Prenatal Genetics Testing Requisition Form

GeneDx an OPKO Health Company

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), specifiy tissue:_____
❒ DNA, Specify Source:_____
❒ Maternal blood for MCC (please send for ALL studies)
❒ Paternal blood (for aCGH studies only)

If other samples submitted

Relationship to fetus Name Sample type

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.

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 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

Payment Options

❒ Insurance Bill

PATIENT STATUS – ONE MUST BE CHECKED ❒ Hospital Inpatient ❒ Hospital Outpatient ❒ Not a Hospital Patient

Referral/Prior Authorization # _____
Please attach copy of Referral/authorization

Insurance Carrier Policy Name 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 GeneDx to inform my Plan of my test result only if test results are required for preauthorization of or payment for reflex/additional testing. 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 _____

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

Please bill my credit card (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

GeneDx an OPKO Health Company

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) Diagnosis:_____

ICD-10 codes:_____ ❒ IVF Pregnancy ❒ Maternal Diabetes mellitus ❒ Egg Donor

Ultrasound Information/Findings

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

General	❒ Fetal hydrops	❒ Intrauterine growth retardation		
CNS	❒ Agenesis of the corpus callosum ❒ Ventriculomegaly (hydrocephalus)	❒ Neural tube defect ❒ Holoprosencephaly	❒ Dandy Walker malformation ❒ Other Brain malformation	
Head/Neck	❒ Cystic hygroma ❒ Macrocephaly ❒ Cleft lip/palate	❒ Eye abnormality ❒ Microcephaly	❒ Increased nuchal translucency _____mm ❒ Increased nuchal fold _____mm	
Cardiac	❒ Tetralogy of Fallot	❒ Congenital heart defect *(please specify if known)*	❒ ASD/VSD	
Skeletal	❒ Club foot ❒ Upper limb deformity	❒ Polydactyly/Syndactyly ❒ Leg bowing	❒ Ectrodactyly ❒ Short limbs	❒ Abnormal ribs and/or small chest circumference ❒ Bowed or fractured bones
Uro-Genital	❒ Ambiguous genitalia ❒ Other renal abnormality	❒ Renal agenesis	❒ Horseshoe kidney	❒ Renal cysts
Gastrointestinal	❒ Duodenal atresia ❒ Other malformation	❒ Gastroschisis	❒ Cong. diaphragmatic hernia	
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 (*BRAF, HRAS, KRAS, MAP2K1, MAP2K2, NRAS, PTPN11, RAF1, RIT1, SOS1, SHOC2* (*S2G* mutation only))

Disorders of Sex Differentiation

❒ 907 Antley Bixler Syndrome (*POR*)
❒ 2201 Androgen Insensitivity Syndrome (*AR*)
❒ 409 XY Female Gonadal Dysgenesis (*SRY*)
❒ 702 5-Alpha Reductase Deficiency * (*SRD5A2*)
❒ 746 46, XY Gonadal Dysgenesis (*NR5A1*-related)
❒ J719-6 Prenatal 46,XY Disorders of Sex Development Panel (10 genes)

Skeletal and Limb Abnormalities

❒ 937 Prenatal Limb Abnormalities Panel * (*NIPBL, SALL1, SALL4, TBX5, TP63*)
❒ 949 Prenatal Skeletal Dysplasia Panel * (23 genes)
❒ 738 Cornelia de Lange Syndrome * (*NIPBL*-related)
❒ 2523 Townes-Brocks Syndrome (*SALL1*)
❒ 408 Duane-Radial-Ray Syndrome * (*DRRS; SALL4*)
❒ 2363 Holt-Oram Syndrome (*TBX5*)
❒ 407 Ectrodactyly-ED-Clefting/Hay-Wells Syndrome (*TP63*, p63)
❒ 3383 Campomelic Dysplasia * (*SOX9*)

Other Panels and Full Gene Testing for Multiple Congenital Anomalies

❒ 428 Anophthalmia/Microphthalmia: Sequencing (*SOX2 *, OTX2 * and VSX2*)
❒ 2373 Holoprosencephaly * (*SHH, ZIC2, SIX3, TGIF*)
❒ 934 Tuberous Sclerosis * (*TSC1* and *TSC2*)
❒ 2262 CHARGE Syndrome (*CHD7*)
❒ 2503 Smith-Lemli-Opitz Syndrome * (*DHCR7*)
❒ 2553 X-linked Hydrocephalus/MASA/CRASH Syndrome (*L1CAM*): MALES
❒ 2553E X-linked Hydrocephalus/MASA/CRASH Syndrome (*L1CAM*): FEMALES *
❒ 663 Adrenal Hypoplasia Congenita (AHC), X-linked (*NR0B1*)

Brain Malformations

❒ J793 Prenatal Lissencephaly Panel (24 genes)
❒ J802 Prenatal Pontocerebellar Hypoplasia Panel (18 genes)
❒ J803 Prenatal Joubert Syndrome and Related Disorders Panel (25 genes)

*: 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

❒ 433 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._____
❒ 455 FISH, DiGeorge syndrome

Chromosome Analysis (by specimen type)

❒ 4341 Amniotic Fluid
❒ 4342 CVS
❒ 4346 PUBS
❒ 4344 Products of Conception (POC)
❒ 4343 Peripheral Blood
❒ 4345 Peripheral Blood (rule out mosaicism)

Biochemical and Other Tests

❒ 1122-1 Amniotic fluid AFP (1952-1 automatic reflex to AChE testing if AF-AFP is elevated)
❒ 437 Zygosity Testing

**To provide CMA testing on patients from New York state, routine cytogenetics must also be performed.

❒ New York clients, check here to indicate that the provider accepts this responsibility.

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)).

Informed Consent and Authorization Form

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. 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.

Informed Consent and Authorization Form

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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.

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 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.