

## 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) \_\_\_\_\_  
 CVS  Amniotic Fluid  Fetal Blood (PUBS)  
 Cultured amniocytes  Cultured CV  
 Products of Conception (POC), specify tissue: \_\_\_\_\_  
 DNA, specify source: \_\_\_\_\_  
 Contact GeneDx by email (WESPrenatal@genedx.com) to discuss a case or to inform us that samples will be sent. Parents' samples must be sent with the proband sample in a Prenatal Kit.

## Clinical Diagnosis:

ICD-10 Codes: \_\_\_\_\_  
 DETAILED MEDICAL RECORDS, CLINICAL SUMMARY, AND FAMILY HISTORY MUST BE SENT WITH REQUISITION FORM AND SAMPLES.

## 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 # \_\_\_\_\_

## Test Requested

959 XomeDxPrenatal Targeted (Trios only)  
 J499 XomeDxPrenatal Comprehensive (Trios only)  
 460 Prenatal GenomeDx (whole genome SNP array) run reflexively with XomeDxPrenatal (Select XomeDxPrenatal option)  
 959 XomeDxPrenatal Targeted  
 J499 XomeDxPrenatal Comprehensive  
 460 Prenatal GenomeDx (whole genome SNP array) run concurrently with XomeDxPrenatal (Select XomeDxPrenatal option)  
 959 XomeDxPrenatal Targeted  
 J499 XomeDxPrenatal Comprehensive

ACMG secondary findings, as discussed in the Informed Consent and Authorization Form, are only returned for the fetus if the XomeDxPrenatal test is completed.

## Biological Parent Sample Information

**Mother:**  Asymptomatic  Symptomatic

First name \_\_\_\_\_ Last name \_\_\_\_\_ DOB \_\_\_\_\_

**Father:**  Asymptomatic  Symptomatic

First name \_\_\_\_\_ Last name \_\_\_\_\_ DOB \_\_\_\_\_

**Other:**  Asymptomatic  Symptomatic

First name \_\_\_\_\_ Last name \_\_\_\_\_ DOB \_\_\_\_\_  
 Relationship to Proband: \_\_\_\_\_

If parental relationships (egg or sperm donor, non-paternity, non-maternity) are inaccurate and/or GeneDx does not receive samples on both biological parents, testing will be canceled.

## Statement of Medical Necessity

This test is medically necessary for the diagnosis or detection of a disease, illness, impairment, symptom, syndrome or disorder. The results will determine my patient's medical management and treatment decisions. The person listed as the Ordering Physician is authorized by law to order the tests(s) requested herein. I confirm that I have provided genetic testing information to the patient and the patient has consented to genetic testing. I have provided genetic counseling to this individual/this individual's family regarding the implications of receiving secondary findings. I have explained the potential benefits and limitations of receiving secondary findings, and I have answered this person's questions.

Medical Professional Signature (required) \_\_\_\_\_ Date \_\_\_\_\_

## 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 \_\_\_\_\_

## For GeneDx use only:

Account # \_\_\_\_\_ Account Name \_\_\_\_\_

First Name \_\_\_\_\_ Last Name \_\_\_\_\_ Date of Birth (mm/dd/yy) \_\_\_\_\_

### Clinical Information (DETAILED MEDICAL RECORDS MUST BE ATTACHED)

Gender/Chromosome Analysis (if known): \_\_\_\_\_ Gestational age: \_\_\_\_\_ (weeks)  IVF Pregnancy  Sperm Donor  Egg Donor

**Clinical diagnosis:** \_\_\_\_\_  
**ICD-10 codes:** \_\_\_\_\_  
**DETAILED MEDICAL RECORDS, CLINICAL SUMMARY AND FAMILY HISTORY MUST BE ATTACHED.**  
**CLINICAL INFORMATION IS CRUCIAL FOR ACCURATE INTERPRETATION OF RESULTS.**

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

### Ultrasound Information/Findings

Date of Ultrasound: \_\_\_/\_\_\_/\_\_\_ GA at time of Ultrasound: \_\_\_\_\_ Weeks \_\_\_\_\_ Days Date of collection: \_\_\_/\_\_\_/\_\_\_ Last menstrual period: \_\_\_/\_\_\_/\_\_\_

<b>General</b>	<input type="checkbox"/> Fetal hydrops	<input type="checkbox"/> Intrauterine growth retardation	<input type="checkbox"/> Polyhydramnios	<input type="checkbox"/> Oligohydramnios
<b>CNS</b>	<input type="checkbox"/> Agenesis of the corpus callosum <input type="checkbox"/> Holoprosencephaly	<input type="checkbox"/> Neural tube defect <input type="checkbox"/> Other brain malformation	<input type="checkbox"/> Dandy-Walker malformation <input type="checkbox"/> Other	<input type="checkbox"/> Ventriculomegaly (hydrocephalus)
<b>Head/Neck</b>	<input type="checkbox"/> Cystic hygroma <input type="checkbox"/> Macrocephaly <input type="checkbox"/> Cleft lip/palate	<input type="checkbox"/> Eye abnormality <input type="checkbox"/> Microcephaly <input type="checkbox"/> Other	<input type="checkbox"/> Increased nuchal translucency _____ mm <input type="checkbox"/> Increased nuchal fold _____ mm	
<b>Cardiac</b>	<input type="checkbox"/> Tetralogy of Fallot <input type="checkbox"/> Other	<input type="checkbox"/> ASD/VSD	<input type="checkbox"/> Congenital heart defect (please specify if known) _____	
<b>Skeletal</b>	<input type="checkbox"/> Club foot <input type="checkbox"/> Upper limb deformity	<input type="checkbox"/> Polydactyly/Syndactyly <input type="checkbox"/> Leg bowing	<input type="checkbox"/> Ectrodactyly <input type="checkbox"/> Short limbs	<input type="checkbox"/> Abnormal ribs and/or small chest circumference <input type="checkbox"/> Bowed or fractured bones <input type="checkbox"/> Other
<b>Uro-Genital</b>	<input type="checkbox"/> Ambiguous genitalia <input type="checkbox"/> Other	<input type="checkbox"/> Renal agenesis	<input type="checkbox"/> Horseshoe kidney	<input type="checkbox"/> Renal cysts
<b>Gastrointestinal</b>	<input type="checkbox"/> Duodenal atresia <input type="checkbox"/> Echogenic bowel	<input type="checkbox"/> Gastroschisis <input type="checkbox"/> Other	<input type="checkbox"/> Congenital diaphragmatic hernia	
<b>Other</b>	<input type="checkbox"/> (please specify) _____			

**Other testing for this pregnancy (summarize or attach reports):**

- Chromosomes/FISH: \_\_\_\_\_
- Array CGH: \_\_\_\_\_
- Fetal echo: \_\_\_\_\_
- Fetal MRI/CT: \_\_\_\_\_
- Other relevant results (clinical or research): \_\_\_\_\_

**Previous pregnancy or family history of:**

- ONTD
- Aneuploidy (please specify) \_\_\_\_\_
- Genetic disorders (please explain below)
- Other: \_\_\_\_\_ (Attach Pedigree if Available)

**Please explain pregnancy or family history:**

- AMA
- Abnormal maternal serum screen for \_\_\_\_\_
- Abnormal NIPS for \_\_\_\_\_

### Billing Information

**Institutional Bill**

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

BILLING STAMP

**Patient Bill**

I understand that my credit card will be charged the full amount for the testing.

Name as it appears on card \_\_\_\_\_  
 Account Number \_\_\_\_\_ Expiration date \_\_\_\_\_ CVC \_\_\_\_\_  
 Signature \_\_\_\_\_ Date \_\_\_\_\_

**Please bill my credit card (all major cards accepted)**

Amount \_\_\_\_\_  
 MasterCard  Visa  Discover  American Express

Account # \_\_\_\_\_ Account Name \_\_\_\_\_

First Name \_\_\_\_\_

Last Name \_\_\_\_\_

Date of Birth (mm/dd/yy) \_\_\_\_\_

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](http://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](http://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](http://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](http://www.genome.gov/10002077).

Account # \_\_\_\_\_ Account Name \_\_\_\_\_

First Name \_\_\_\_\_

Last Name \_\_\_\_\_

Date of Birth (mm/dd/yy) \_\_\_\_\_

## 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.

## WES Secondary Findings & Opt-Out

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

**Secondary findings** are variants, identified by the 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 whole 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 coding exons of the genes (for which a minimum of 10X coverage was achieved by the XomeDx, XomeDxPlus, XomeDxPrenatal or XomeDxXpress test), as recommended by the ACMG.

## What will be reported for relatives (if tested with 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 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 whole exome 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](mailto: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.**