

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) _____
Sample Type:
 CVS Amniotic Fluid Fetal Blood (PUBS)
 Cultured CV Cultured amniocytes
 Products of Conception (POC), specify tissue: _____
 DNA, Specify Source: _____
 Maternal blood for MCC (please send for ALL studies)
 Paternal blood (please send for ALL molecular studies)
 Father's: Last Name _____ First Name _____ DOB _____
If other samples submitted
 Relationship to fetus _____ Name _____ Sample type _____
Clinical Diagnosis: _____ **ICD-10 Codes:** _____

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 # _____

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.

Signature of Physician or Other Authorized NPI Provider (required) _____ Date _____

Patient Consent (sign here)

I have read the attached 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. **More information is available on our website: www.genedx.com**
 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** _____

PATIENT STATUS – ONE MUST BE CHECKED: Hospital Inpatient Hospital Outpatient Not a Hospital Patient Hospital Patient Date of Discharge: _____

Payment Options

Insurance Bill Referral/Prior Authorization # _____
Please attach copy of Referral/authorization
 Insurance Carrier _____ Policy Name _____ Hold sample for Benefit Investigation (only if OOP cost is >\$100) GeneDx Benefit Investigation # _____
 Insurance ID # _____ Group # _____ Name of Insured _____ Date of Birth _____ Insurance Address _____ City _____ State _____ Zip _____
 Secondary Insurance Carrier Name _____ Insurance ID# _____ Group # _____ Name of Insured _____ Date of Birth _____ Relationship to Insured Child Spouse Self Other _____
 Relationship to Insured Child Spouse Self Other _____
Please include a copy of the front and back of the patient's insurance card (include secondary when applicable)
If you would like to expedite an assessment of your possible eligibility for GeneDx's financial assistance program (FAP), please provide the number of your household members _____ and the annual income of your household \$ _____. GeneDx may require additional information from you to complete an application for GeneDx's financial assistance program.
 I represent that I am covered by insurance and authorize GeneDx, Inc. to give my designated insurance carrier, health plan, or third party administrator (collectively "Plan") the information on this form and other information provided by my health care provider necessary for reimbursement. I authorize Plan benefits to be payable to GeneDx. I understand that GeneDx will attempt to contact me if my out-of-pocket responsibility will be greater than \$100 per test (for any reason, including co-insurance and deductible, or non-covered services). If GeneDx is unsuccessful in its attempts to contact me, I understand that it will be my responsibility to contact GeneDx to determine my out-of-pocket cost and to pay my out-of-pocket responsibility. I will cooperate fully with GeneDx by providing all necessary documents needed for Plan billing and appeals. I understand that I am responsible for sending GeneDx any and all of the money that I receive directly from my Plan in payment for this test. Reasonable collection and/or attorney's fees, including filing and service fees, shall be assessed if the account is sent to collection but said fees shall not exceed those permitted by state law. I permit a copy of this authorization to be used in place of the original.
Patient Signature (required) _____ **Date** _____

Institutional Bill

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

Patient Bill

Amount _____
 If I have insurance coverage for this testing, I am electing to be treated as a self-pay patient for this testing. As such, I agree that neither GeneDx nor I will submit a claim to my insurance for this testing.
Please bill my credit card for the full amount stated above (all major cards accepted)
 MasterCard Visa Discover American Express
 Name as it appears on card _____
 Account Number _____ Expiration date _____ CVC _____
Signature _____ **Date** _____
For GeneDx Use Only

Prenatal Genetics Testing Requisition Form

Account # _____ Account Name _____

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

Clinical Information (DETAILED MEDICAL RECORDS MUST BE ATTACHED)

Fetal Gender: M F Discrepant: _____ Chromosome Analysis (if known): _____ Diagnosis: _____
ICD-10 codes: _____ IVF Pregnancy Maternal Diabetes mellitus Egg Donor Sperm Donor

Ultrasound Information/Findings

Date of Ultrasound: ___/___/___ GA at time of Ultrasound: _____ Weeks _____ Days Date of collection: ___/___/___ Last menstrual period: ___/___/___

| | | | | | |
|-------------------------|--|--|--|--|--|
| General | <input type="checkbox"/> Fetal hydrops | <input type="checkbox"/> Intrauterine growth retardation | | | |
| CNS | <input type="checkbox"/> Agenesis of the corpus callosum | <input type="checkbox"/> Neural tube defect | <input type="checkbox"/> Lissencephaly | <input type="checkbox"/> Other brain malformation: _____ | |
| | <input type="checkbox"/> Ventriculomegaly (hydrocephalus) | <input type="checkbox"/> Holoprosencephaly | <input type="checkbox"/> Dandy Walker malformation | | |
| Head/Neck | <input type="checkbox"/> Increased nuchal translucency _____mm | <input type="checkbox"/> Cystic hygroma | <input type="checkbox"/> Macrocephaly | <input type="checkbox"/> Eye abnormality | |
| | <input type="checkbox"/> Increased nuchal fold _____mm | <input type="checkbox"/> Cleft lip/palate | <input type="checkbox"/> Microcephaly | | |
| Cardiac | <input type="checkbox"/> Tetralogy of Fallot | <input type="checkbox"/> ASD | <input type="checkbox"/> VSD | <input type="checkbox"/> Other cardiac anomaly (specify if known): _____ | |
| Skeletal | <input type="checkbox"/> Upper limb deformity | <input type="checkbox"/> Polydactyly | <input type="checkbox"/> Club foot | <input type="checkbox"/> Fractured bones | <input type="checkbox"/> Abnormal ribs |
| | <input type="checkbox"/> Ectrodactyly | <input type="checkbox"/> Syndactyly | <input type="checkbox"/> Short limbs: (%ile____) | <input type="checkbox"/> Bowed bones | <input type="checkbox"/> Small chest circumference (TC ratio:____) |
| Uro-Genital | <input type="checkbox"/> Ambiguous genitalia | <input type="checkbox"/> Renal agenesis | <input type="checkbox"/> Horseshoe kidney | <input type="checkbox"/> Renal cysts | <input type="checkbox"/> Other renal abnormality: _____ |
| Gastrointestinal | <input type="checkbox"/> Duodenal atresia | <input type="checkbox"/> Gastroschisis | <input type="checkbox"/> Diaphragmatic hernia | <input type="checkbox"/> Omphalocele | <input type="checkbox"/> Other malformation: _____ |
| Other | | | | | |

Previous pregnancy or family history of:
 ONTD Genetic disorders (please explain below)
 Down Syndrome Other: _____ (Attach Pedigree if Available)

Please explain pregnancy or family history:
 AMA Abnormal maternal serum screen for _____
 Abnormal NIPT for _____

Please choose test(s) and provide clinical information in the appropriate section above. GeneDx performs maternal contamination studies for prenatal tests, so a maternal blood sample is requested for prenatal tests. All tests will be performed concurrently unless order of testing is specified.

Prenatal Genetic Testing Menu

Sequencing Tests for Fetuses with Abnormal Ultrasound Findings

- Noonan Spectrum and RASopathies**
 357 Prenatal Noonan Spectrum Disorders Panel
- Skeletal and Limb Abnormalities**
 937 Prenatal Limb Abnormalities Panel *
 949 Prenatal Skeletal Dysplasia Panel *
 3383 Campomelic Dysplasia * (SOX9)
 738 Cornelia de Lange Syndrome * (NIPBL-related)
 408 Duane-Radial-Ray Syndrome * (DRRS; SALL4)
 407 Ectrodactyly-ED-Clefting/Hay-Wells Syndrome (TP63, p63)
 2363 Holt-Oram Syndrome (TBX5)
 2523 Townes-Brocks Syndrome (SALL1)
- Brain Malformations**
 J803 Prenatal Joubert Syndrome and Related Disorders Panel
 J793 Prenatal Lissencephaly Panel
 J802 Prenatal Pontocerebellar Hypoplasia Panel
 2373 Holoprosencephaly Panel*
 2553E X-linked Hydrocephalus/MASA/CRASH Syndrome (LICAM): FEMALES *
 2553 X-linked Hydrocephalus/MASA/CRASH Syndrome (LICAM): MALES
- Disorders of Sex Differentiation**
 J719-6 Prenatal 46, XY Disorders of Sex Development Panel
 702 5-Alpha Reductase Deficiency * (SRD5A2)
 2201 Androgen Insensitivity Syndrome (AR)
 907 Antley Bixler Syndrome (POR)
 J720 SRY Present/Absent Testing
 409 XY Female Gonadal Dysgenesis (SRY)
 746 46, XY Gonadal Dysgenesis (NR5A1-related)
- Other Panels and Full Gene Testing for Multiple Congenital Anomalies**
 663 Adrenal Hypoplasia Congenita (AHC), X-linked (NR0B1)
 428 Anophthalmia/Microphthalmia: Sequencing (SOX2 *, OTX2 * and VSX2)
 2262 CHARGE Syndrome (CHD7)
 2503 Smith-Lemli-Opitz Syndrome * (DHCR7)
 934 Tuberous Sclerosis * (TSC1 and TSC2)

* Entire panel/test or specific gene includes deletion/duplication testing. For SRD5A2, DHCR7, and skeletal dysplasias, del/dup testing only completed if sequencing identifies a single pathogenic variant in AR disorders.

Cytogenetics and Biochemical Tests

- Cytogenetic Tests**
 3582 Rapid aneuploidy FISH (13, 18, 21, X, Y)
 410 Prenatal Targeted Chromosomal Microarray (CMA)
 Run as REFLEX test if _____ test(s) are negative
 460 Prenatal/POC Whole Genome Chromosomal Microarray (CMA)
 J542 FISH Follow-up after CMA (Test 410 or 460)
 Please specify chromosomal region: 1. _____ 2. _____
- Chromosome Analysis (select specimen type)**
 2136 Karyotype: Amniotic Fluid
 A587 Karyotype: CVS
 1053 Karyotype: Products of Conception (POC)
 0559 Karyotype: Peripheral Blood
- Biochemical and Other Tests**
 2122 Amniotic fluid AFP (1952-I automatic reflex to AChE testing if AF-AFP is elevated)
 437 Zygosity Testing

Prenatal Testing for Known Familial Mutation(s)

- Relative tested at GeneDx (provide ID number below)
 Relative tested at another lab (Please send positive control sample and copy of report)
 Positive control samples (no report issued for patient/relative)

GeneDx ID # _____ First name _____ Last name _____
 Gene or locus _____ Mutations(s) _____
 Relationship to fetus _____

Please write any special instructions, i.e., in which order to perform tests

- Notes:**
 (1) If sufficient fetal material is submitted, most testing can be performed concurrently. If no other instructions are given, all tests will be performed concurrently.
 (2) If you choose to have the testing done in a particular order ('reflex testing'), indicate the order of tests by numbering the tests (Example: (1) Chromosome analysis, CVS;
 (2) Prenatal targeted array (if chromosomes normal); (3) Noonan syndrome testing (if array normal)).

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 certain changes in DNA affecting the structure or number of chromosomes. Genetic testing is a laboratory test that tries to identify these 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 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.

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 by 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 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.

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.

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, the limitations of genetic testing, as well as information about how specimens and information are stored and used.