XOMEDX*PRENATAL* TEST REQUISITION FORM

GeneDx an OPKO Health Company

PATIENT INFORMATION

First name | Last name

Sex ◯ Male ◯ Female
Gender identification (optional): _____ | Date of birth (mm/dd/yy)

Ancestry ◯ White/Caucasian ◯ Hispanic ◯ Black/African American ◯ Native American ◯ East Asian ◯ South Asian ◯ Middle Eastern ◯ Ashkenazi Jewish ◯ Other: _____

Email

Address

City | State | Zip code

Primary phone

ACCOUNT INFORMATION

Account number | **Account name**

Phone | Fax

Address | City

State | Zip code | Country

Ordering provider

Name | Role/Title

Phone | NPI

Email address (for report access)

Reporting Preference: ◯ Portal ◯ Fax ◯ Email ◯ Care Evolve *If unmarked, we will use the account's default preferences or fax to new clients.*

Additional Reporting Providers ◯ Same as ordering provider

Name | Role/Title

Phone | NPI

Email address (for report access)

Additional clinical or laboratory contact (optional)

Name | Email address (for report access)

SEND ADDITIONAL REPORT COPIES TO

Healthcare provider/Acct # | Fax #/Email

SAMPLE INFORMATION

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

◯ Chorionic villi (CV) ◯ Amniotic Fluid ◯ Fetal Blood (PUBS)
◯ Cultured CV ◯ Cultured amniocytes
◯ 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.

ICD-10 codes (required): | **Clinical diagnosis:** | **Age at initial presentation:**

STATEMENT OF MEDICAL NECESSITY

By submission of this test requisition and accompanying sample(s), I: (i) authorize and direct GeneDx to perform the testing indicated; (ii) certify that the person listed as the ordering provider is authorized by law to order the test(s) requested; (iii) certify that any custom panel and/or ordered test(s) requested on this test requisition form are reasonable and medically necessary for the diagnosis and/or treatment of a disease, illness, impairment, symptom, syndrome or disorder; (iv) the test results will determine my patient's medical management and treatment decisions of this patient's condition on this date of service; (v) have obtained this patient's and relatives', when applicable, written informed consent to undergo any genetic testing requested; and (vi) that the full and appropriate diagnosis code(s) are indicated to the highest level of specificity.

Signature of provider (required) **Date**

PATIENT CONSENT

By signing this form I acknowledge as the patient that I have read the attached informed consent document and that I authorize GeneDx to perform genetic testing as described. For tests that evaluate data from multiple family members concurrently, such as me and my spouse or partner, results from these family members may be included in a single comprehensive report that will be made available to all tested individuals and their health care providers I have been informed that GeneDx may contact me or my healthcare provider about research opportunities in the future. For the insurance bill, I understand and authorize GeneDx to share information with the designated insurance carrier for reimbursement. I understand that GeneDx will attempt to contact me if my estimated out-of-pocket responsibility will be greater than $100 per test. If GeneDx is unsuccessful in its attempts to contact me, it will be my responsibility to contact GeneDx to determine and pay the out-of-pocket cost. More information, including the GeneDx Notice of Privacy Policies, is available on GeneDx's website: www.genedx.com

◯ By checking this box, I confirm that I am a New York state resident, and I give permission for GeneDx to retain any remaining sample longer than 60 days after the completion of testing, and to be used as a de-identified sample for test development and improvement, internal validation, quality assurance, and training purposes. Otherwise, New York law requires GeneDx to destroy my sample after 60 days, and it cannot be used for the studies listed above.

◯ 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 ACMG secondary findings.

Signature of Patient/Guardian (required) Date | Signature of Relative A/Guardian (required) Date | Signature of Relative B/Guardian (required) Date

PATIENT STATUS – ONE MUST BE CHECKED: ◯ **Hospital outpatient** ◯ **Hospital inpatient** **Date of discharge:** _____ ◯ **Not a hospital patient**

◯ **Patient Bill** | Amount _____

If Patient Bill is selected, I am electing to be treated as a self-pay patient for this testing. I agree that neither GeneDx nor I will submit a claim to my insurance for this testing, if I have insurance. GeneDx will send an invoice to the patient listed above.

◯ **GeneDx Affiliate Code:**

◯ **Institutional Bill**

GeneDx account # | Hospital/Lab name

Place sticker/stamp here

CLINICAL INFORMATION

Account #	Account Name

First Name	Last Name	Date of Birth

CLINICAL INFORMATION (DETAILED MEDICAL RECORDS MUST BE ATTACHED)

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

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: ___/___/___

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

_____

Signature of provider (required) Date

XOMEDX*PRENATAL* TEST REQUISITION FORM

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TEST REQUESTED

TEST CODE	TEST NAME
◯ 959	XomeDx*Prenatal* Targeted (Trios only)
◯ J499	XomeDx*Prenatal* Comprehensive (Trios only)
◯ 460	Prenatal GenomeDx (whole genome SNP array) run reflexively with XomeDx*Prenatal* (Select XomeDx*Prenatal* option) ◯ 959 XomeDx*Prenatal* Targeted ◯ J499 XomeDx*Prenatal* Comprehensive
◯ 460	Prenatal GenomeDx (whole genome SNP array) run concurrently with XomeDx*Prenatal* (Select XomeDx*Prenatal* option) ◯ 959 XomeDx*Prenatal* Targeted ◯ J499 XomeDx*Prenatal* Comprehensive

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

BIOLOGICAL PARENT SAMPLE INFORMATION

Mother:

First name _____ Last name _____ DOB _____ ◯ Asymptomatic ◯ Symptomatic

Father:

First name _____ Last name _____ DOB _____ ◯ Asymptomatic ◯ Symptomatic

Other:

First name _____ Last name _____ DOB _____ ◯ Asymptomatic ◯ Symptomatic

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.

INFORMED CONSENT

Account #	Account Name

First Name	Last Name	Date of Birth

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 complete gene lists, 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.

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 heath 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. The authorization is optional, and testing will be unaffected if I do not check the box for the New York authorization language. No tests other than those authorized shall be performed on the biological sample.

Database Participation

De-identified heath 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 database, 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 healthcare 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 {my/my child} or {my/my child's} heirs.

INFORMED CONSENT

Account #	Account Name

First Name	Last Name	Date of Birth

WES Secondary Findings & Opt-Out

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

What will be reported for relatives (if tested with 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 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 whole exome sequencing will not be reported.