



Patient information

First name _____ Last name _____
 Gender: Female Male Unknown
 Date of birth (mm/dd/yy) _____
 Mailing address _____
 City _____ State _____ Zip Code _____
 Home phone _____ Work Phone _____

Ethnic Background

Maternal

African American Ashkenazi Jewish Asian Caucasian
 Hispanic Native American Other Jewish Other _____

Paternal

African American Ashkenazi Jewish Asian Caucasian
 Hispanic Native American Other Jewish Other _____

Sample information

Medical record # _____ Specimen ID# _____
 Date sample obtained (mm/dd/yy) _____
Specimen Type
 Blood in EDTA (Adults 8-10 ml; Children 4-6 ml; Infants 2-3 ml)
 Fresh/Frozen tissue (>50mg): Muscle Liver Other _____
AND Blood in EDTA
 DNA (discouraged for this test) (>5 ug): Tissue source _____
 concentration _____ (ug/ml) total Volume _____ (ul)
 Fetal sample (call before sending): Tissue source _____

Reporting Address

Physician / Genetic counselor _____
 Mailing address _____
 City _____ State _____ Zip code _____
 Phone _____ Fax (important) _____
 Email _____ Beeper _____

Duplicate Report Address

Physician / Genetic counselor _____
 Mailing address _____
 Phone _____ Fax (important) _____
 Email _____

Test requested

Please select choice and provide clinical information on page 2

- 390 Test for 16 common mtDNA point mutations + deletion/duplication analysis for mtDNA and 116 nuclear genes
- 392 Full sequence analysis of the mitochondrial genome
- 393 MtDNA deletion/over-replication analysis
- 394 POLG gene sequencing

Coming Soon!

Next-Gen sequencing of 20+ relevant nuclear genes

Testing for known mutation(s)

- 9011 Testing for ONE known familial mutation in a nuclear gene
- 9012 Testing for TWO known familial mutations in a nuclear gene
- 906 Testing for ONE known familial exon-level deletion/duplication
- 9014 Testing for ONE mtDNA mutation (of the 16 common mtDNA mutations only)
- 903 Testing for ONE mtDNA mutation (custom mutation)

Gene: _____ Mutation: _____
 Proband Name: _____
 Proband GeneDx #: _____
 Relationship to proband: _____

Reason for testing - please complete (required):

- Symptomatic Asymptomatic
- Prenatal (for nuclear genes only)
- Positive control sample (no report issued) for patient/relative:

GeneDx ID _____ First name _____ Last name _____

Clinical information and family history is crucial for accurate interpretation of results. Please provide relevant information on page 2

Clinical diagnosis: _____

Family History:

Family member(s) affected: No Yes
 Relationship to proband: _____
 Pedigree: _____

Ordering Checklist

- Test requisition form Completed clinical information form (page 2)
- Completed payment option form (page 3) Informed consent (page 4)
- Specimen tube appropriately labeled

For office use only:



Patient information

Last name

First name

Middle

Clinical Information is crucial for accurate interpretation of test results (Check all that apply):

General

- Developmental delay
- Mental retardation
- Intrauterine Growth Retardation (IUGR)
- Short stature
- Failure to thrive

Neurologic & Muscular

- Hypotonia
- Psychomotor regression
- Seizures
- Myoclonus
- Ataxia
- Exercise intolerance / easy fatigue
- Muscle weakness
- Stroke / stroke-like episodes
- Autism / autistic-like behavior
- Recurrent headache / migraine
- Psychiatric symptoms
- Other _____

Gastrointestinal and Liver

- Recurrent vomiting
- Gastrointestinal reflux
- Delayed gastric emptying
- Chronic diarrhea
- Constipation
- Chronic intestinal pseudo-obstruction
- Hepatic failure
- Elevated transaminases

Cardiovascular

- Cardiomyopathy
- Arrhythmia or conduction defect

Endocrine

- Diabetes melitus: Type I Type II
- Hypothyroidism
- Hypoparathyroidism
- Other _____

Ophthalmologic and auditory

- CPEO (ophthalmoplegia)
- Ptosis
- Blindness
- Subacute bilateral visual failure
- Optic atrophy
- Retinitis pimentosa
- Sensorineural hearing loss
- Ototoxicity (Aminoglycoside-induced)

Metabolic

- Ketosis
- Lactic acidemia / High CSF lactate
- High lactate/pyruvate ratio (>50:1)
- Elevated pyruvate
- Elevated alanine
- Organic aciduria, Tiglyglycine
- Low plasma carnitine
- CPK abnormalities
- Other _____

Muscle Biopsy

- Abnormal histology _____
- Abnormal ultrastructure (EM) _____
- Abnl. respiratory enzymes _____
- Large mitochondria / mitochondrial proliferation
- COX deficiency
- Ragged Red Fiber

Other clinical/laboratory findings

- SIDS / Unexplained death
- Anemia / Neutropenia / Pancytopenia
- Renal tubulopathy
- Urinary retention
- Hair abnormalities
- Basal ganglia abnormalities
- Paraganglioma / pheochromocytoma
- Other, specify _____

Payment Options

I. Institutional Billing Information:

PO#/Department Code

Hospital/Lab Name

Contact Name

Address

City State Zip Code

Phone Fax

INSTITUTIONAL BILLING ADDRESS STAMP

2. Payment by credit card

The full amount of the test fee is charged at the time of sample submission.

Name as it appears on card

Billing address

City State Zip Code

Phone

Mastercard Visa Discover American Express

Account Number

Expiration date 3/4 Digit Security Code

Please bill my credit card in the amount of \$_____ for diagnostic laboratory tests performed by GeneDx, Inc.

Signature (Required) Date

3. Payment by check or money order:

Minimum of 75% of the cost of the test is required at the time of sample submission, with the remainder of the fee billed at the time of test completion.*

Check or money order enclosed in the amount of \$_____.

*** For patients from outside the United States, 100% of the fee is due at the time of sample submission**

4. Insurance Billing:

GeneDx cannot bill Medicare. GeneDx is not a participating member with any Medicaid/MediCal program.

GeneDx does not bill Insurance Companies directly unless

all of the following is submitted:

- Credit card information (complete part 2) to which any outstanding balance may be billed;
- An authorization number or letter of agreement from the insurance company.
 - The letter of agreement should be directed to GeneDx
 - detail the reimbursement rate
 - the name of the department or individual to whom the bill will be sent (including address, phone and fax numbers)
 - the patient's name and policy number.
- Copy of both sides of the insurance card.
- ICD9 codes (to be provided by physician) _____

I UNDERSTAND THAT I AM RESPONSIBLE IN ALL CASES FOR ALL FEES NOT COVERED BY INSURANCE.

Signature (Required)

Note

IF YOU plan to apply on your own to your insurance carrier for reimbursement of your expenses for this test, the following information may be helpful in the case that GeneDx is requested by the carrier to prepare supporting documentation for you to use in your insurance claim:

Insurance Carrier
Is this a Blue Cross/Blue Shield Plan? YES NO

Subscriber Name
Is this a Medicaid plan? YES NO

Subscriber DOB

Informed Consent for Molecular Testing for Mitochondrial Disorders

My signature of informed consent certifies that I have been provided with the following facts about the genetic test for which I am giving permission. I have had the opportunity to discuss the benefits, risks and limitations.

Why is this test performed?

1. Many genetic diseases are caused by small mutation or changes in an individual's genes. On the other hand, some genetic disorders are caused by a deletion or duplication of a larger section of a gene, or of the entire gene. Genetic diseases can be caused by both small and large mutations.
2. In this genetic test for mitochondrial disorders [my/my child's] DNA will be studied to see if a mutation and/or deletion/duplication have occurred in a gene or genes associated with mitochondrial function. This genetic test for mitochondrial disorders cannot be expected to identify any changes (small or large) in genes not included in this test.
3. I understand that this test is performed to determine if I and/or my child have a mutation in a gene associated with a specific genetic disease, and whether I am/we are affected with, or at an increased risk to someday be affected with, this genetic disease.
4. This test is not the only way to look for changes in my DNA and my physician may recommend this test before or after ordering other genetic tests for mitochondrial disorders.

What might I find out from this test?

5. I might learn that no change (mutation, deletion or duplication) was found in the gene(s) studied. This outcome does not mean that [I do not/my child does not] have a genetic disease.
6. I might learn that a specific gene or genes do have a change (mutation, deletion or duplication), thus explaining the cause of the disorder I already know [I have/my child has] been diagnosed with.
7. I might learn that a gene change was found in my gene(s) that is associated with possible long-term medical problems that I do not already know about. My physician will be informed of any long-term risks that may be associated with the findings of this test, according to current medical understanding.
8. I am aware that the results of this test may have implications for other family members as well.
9. I might learn that I have a gene change (gene variant) whose clinical consequences or relationship to medical problems is currently unknown. The variant could be a normal change in the gene, or could be a disease-causing mutation.
10. When a gene variant is found, it is important to find out if the gene variant (change) is also found in other family members. Testing the biological parents of the affected individual, and/or testing other affected and/or unaffected family members may be necessary to determine if the gene variant is disease-causing or a genetic change not associated with a disease.

What are the limitations of the test?

11. In some cases, genetic tests are unable to identify a change in a gene even though the change may exist. This event may be due to the current lack of knowledge about a gene's complete structure, or due to the fact that there may be other genes associated with a disease that have not yet been identified. In other cases, there is an inability of the current technology (test method), to identify certain types of changes in the gene(s).
12. An error in the diagnosis of a disease may occur if the true biological relationships of the family members being tested are not as stated. This is particularly important when specimens from parents of an affected child are submitted to the laboratory for testing. For example, if the stated father of an individual is not the true biological father (non-paternity), this test may detect non-paternity, and it may be necessary to report this finding to the individual(s) who requested testing. Non-disclosure regarding any biological relationships may result in incorrect result interpretation, incorrect diagnoses in the family members and/or inconclusive test results.
13. I understand that the genetic test performed by GeneDx is specific for this disease and in no way guarantees my health or the health of my living or unborn children. This test does not have the ability to detect all of the long-term medical risks that [I/my child] might experience. The accuracy of genetic testing is dependent on the clinical diagnosis made elsewhere, and GeneDx cannot be responsible for incorrect clinical diagnoses.

After testing is complete, for test improvement and training purposes, the anonymized submitted specimen and clinical information may be used. Confidentiality of each sample is maintained. Declining to participate shall not affect the quality of services you receive. If you wish to not have your blood/DNA sample used for this purpose, although it is anonymous, please check this box.

If not checked, consent is implied.

Informed Consent

Sign here to provide consent for Mitochondrial testing at GeneDx:

Signature: _____ **Date:** _____