# GeneDx — Cytogenetics Test Requisition Form

an OPKO Health Company

*For our PRENATAL cytogenetic and molecular testing, please use the prenatal submission form. For other testing services, please use the specific submission forms also available at www.genedx.com/forms.*

## 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

## 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 #

## Sample Information

Medical record #  Specimen ID #  Date sample obtained (mm/dd/yy)

### Sample Type

❒ Blood in EDTA (for array CGH; purple top - single tube of 2-5mL)

❒ Blood in sodium heparin (for FISH and chromosome analysis; green top-single tube of 2-5mL)

❒ DNA: source _____ (tissue?) / concentration_____ (µg/mL)

❒ Buccal Swab

***If other samples submitted***

Relationship to patient  Name  Sample type

Relationship to patient  Name  Sample type

**Clinical Diagnosis:**_____ **ICD-10 Codes:**_____

**Age at Initial Presentation:**_____

## 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 Provider 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 Estimated 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

Relationship to Insured ❒ Child ❒ Spouse ❒ Self ❒ Other _____

Secondary Insurance Carrier Name  Insurance ID#  Group #  Name of Insured  Date of Birth

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)***

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 estimated 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**

# Cytogenetics Test Requisition Form

GeneDx an OPKO Health Company

Account # Account Name

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

## Test requested

**Please select choice and provide clinical information below**

❒ 910 GenomeDx: Whole-Genome Chromosomal Microarray (CMA)

❒ J542 FISH Follow-up after GenomeDx (Test 910)

Please specify chromosomal region: 1._____ 2._____

❒ 0559 Chromosome Analysis, Peripheral blood (routine study)

❒ Rule out mosaicism for _____

(Must indicate suspected mosaic chromosomal abnormality)

❒ 336 FISH (Please specify chromosomal region or disorder)

1._____ 2._____

❒ Parental Follow-up Testing for copy number variant(s)

❒ 905 Known Familial Deletion/Duplication Testing

Gene(s):_____ Mutation(s): _____

Proband Name: _____

Proband GeneDx Acc#:_____ Relationship to proband: _____

❒ Positive control included: **Positive control is required if previous test was performed at another lab.**

❒ Family Member Test Report included - A clear copy of the test report on the mutation positive family member is recommended if previous test was performed at another lab.

**Expedited Testing.** I would like expedited testing for the following reason:

❒ Pregnancy (gestational age: _____ weeks) ❒ Transplantation ❒ Other: _____

## Patient Clinical Information:

**ICD-10 Codes:** _____

Diagnosis:_____

Dysmorphic features: _____

Congenital heart disease: _____

Other medical problems: _____

Suspected syndrome(s): _____

Previous cytogenetics:_____ (attach copy)

Clinical and family history: _____

**This information is crucial for interpretation of array CGH results. Please check all that apply (if standard karyotype done, list below):**

**Perinatal History:**
- ❒ Prematurity
- ❒ IUGR
- ❒ Oligohydramnios
- ❒ Polyhydramnios
- ❒ Other:_____

**Growth:**
- ❒ Failure to thrive
- ❒ Overgrowth
- ❒ Short stature
- ❒ Other:_____

**Development:**
- ❒ Fine motor delay
- ❒ Gross motor delay
- ❒ Speech delay
- ❒ Other:_____

**Cognitive:**
- ❒ Learning disability
- ❒ Mental retardation

List IQ/DB, if known:_____
- ❒ Other:_____

**Behavioral:**
- ❒ Autistic features
- ❒ Autism spectrum disorder
- ❒ Oppositional-defiant disorder
- ❒ Obsessive-compulsive disorder
- ❒ Other:_____

**Neurological:**
- ❒ Ataxia
- ❒ Dystonia
- ❒ Chorea
- ❒ Hypotonia
- ❒ Neural tube defect
- ❒ Seizures
- ❒ Spasticity
- ❒ Structural brain anomaly
- ❒ Other:_____

**Cardiac:**
- ❒ ASD
- ❒ AV canal defect
- ❒ Coarctation of aorta
- ❒ Hypoplastic left heart
- ❒ Tetralogy of Fallot
- ❒ VSD
- ❒ Other:_____

**Craniofacial:**
- ❒ Cleft lip
- ❒ Cleft palate
- ❒ Coloboma of eye
- ❒ Craniosynostosis
- ❒ Dysmorphic facial features
- ❒ Ear malformation
- ❒ Macrocephaly
- ❒ Microcephaly

List HC, if known:_____
- ❒ Other:_____

**Cutaneous:**
- ❒ Hyperpigmentation
- ❒ Hypopigmentation
- ❒ Other:_____

**Musculoskeletal:**
- ❒ Contractures
- ❒ Club foot
- ❒ Diaphragmatic hernia
- ❒ Limb anomaly
- ❒ Polydactyly
- ❒ Scoliosis
- ❒ Syndactyly
- ❒ Vertebral anomaly
- ❒ Other:_____

**Gastrointestinal:**
- ❒ Gastroschisis
- ❒ Hirschsprung disease
- ❒ Omphalocele
- ❒ Pyloric stenosis
- ❒ Tracheoesophageal fistula
- ❒ Other:_____

**Genitourinary:**
- ❒ Ambiguous genitalia
- ❒ Hydronephrosis
- ❒ Hypospadias
- ❒ Kidney malformation
- ❒ Undescended testis
- ❒ Urethral obstruction
- ❒ Other:_____

**Family History:**
- ❒ Parents with >=2 miscarriages
- ❒ Other relatives with similar clinical history *(please explain below)*

# Informed Consent

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.**