

Patient Information

First name _____ Last name _____
 Gender Male Female Date of birth (mm/dd/yy) _____
 Ancestry Caucasian Eastern European Northern European
 Western European Native American Middle Eastern
 African American Asian Pacific Islander
 Caribbean Central/South American
 Ashkenazi Jewish Hispanic Other: _____

Mailing address _____
 City _____ State _____ Zip code _____
 Home phone _____ Work phone _____
 Email _____ Patient's primary language if not English _____

Sample Information

Medical record # _____ Specimen ID _____ Date sample obtained (mm/dd/yy) _____
 Blood in EDTA (5-6 mL in lavender top tube)
 Other _____ (Call lab)
 Patient has had a blood transfusion Yes No Date of last transfusion __/__/__
 (2-4 weeks of wait time is required for some testing) Specimens are not accepted for patients who have had allogeneic bone marrow transplants

Clinical Information

Clinical diagnosis: _____
ICD-10 codes: _____
DETAILED MEDICAL RECORDS, CLINICAL SUMMARY, PICTURES AND FAMILY HISTORY MUST BE SENT WITH REQUISITION FORM AND SAMPLES.

Test Ordered

J774 GenomeSeqDx (Trios Only)

Biological Parent Sample Information

Mother: To be sent later*
 First name _____ Last name _____ DOB _____
 Asymptomatic Symptomatic
Father: To be sent later*
 First name _____ Last name _____ DOB _____
 Asymptomatic Symptomatic
Other: To be sent later*
 Relationship to Proband: _____
 First name _____ Last name _____ DOB _____
 Asymptomatic Symptomatic
***ADDITIONAL SAMPLES MUST BE RECEIVED WITHIN 3 WEEKS**

Patient Consent (sign here or on the consent document)

I have read the Informed Consent document and I give permission to GeneDx to perform genetic testing as described. I also give permission for my specimen and clinical information to be used in de-identified studies at GeneDx to improve genetic testing and for publication, if appropriate. My name or other personal identifying information will not be used in or linked to the results of any studies and publications. I also give GeneDx permission to inform me or my health care provider in the future about research opportunities, including treatments for the condition in my family.

Check this box if you wish to opt out of being contacted for research studies.
 Check this box if you do not wish to receive secondary findings.
 Check this box if you are a New York state resident, and give permission for GeneDx to retain any remaining sample longer than 60 days after the completion of testing.

Patient/Guardian Signature _____

Date _____

Ordering Account Information

Acct # _____ Account Name _____
 Reporting Preference*: Care Evolve Fax Email
**If unmarked, we will use the account's default preferences or fax to new clients.*

Physician _____ NPI # _____
 Genetic Counselor _____
 Street address 1 _____
 Street address 2 _____
 City _____ State _____ Zip code _____
 Phone _____ Fax (important) _____
 Email _____ Beeper _____

Send Additional Report Copies To:

Physician or GC/Acct # _____ Fax#/Email/CE # _____
 Physician or GC/Acct # _____ Fax#/Email/CE # _____

Billing Information (GeneDx does NOT bill insurance for this test)

Institutional Bill

GeneDx Account # _____
 Hospital/Lab Name _____
 Contact Name _____
 Address _____
 City _____ State _____ Zip Code _____
 Phone _____ Fax _____

Patient Bill/Self-Pay

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 \$ _____

Payment by credit card (all major cards accepted)

MasterCard Visa Discover American Express

Name as it appears on card _____

Account Number _____ Expiration date _____ CVC _____

Amount _____ **Signature** _____ Date _____

For GeneDx Use Only

Account # _____ Account Name _____

First Name _____

Last Name _____

Date of Birth (mm/dd/yy) _____

Clinical information is crucial for accurate interpretation of results. Please attach medical records.

Please check all that apply. This is not a substitute for submitting clinical records.

Perinatal history

- Prematurity
- IUGR
- Oligohydramnios
- Polyhydramnios
- Cystic hygroma/increased NT

Growth

- Failure to thrive
- Growth retardation/short stature
- Overgrowth
- Macrocephaly
- Microcephaly

Physical/Cognitive Development

- Fine motor delay
- Gross motor delay
- Speech delay
- Intellectual disability/MR
IQ: _____
- Learning disability
- Developmental regression

Behavioral

- Autism spectrum disorder
- Autistic features
- Obsessive-compulsive disorder
- Stereotypic behaviors
- Other psychiatric symptoms

Craniofacial/Ophthalmologic/Auditory

- Cataracts
- Cleft lip/palate
- Coloboma of eye
- CPEO (ophthalmoplegia)
- Ptosis
- Blindness
- Optic atrophy
- Retinitis pigmentosa
- Hearing loss
- Ototoxicity (aminoglycoside-induced)
- External ear malformation
- Facial dysmorphism - please describe:

Cardiac/congenital heart malformations

- ASD
- VSD
- Coarctation of aorta
- Hypoplastic left heart
- Tetralogy of Fallot
- Cardiomyopathy
- Arrhythmia/conduction defect
- Other: _____

Cancer/Malignancy

- Age of onset: _____
- Tumor type: _____
- Location(s): _____
- Affected relatives: _____

Skin/Hair

- Abnormal hair: _____
- Quality/Quantity: _____
- Hair distribution: _____
- Abnormal nails: _____
- Abnormal pigmentation: _____
- Abnormal connective tissue: _____
- Blistering
- Ichthyosis
- Skin tumors/Malignancies
- Other: _____

Brain malformations/abnormal imaging

- Agenesis of the corpus callosum
- Holoprosencephaly
- Lissencephaly
- Cortical dysplasia
- Heterotopia
- Hydrocephalus
- Brain atrophy
- Periventricular leukomalacia
- Hemimegalencephaly
- Abnormalities of basal ganglia
- Other: _____

Neurological/Muscular

- Ataxia
- Chorea
- Dystonia
- Hypotonia
- Hypertonia
- Seizures (type: _____)
- Spasticity
- Exercise intolerance/easy fatigue
- Muscle weakness
- Stroke/stroke-like episodes
- Recurrent headache/migraine

Gastrointestinal

- Gastroschisis/omphalocele
- Pyloric stenosis
- Tracheoesophageal fistula
- Delayed gastric emptying
- Eosinophilic esophagitis
- Gastrointestinal reflux
- Recurrent vomiting
- Chronic diarrhea
- Constipation
- Chronic intestinal pseudo-obstruction
- Hirschsprung disease
- Hepatic failure
- Elevated transaminases

Skeletal/Limb abnormalities

- Contractures
- Club foot
- Polydactyly
- Syndactyly
- Scoliosis
- Vertebral anomaly
- Other: _____

Genitourinary abnormalities

- Ambiguous genitalia
- Hypospadias
- Hydronephrosis
- Undescended testis
- Kidney malformation
- Renal agenesis
- Renal tubulopathy
- Other: _____

Endocrine

- Diabetes mellitus:
 Type I Type II
- Hypothyroidism
- Hypoparathyroidism
- Pheochromocytoma/paraganglioma

Metabolic

- Ketosis
- Lactic acidemia/high CSF lactate
- Elevated pyruvate
- Elevated alanine
- Organic aciduria
- Low plasma carnitine
- CPK abnormalities

Hematologic/Immunologic

- Anemia/neutropenia/pancytopenia
- Immunodeficiency
- Other: _____

Other testing (summarize or attach reports):

- Chromosomes/FISH: _____
- Array CGH: _____
- Fragile X syndrome: _____
- Muscle biopsy: _____
- Other relevant results (clinical or research):

Additional relevant clinical info: _____

I understand that my health care provider has ordered the following genetic testing for {me/my child}: _____.

General Information About Genetic Testing

What is genetic testing?

DNA provides instructions for our body's growth and development. Genes are distinct sequences of DNA, and are arranged on chromosomes. The DNA in a gene contains instructions for making proteins, which determine things like growth and metabolism as well as traits like eye color and blood type. Genetic disorders are caused by harmful changes in DNA or from changes in the structure or number of chromosomes. Genetic testing is a laboratory test that tries to identify these harmful changes in chromosomes or the DNA. Genetic testing can be a diagnostic test, which is used to identify or rule out a specific genetic condition. Genetic screening tests are used to assess the chance for a person to develop or have a child with a genetic condition. Genetic screening tests are not typically diagnostic and results may require additional diagnostic testing.

The purpose of this test is to see if I, or my child, may have a genetic variant or chromosome rearrangement causing a genetic disorder or to determine the chance that I, or my child, will develop or pass on a genetic disorder in the future. 'My child' can also mean my unborn child, for the purposes of this consent.

Additional information about the specific test being ordered is available from my health care provider or I can go to the GeneDx website, www.genedx.com. This information includes the specific types of genetic disorders that can be identified by the genetic test, the likelihood of a positive result, and the limitations of genetic testing.

If {I/my child} already know the specific gene variant(s) or chromosome rearrangement that causes the genetic disorder in my family, I will inform the laboratory of this information.

What could I learn from this genetic test?

The following describes the possible results from the test:

1) Positive: A positive result indicates that a genetic variant has been identified that explains the cause of {my/my child's} genetic disorder or indicates that {I/my child} am at increased risk to develop the disorder in the future. It is possible to test positive for more than one genetic variant.

2) Negative: A negative result indicates that no disease-causing genetic variant was identified for the test performed. It does not guarantee that {I/my child} will be healthy or free from genetic disorders or medical conditions. If {I/my child} test negative for a variant known to cause the genetic disorder in other members of {my/my child's} family, this result rules out a diagnosis of the same genetic disorder in {me/my child} due to this specific change.

3) Inconclusive/Variant of Uncertain Significance (VUS): A finding of a variant of uncertain significance indicates that a genetic change was detected, but it is currently unknown whether that change is associated with a genetic disorder either now or in the future. A variant of uncertain significance is not the same as a positive result and does not clarify whether {I/my child} is at increased risk to develop a genetic disorder. The change could be a normal genetic variant or it could be disease-causing. Further analysis may be recommended, including testing both parents and other family members. Detailed medical records or information from other family members also may be needed to help clarify results.

4) Unexpected results: In rare instances, this test may reveal an important genetic change that is not directly related to the reason for ordering this test. For example, this test may tell me about the risk for another genetic condition {I/my child} is not aware of or it may indicate differences in the number or rearrangement of sex chromosomes. This information may be disclosed to the ordering health care provider if it likely impacts medical care.

Result interpretation is based on currently available information in the medical literature, research and scientific databases. Because the literature, medical and scientific knowledge are constantly changing, new information that becomes available in the future may replace or add to the information GeneDx used to interpret {my/my child's} results. Providers can contact GeneDx at any time to discuss the classification of an identified variant. In addition, I or {my/my child's} health care providers may monitor publicly available resources used by the medical community, such as ClinVar (www.clinvar.com), to find current information about the clinical interpretation of my/my child's variant(s).

For tests that evaluate data from multiple family members, my spouse, or partner concurrently, results may be included in a single comprehensive report.

What are the risks and limitations of this genetic test?

- Genetic testing is an important part of the diagnostic process. However, genetic tests may not always give a definitive answer. In some cases, testing may not identify a genetic variant even though one exists. This may be due to limitations in current medical knowledge or testing technology.
- Accurate interpretation of test results may require knowing the true biological relationships in a family. Failing to accurately state the biological relationships in {my/my child's} family may result in incorrect interpretation of results, incorrect diagnoses, and/or inconclusive test results. In some cases, genetic testing can reveal that the true biological relationships in a family are not as they were reported. This includes non-paternity (the stated father of an individual is not the biological father) and consanguinity (the parents of an individual are related by blood). It may be necessary to report these findings to the health care provider who ordered the test.
- Genetic testing is highly accurate. Rarely, inaccurate results may occur for various reasons. These reasons include, but are not limited to: mislabeled samples, inaccurate reporting of clinical/medical information, rare technical errors, or unusual circumstances such as bone marrow transplantation, or the presence of change(s) in such a small percentage of cells that the change(s) may not be detectable by the test (mosaicism).
- This test does not have the ability to detect all of the long-term medical risks that {I/my child} might experience. The result of this test does not guarantee my health or the health of my child/fetus. Other diagnostic tests may still need to be done, especially when only a genetic screening test has been performed previously.
- Occasionally, an additional sample may be needed if the initial specimen is not adequate.

Patient Confidentiality and Genetic Counseling

It is recommended that I receive genetic counseling before and after having this genetic test. I can find a genetic counselor in my area here: www.nsgc.org. Further testing or additional consultations with a health care provider may be necessary.

To maintain confidentiality, the test results will only be released to the referring health care provider, to the ordering laboratory, to me, to other health care providers involved in {my/my child's} diagnosis and treatment, or to others as entitled by law. The United States Federal Government has enacted several laws that prohibit discrimination based on genetic test results by health insurance companies and employers. In addition, these laws prohibit unauthorized disclosure of this information. For more information, I understand that I can visit www.genome.gov/10002077.

Account # _____ Account Name _____

First Name _____ Last Name _____ Date of Birth (mm/dd/yyyy) _____

International Specimens

If {I/my child} reside outside the United States, I attest that by providing a sample for testing, I am not knowingly violating any export ban or other legal restriction in the country of {my/my child's} residence.

Specimen Retention

After testing is complete, the de-identified submitted specimen may be used for test development and improvement, internal validation, quality assurance, and training purposes. DNA specimens are not returned to individuals or to referring health care providers unless specific prior arrangements have been made.

I understand that samples from residents of New York State will not be included in the de-identified research studies described in this authorization and will not be retained for more than 60 days after test completion, unless specifically authorized by my selection below. The authorization is optional, and testing will be unaffected if I do not check the box for the New York authorization language.

Database Participation

De-identified health history and genetic information can help health care providers and scientists understand how genes affect human health. Though {I/my child} may not personally benefit, sharing this information helps health care providers to provide better care for their patients and researchers to make discoveries. GeneDx shares this type of information with health

care providers, scientists, and health care databases. No personal identifying information will be shared, as it will be replaced with a unique code.

Even though only a code is used for the reporting to the databases, there is a risk that {I/my child} could be identified based on the genetic and health information that is shared. GeneDx believes that this is unlikely, though the risk is greater if I have already shared {my/my child's} genetic or health information with public resources, such as genealogy websites.

Recontact for Research Participation

Separate from the above, GeneDx may collaborate with scientists, researchers and drug developers to advance knowledge of genetic diseases and to develop new treatments. If there are opportunities to participate in research relevant to the disorder in {my/my child's} family, and if I have consented for recontact, GeneDx may allow my health care provider to be recontacted for research purposes, such as the development of new testing, drug development, or other treatment modalities. In some situations, such as if my health care provider is not available, I may be contacted directly.

Any research that results in medical advances, including new products, tests or discoveries, may have potential commercial value and may be developed and owned by GeneDx or the collaborating researchers. If any individuals or corporations benefit financially from these studies, no compensation will be provided to {me/my child} or {my/my child's} heirs.

Exome/Genome Sequencing Secondary Findings & Opt-Out

As many different genes and conditions are analyzed in the GenomeSeqDx, XomeDx, XomeDxPlus, XomeDxPrenatal and XomeDxXpress tests, these tests may reveal some findings not directly related to the reason for ordering ES/GS. Such findings are called "incidental" or "secondary" and can provide information that was not anticipated.

Secondary findings are variants, identified by the GenomeSeqDx, XomeDx, XomeDxPlus, XomeDxPrenatal and XomeDxXpress tests, in genes that are unrelated to the individual's reported clinical features.

The American College of Medical Genetics and Genomics (ACMG) has recommended that secondary findings identified in a specific subset of medically actionable genes associated with various inherited disorders be reported for all probands undergoing genome or exome sequencing. Please refer to the latest version of the [ACMG Recommendations for Reporting of Secondary Findings in Clinical Exome and Genome Sequencing](#) for complete details of the genes and associated genetic disorders. Reportable secondary findings will be confirmed by an alternate test method.

What will be reported for the proband

- All known and/or expected pathogenic variants identified in the genes (for which a minimum of 10X coverage was achieved by the GenomeSeqDx, XomeDx, XomeDxPlus, XomeDxPrenatal or XomeDxXpress test), as recommended by the ACMG.

What will be reported for relatives (if tested with GenomeSeqDx, XomeDx, XomeDxPlus, XomeDxPrenatal or XomeDxXpress)

- The presence or absence for any secondary findings reported for the proband will be provided for all relatives tested by GenomeSeqDx, XomeDx, XomeDxPlus, XomeDxPrenatal or XomeDxXpress.

Limitations

- Pathogenic variants may be present in a portion of the gene not covered by this test and therefore are not reported. **The absence of reportable secondary findings for any particular gene does not mean there are no pathogenic variants in that gene.**
- Pathogenic variants that may be present in a relative, but are not present in the proband, will not be identified, or reported.
- Only changes at the sequence level will be reported in the secondary findings report. Larger deletions/duplications, abnormal methylation, triplet repeat or other expansion variants, or other variants not routinely identified by clinical exome and genome sequencing will not be reported.

Patient Consent (sign here or on page 1 of the test requisition form)

I have read the Informed Consent document and I give permission to GeneDx to perform genetic testing as described. I also give permission for my specimen and clinical information to be used in de-identified studies at GeneDx to improve genetic testing and for publication, if appropriate. My name or other personal identifying information will not be used in or linked to the results of any studies and publications. I also give GeneDx permission to inform me or my health care provider in the future about research opportunities, including treatments for the condition in my family.

- Check this box if you wish to opt out of being contacted for research studies.
- Check this box if you do not wish to receive secondary findings.
- Check this box if you are New York state resident, and give permission for GeneDx to retain any remaining sample longer than 60 days after the completion of testing.

Patient/Guardian Signature

Date (mm/dd/yyyy)

If I wish to change my decisions or have any questions, I understand that I may contact the laboratory via email at genedx@genedx.com or by phone at +1-301-519-2100, or if I am located in the United States, toll free at +1-888-729-1206.