- ☐ Generalized atrophy
- ☐ Leukodystrophy
- ☐ Molar tooth sign
- ☐ 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
- ☐ Telangiectasies
- ☐ Ichthyosis
- ☐ Other: _____

**Family history of movement disorder or other relevant findings?**
☐ YES (see below) ☐ NO ☐ UNKNOWN
Please specify: _____

**Biochemical testing performed?**
☐ YES (see below) ☐ NO ☐ UNKNOWN
**Results of biochemical testing:**
- ☐ Elevated AFP
- ☐ Normal results for the following tests: _____
- ☐ Other: _____

**Cardiac findings present?**
☐ YES (see below) ☐ NO ☐ UNKNOWN
Please specify: _____

**Gastrointestinal (GI) issues present?**
☐ YES (see below) ☐ NO ☐ UNKNOWN
Please specify: _____

**Growth abnormality present?**
☐ YES (see below) ☐ NO ☐ UNKNOWN
Please specify: _____

**Musculoskeletal findings present?**
☐ YES (see below) ☐ NO ☐ UNKNOWN
- ☐ Muscle weakness
- ☐ Other: _____

**Neoplasms or immunodeficiency?**
☐ YES (see below) ☐ NO ☐ UNKNOWN
Please specify: _____

**Vision/hearing abnormality present?**
☐ YES (see below) ☐ NO ☐ UNKNOWN
- ☐ Oculomotor apraxia
- ☐ Optic atrophy
- ☐ Nystagmus
- ☐ Retinopathy
- ☐ 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.

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# BILLING OPTIONS

*There are some tests for which we do not offer insurance billing. Please consult our website and quick guide (list of tests, costs, TAT and CPT codes) or contact us for more information.*
*All samples received with incomplete billing information will delay processing time.*
*Test canceled while "in progress" will be billed for the amount of work completed up to that point.*
*Please forward all client billing questions to: venessa.gamboa@uchospitals.edu or call 312-213-5441.*

**Patient Name:** *Last*_____ *First* _____(MI):_____Date of Birth: _____

## 1.) Institutional Billing

Billing Institution and Client Account Code:_____PO#:_____

Financial Contact:_____Phone:_____Fax:_____

Address:_____City:_____State:_____Zip: _____

Email (required): _____

## 2.) Insurance Billing

*(We do NOT accept Illinois or any out-of-state Medicaid. Please note we do not bill insurance for all our testing options. Please see our website for more details. Prices listed on our website are not applicable for insurance billing, please contact us for insurance pricing information.) A legible photocopy of the front and back of the insurance card and insurance authorization must be included.*

ICD-10 Diagnosis Code(s):_____(***Must be provided or insurance cannot be filed.***) Policyholder Name:_____Date of Birth:_____Sex: ☐ Male ☐ Female

Policyholder Address:_____City:_____State:_____Zip:_____

Relationship to the Patient: ☐ Self ☐ Spouse ☐ Dependent ☐ Other Preauthorization # (*if applicable*): _____

Name of Primary Insurance:_____Policy No._____Group No.: _____

Insurance Address:_____City:_____State:_____Zip:_____

PCP/Referring Physician Name:_____NPI #: _____

Name of Secondary Insurance:_____Policy No.:_____Group No.: _____

Insurance Address:_____City:_____State:_____Zip: _____

The policy holder's signature to the following statement: I hereby authorize any physician who treated or attended to me or my dependent(s) to furnish any medical information requested. In consideration of services rendered, I hereby transfer and assign to the University of Chicago Genetic Services Laboratories any benefits of insurance I may have. I assume responsibility for the balance of the cost of testing not paid by my insurance company. A photocopy of this authorization shall be considered as effective and valid as original.

Authorized Signature:_____Date:_____

**See our QuickGuide to Genetic Testing for complete list of Costs, TAT and CPT Codes.**

# MOVEMENT DISORDER EXOME PANEL CONSENT FORM

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

***REQUIRED FOR EXOME PANEL 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 offers different exome panels for movement disorders, including an Ataxia Exome Panel, a Dystonia Exome Panel, and a Hereditary Spastic Paraplegia Exome Panel. For these exome panel tests, analysis of the exome sequencing data is limited to a pre-defined list of genes that have been associated with the movement disorder indicated for each specific panel. For cases without a clearly pathogenic variant identified in the pre-defined list of panel 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 movement disorder in the patient's family.

## Accuracy

The analysis performed is specific to genes associated with the movement disorder indicated in the name of the panel requested. 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 90-95% of exons are targeted in the diagnostic Exome Panel 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 panel 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 panel 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 panel gene list.

## What is Not Reported?

- Variants that occur in genes outside of the pre-defined panel 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 panel gene list.
- Carrier status for recessive disorders, with the exception of carrier status for genes that are included on the pre-defined panel 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 Movement Disorder Exome Panel

**I consent to a "Movement Disorder" Exome Panel being performed on my sample, as requested by my healthcare provider. I understand that the genes and variants reviewed as part of exome panel analysis will be dependent on the panel selected by my healthcare provider (Ataxia Exome Panel, Dystonia Exome Panel, or Hereditary Spastic Paraplegia Panel).**

**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:** _____

## 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.

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 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*

## 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:** _____