XomeDx Test Requisition Form

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)

❒ Blood in EDTA (5-6 mL in lavender top tube)

❒ DNA (>20 ug): Tissue source _____ concentration ___ (ug/ml) Vol ___(ul)

❒ Oral Rinse (At least 30 mL of Scope oral rinse in a 50 mL centrifuge tube)

❒ Dried Blood Spots (2 cards) - Not accepted for any testing with a del/dup component

❒ Buccal Swab

❒ 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 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

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 do not wish to receive ACMG 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

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

XomeDx Clinical Information Form

GeneDx an OPKO Health Company

Account # Account Name

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

Clinical diagnosis: _____

ICD-10 codes: _____

DETAILED MEDICAL RECORDS, CLINICAL SUMMARY, PICTURES 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.

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/Ophthalmalogic/Auditory
- ❒ Cataracts
- ❒ Cleft lip/palate
- ❒ Coloboma of eye
- ❒ CPEO (opthalmoplegia)
- ❒ 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

Hemotologic/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: _____

_____

_____

_____

_____

XomeDx Test Requisition Form

Account # Account Name

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

XomeDx Testing Options

❒ 561a XomeDx Trio† ❒ 690a XomeDx*Plus* Trio† (XomeDx + 690c mtDNA)
❒ 561e XomeDx Duo† ❒ 690e XomeDxPlus Duo† (XomeDx + 690c mtDNA)
❒ 561b XomeDx Proband ❒ 690b XomeDx*Plus* Proband (XomeDx + 690c mtDNA)

†If a Trio or Duo test is ordered, please fill out the Biological Parent Sample Information section below.

XomeDx Reflex Testing

❒ Test code(s): _____

Reflex to (choose one):
❒ 561a XomeDx Trio† ❒ 690a XomeDx*Plus* Trio† (XomeDx + 690c mtDNA)
❒ 561e XomeDx Duo† ❒ 690e XomeDxPlus Duo† (XomeDx + 690c mtDNA)
❒ 561b XomeDx Proband ❒ 690b XomeDx*Plus* Proband (XomeDx + 690c mtDNA)

Custom XomeDx*Slice* Testing Options

❒ 706 XomeDx*Slice* (1-150 genes, Proband only)
Approved Slice ID: _____

❒ J757 XomeDx*Slice* Xpanded (>150 genes, Proband or Trio)†
Approved Slice ID: _____
† If a Trio test is ordered, please fill out the Biological Parent Sample Information section below.

Skin Disorder Slices

❒ 707 XomeDx*Slice* Epidermolysis Bullosa (EB) (Proband Only)
❒ 708 XomeDx*Slice* Congenital Ichthyosis (Proband Only)

Testing for Known Familial Variants Identified through XomeDx

❒ 9011 Testing for ONE known familial variant
❒ 9012 Testing for TWO known familial variants in the same gene Gene(s): _____
❒ 905 Testing for one known familial deletion or duplication Variant(s): _____

Proband Name: _____ Proband GeneDx Acc#: _____ Relationship to Proband: _____

Reanalysis of XomeDx Testing Options

These test options are only appropriate if the patient previously had a XomeDx test (full exome analysis) at GeneDx. We recommend waiting at least one year from original/prior analysis before ordering a Reanalysis.

❒ 660 XomeDx First Time Reanalysis (no charge)
❒ 947 XomeDx Subsequent Reanalysis (charged)
Is there new clinical information available? ❒ Yes ❒ No

Biological Parent Sample Information

Mother: ❒ Not available ❒ To be sent later* ❒ At GeneDx
First name Last name DOB ❒ Asymptomatic ❒ Symptomatic

Father: ❒ Not available ❒ To be sent later* ❒ At GeneDx
First name Last name DOB ❒ Asymptomatic ❒ Symptomatic

Other: ❒ Not available ❒ To be sent later* ❒ At GeneDx
First name Last name DOB ❒ Asymptomatic ❒ Symptomatic

***ADDITIONAL SAMPLES MUST BE RECEIVED WITHIN 3 WEEKS. Ordered test codes may require adjusting to appropriately correspond with relative samples received. A change in the ordered test will impact billing, including prior benefits investigations.**

ACMG secondary findings, as discussed in the Informed Consent and Authorization Form, are only returned for the patient if an XomeDx test (full exome analysis) is completed.

ES/WGS 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 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.

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

Exome/Genome Sequencing Secondary Findings & Opt-Out

As many different genes and conditions are analyzed in the GenomeSeqDx, XomeDx, XomeDx*Plus*, XomeDx*Prenatal* and XomeDx*Xpress* 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, XomeDx*Plus*, XomeDx*Prenatal* and XomeDx*Xpress* 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, XomeDx*Plus*, XomeDx*Prenatal* or XomeDx*Xpress* test), as recommended by the ACMG.

What will be reported for relatives (if tested with GenomeSeqDx, XomeDx, XomeDx*Plus*, XomeDx*Prenatal* or XomeDx*Xpress*)

- The presence or absence for any secondary findings reported for the proband will be provided for all relatives tested by GenomeSeqDx, XomeDx, XomeDx*Plus*, XomeDx*Prenatal* or XomeDx*Xpress*.

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.

GeneDx
an OPKO Health Company

A. Notifier:

B. Patient Name: **C. Identification Number:**

Advance Beneficiary Notice of Noncoverage (ABN)

NOTE: If Medicare doesn't pay for **D.**_____ below, you may have to pay. Medicare does not pay for everything, even some care that you or your health care provider have good reason to think you need. We expect Medicare may not pay for the **D.**_____ below.

D.	E. Reason Medicare May Not Pay:	F. Estimated Cost

WHAT YOU NEED TO DO NOW:

- Read this notice, so you can make an informed decision about your care.
- Ask us any questions that you may have after you finish reading.
- Choose an option below about whether to receive the **D.**_____ listed above.
  Note: If you choose Option 1 or 2, we may help you to use any other insurance that you might have, but Medicare cannot require us to do this.

G. OPTIONS: Check only one box. We cannot choose a box for you.

☐ **OPTION 1.** I want the **D.**_____ listed above. You may ask to be paid now, but I also want Medicare billed for an official decision on payment, which is sent to me on a Medicare Summary Notice (MSN). I understand that if Medicare doesn't pay, I am responsible for payment, but **I can appeal to Medicare** by following the directions on the MSN**.** If Medicare does pay, you will refund any payments I made to you, less co-pays or deductibles.

☐ **OPTION 2.** I want the **D.**_____ listed above, but do not bill Medicare. You may ask to be paid now as I am responsible for payment. **I cannot appeal if Medicare is not billed**.

☐ **OPTION 3.** I don't want the **D.**_____ listed above. I understand with this choice I am **not** responsible for payment, and **I cannot appeal to see if Medicare would pay.**

H. Additional Information:

This notice gives our opinion, not an official Medicare decision. If you have other questions on this notice or Medicare billing, call **1-800-MEDICARE** (1-800-633-4227/**TTY:** 1-877-486-2048). Signing below means that you have received and understand this notice. You also receive a copy.

I. Signature:	J. Date:

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