PREVENTION GENETICS

PREVENTIONGENETICS USE ONLY

Test information is available on our website:
www.PreventionGenetics.com

All testing must be ordered by a qualified Healthcare Provider

THIS FORM MUST ACCOMPANY ALL SPECIMENS

DIAGNOSTIC PGxome® - WHOLE EXOME SEQUENCING
HEALTHCARE PROVIDER STATEMENT

This Statement is required, and applies to Whole Exome Sequencing tests for diagnostic purposes.

PATIENT INFORMATION

LAST (FAMILY) NAME	FIRST NAME	MI	DATE OF BIRTH
			____ / ____ / ____ MONTH DAY YEAR

FAMILY MEMBERS

If a family (duo, trio, etc.) is being tested, please provide family member information:

FAMILY MEMBER'S NAME	RELATIONSHIP
FAMILY MEMBER'S NAME	RELATIONSHIP
FAMILY MEMBER'S NAME	RELATIONSHIP

The following information should be used as a guide to provide informed consent to the patient and/or patient's family. We require the consenting Healthcare Provider sign below. Testing must be ordered by a qualified Healthcare Provider.

PURPOSE

- The purpose of this test is to find the underlying genetic cause for the patient's health condition using Whole Exome Sequencing (WES).

ABOUT PGXOME TEST

- This test involves the sequencing of thousands of genes at the same time, whereas many other genetic tests look at only one gene or a small group of genes. The way we perform the exome test is through a procedure called Next Generation Sequencing (NGS). We confirm important results with another type of sequencing called Sanger sequencing. Copy number variants (CNVs), also known as deletions/duplications, are also detected from NGS data. All reported CNVs are confirmed using another technology such as aCGH, MLPA, or PCR.
- We will need about one teaspoon of blood (3-5 ml of whole blood or DNA extracted from blood) from each individual to perform testing. In rare instances, a second specimen may be requested.
- Results of the test will be presented in an individualized, written report transmitted to the patient's Healthcare Provider(s).
- For additional information about this test, see the Diagnostic PGxome test description on the PreventionGenetics website (https://www.preventiongenetics.com/ClinicalTesting/TestCategory/PGxome.php).

FAMILY TESTING

- Testing of family members is very helpful for interpretation of results. When possible, testing of the patient and two other family members (called a trio), preferably biological parents, should be performed. If one or both biological parents are unavailable, sometimes siblings or other close relatives can be tested. Family testing increases the chance of getting a conclusive result.
- It is very important family genetic relationships are correctly stated because issues such as an undisclosed adoption or uncertain paternity can cause confusion. If you are aware of any such issues in the family, they should be discussed confidentially with your Genetic Counselor or Ordering Physician.
- Family member information (i.e. parental genotype information) helps us interpret the patient's result and will be included in the patient's report for all findings mentioned. If parental status for variants in the patient's report is not desired (for primary and/or secondary findings), please make note of this under "Patient Test Selection" on page 3.
- Separate reports can be issued for family members upon request. If family member(s) desire their own test report, please complete either the "PGxome Health Screen Test Requisition Form" or the "Diagnostic PGxome Test Requisition Form", depending on which form is more appropriate, for each individual who desires a report. Reports for family member(s) incur an additional $990 charge per family member.

REPORT INFORMATION

- Genetic variants are defined as the differences between the patient's DNA and the human reference DNA.
- We will generally only report results that may explain the patient's clinical features.
- In genes that are believed to be associated or possibly associated with the patient's clinical features, we will report all Pathogenic, Likely Pathogenic, and Variants of Uncertain Significance (unknown if they cause disease).
- We may report other findings (aka "Secondary Findings" - see below) depending on the patient's preferences (see bottom of first page of Test Requisition Form). These Secondary Findings may have an important impact on health.
- New research results are continually improving our ability to interpret the WES results. An ordering Healthcare Provider can request a re-interpretation from us.

ISSUING THE REPORT

- Results will be sent directly to the ordering Healthcare Provider(s) and NOT to the patient.
- We recommend genetic counseling and/or clinical genetics consultation before and after testing is completed.
- Patients have the right to receive a copy of their test report. They may obtain a copy from their Healthcare Provider(s) or if a signed patient authorization (form available upon request) is received, from PreventionGenetics.

SECONDARY FINDINGS

- In many patients, WES will reveal one or more additional genetic variants which could be important to the patient's health. These include for example variants predisposing the patient to cancer or heart disease, variants relevant to reproductive planning, and variants which may inform drug prescription. These are termed secondary findings. The patient may or may not wish to be informed of secondary findings.
- The patient and/or patient's family will have a choice on which types of secondary findings are reported (see bottom of first page of Test Requisition Form). Please consider the following carefully. Variants described in these sections will only be reported if the patient OPTS IN.
  - o The American College of Medical Genetics and Genomics recommends that all labs performing WES report pathogenic variants

in 59 genes that cause certain, mostly dominantly inherited disorders (Kalia et al. 2016. Genet Med. Advance online publication. doi:10.1038/gim.2016.190). These disorders are treatable and/or preventable. Included on this list are some cancer predisposition conditions, heart conditions associated with sudden death, and conditions that could result in severe health consequences if surgery is performed with certain anesthetics.

- Genes involved in other inherited disorders are not included on the ACMG list of 59 genes because it is not as clear if treatment or prevention will be effective (examples: Amendola et al. 2015. Genome Res 25(3):305-315; Dorschner et al. 2013. Am J Hum Genet 93(4):631-640). Some of these disorders are very serious, leading to death. Some people may want to know about these disorders while others may prefer not to know. Many of these conditions have adult onset, and in accordance with current professional guidelines (Borry et al. 2006 Clin Genet 70(5):374-81; Lucassen et al. 2010 British Society for Human Genetics; Fallat et al. 2013 Pediatrics 131(3): 620–2; NSGC Position Statement 2017), we do not recommend testing for adult onset conditions in minors (under the age of 18 years). For minors, we recommend that this testing be postponed until the age of 18 years or that access to this portion of their healthcare records be blocked until they reach 18 years.
- WES can also provide pan-ethnic carrier screening for autosomal recessive disorders or X-linked recessive disorders (in females). Such single recessive, pathogenic variants usually don't appreciably affect a patient's health, but may affect reproductive planning. In accordance with current professional guidelines (Borry et al. 2006. Eur J Hum Genet 14(2):133-8; NSGC Position Statement 2012; Ross et al. 2013 Genet Med 15(3):234-245), we do not recommend carrier testing for minors (under the age of 18 years). For minors, we recommend that carrier testing be postponed until the age of 18 years. In rare cases, we will consider special requests for PGxome carrier testing of minors.
- Pharmacogenetic variants are those that influence an individual's response to certain prescription medications. A healthcare provider considering prescribing one of these medications may adjust dosage or choose an alternate medication dependent on the presence of certain pharmacogenetic variants. PreventionGenetics has selected 15 genes known to influence prescription drug response to include in our PGxome based on evidence supporting gene-drug interactions (https://www.pharmgkb.org). For a full gene list, please see the PGxome test description on our website. Although less likely, rare variants in these genes may be identified that confer risk for a Mendelian disease or carrier status. The pharmacogenetics section of the report will only include variants associated with drug response.

- Genetic variants related to complex disease, and mitochondrial disorders (excluding nuclear genes) will not be reported at this time.
- Genetic variants in genes not currently known to be clinically relevant will not be reported.
- If we learn that family relationships are not as expected (for example, non-paternity), this information will be relayed to the healthcare provider(s) for discussion, but will not be included in the patient's report.

DATA

- PreventionGenetics will store the patient's sequence data. This will permit reanalysis and reinterpretation of the data in the future. Upon a physician's request, PreventionGenetics will perform, without additional charge, one reanalysis and reinterpretation of the data within two years of the date on the original test report. Thereafter, reanalysis and reinterpretation may be requested, but a fee will be charged for this service.
- PreventionGenetics recommends DNA sequence information from this test also be stored in the patient's electronic medical record. This will best benefit the patient and family members. Upon request, PreventionGenetics will provide WES data such as a list of sequence variants, a list of genes analyzed, and .bed files with coverage information. PreventionGenetics does not supply software for data review and interpretation.

RISKS

- Blood draw risks include bruising and bleeding. There is also a small chance you may get an infection, have excess bleeding, become dizzy, or faint from the blood draw.
- Learning about test results can be stressful and upsetting.
- The patient and/or patient's family may have concerns about genetic discrimination, including health insurance, life insurance, employment and long-term disability. These should be addressed according to federal and state laws. The Federal Genetic Information Non-discrimination Act (GINA) prohibits the use of genetic information for discrimination in health insurance and employment.

LIMITATIONS

- This test targets most, but not all, of the coding parts of our genes (called exons). All of the exons together is called the exome. The exome only covers approximately 1.5% of all the genetic material. However, testing the exome covers the vast majority of genetic variants which cause single gene (or Mendelian) disorders.
- Interpretation of the test results is limited by the information currently available. Better interpretation could be possible in the future as more data and knowledge about human genetics are accumulated.
- Testing will detect single base pair changes and small and large deletions or duplications, but we are generally not able to detect other types of genetic changes (e.g. rearrangements, inversions, deep intronic variants, methylation abnormalities, or repetitive sequence changes).
- This test will not provide detection of certain genes or specific exons of genes due to complicated technicalities (such as sequence characteristics, interfering pseudogenes, or inadequate coverage). In the case of deletions/duplications, most will be detected including intragenic CNVs and large cytogenetic events. CNVs of 4 exons or more in size are detected with sensitivity approaching 100% through analysis of NGS data. However, sensitivity for detection of CNVs smaller than 4 exons is lower (we estimate ~75%). Sensitivity may vary from gene-to-gene based on exon size, depth of coverage, and characteristics of the region. *Because of these technicalities, this test is not 100% sensitive and will not identify all disease-causing genetic variants.*
- Even if a disease-causing genetic variant associated with the patient's symptoms is identified, it may not allow for predictions regarding severity of the disease or prognosis.
- It is very important your Healthcare Provider(s) provide an accurate family history and clinical information as that information is critical for result interpretation. Detailed clinical information (such as clinical features, a family pedigree, and results of prior testing) is required for testing to proceed.
- Additional limitations to this test will be provided in the Supplementary material included with the test report.

CONFIDENTIALITY

- We take confidentiality and patient privacy very seriously. We follow confidentiality laws related to protected health information and are a CAP and CLIA certified laboratory.

TURNAROUND TIME (TAT)

- The maximum TAT for our PGxome® test is 45 days.

I have provided informed consent to my patient. My patient has had the opportunity to ask questions. Please indicate patient preferences for secondary findings on the Diagnostic PGxome® Test Requisition Form. If family member(s) desire their own test report, please complete the "PGxome® Health Screen Test Requisition Form" for each individual who desires a report.

HEALTHCARE PROVIDER SIGNATURE _____ PRINTED NAME _____ DATE _____

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DIAGNOSTIC PGxome®
TEST REQUISITION FORM

- The primary purpose of this test is for diagnosis. For carrier and disease susceptibility screening, please use our PGxome® Health Screen Test Requisition Form.
- Test information is available at www.PreventionGenetics.com.
- Testing must be ordered by a qualified Healthcare Provider.

ORDERING CHECKLIST (required)
- ☐ Patient and family members (if provided) specimens
- ☐ Healthcare Provider Statement
- ☐ Relevant medical records and family health history (i.e. clinic notes, prior genetic testing, pedigree)

PERSON COMPLETING FORM	CONTACT (PHONE OR EMAIL)	DATE OF REQUEST
		____ / ____ / ____ MONTH / DAY / YEAR

PATIENT INFORMATION

LAST (FAMILY) NAME	FIRST NAME	MI	DATE OF BIRTH
			____ / ____ / ____ MONTH / DAY / YEAR

PATIENT ID	SPECIMEN COLLECTION DATE	SEX
	MONTH ____ / DAY ____ / YEAR ____	☐ Male ☐ Female ☐ Other

SSPECIMEN SOURCE
- ☐ Whole Blood
- ☐ Direct Amniotic Fluid
- ☐ Direct CVS
- ☐ Extracted DNA, Source ____
- ☐ Cultured Cells, Source ____
- ☐ Tissue, Source ____
- ☐ Saliva
- ☐ Other ____

REASON FOR TEST
- ☐ Diagnosis / Affected
- ☐ Presymptomatic / At Risk
- ☐ Carrier Testing

SPECIFY KARYOTYPE

GEOANCESTRY / ETHNICITY

HAS PATIENT BEEN TESTED PREVIOUSLY AT PreventionGenetics?
☐ NO ☐ YES, PG ID# ____

HAS PATIENT'S RELATIVE BEEN TESTED PREVIOUSLY AT PreventionGenetics?
☐ NO ☐ YES
Name and DOB or PG ID# ____

BLOOD TRANSFUSION
☐ NO
☐ Within Last 30 Days, Date and Type
____ / ____ / ____ MONTH / DAY / YEAR
TYPE ____

BONE MARROW TRANSPLANT
☐ NO
☐ YES
____ / ____ / ____ MONTH / DAY / YEAR

ONGOING PREGNANCY
☐ NO
☐ YES
Prenatal Healthcare Statement required for fetal testing of ongoing pregnancies.

OTHER RELEVANT CLINICAL INFORMATION (Labs, biopsies, other genetic testing performed, etc.) ***PLEASE ATTACH PEDIGREE, IF POSSIBLE.***

PATIENT TEST SELECTION

☐ Test Code 5000 ***PATIENT ONLY***

☐ Test Code 5000 ***FAMILY (duo, trio, etc.)***
- ☐ Proband's Form
- ☐ Additional Family Member Form

Secondary (Additional) Findings

Testing may reveal other genetic information unrelated to the patient's phenotype. These are termed Secondary Findings. **Details can be found in the Diagnostic PGxome Healthcare Provider Statement (required). Options for reporting of Secondary Findings are to be marked below.**
- ☐ OPT IN: ACMG 59 GENES
- ☐ OPT IN: OTHER PREDISPOSITIONS / DIAGNOSES
- ☐ OPT IN: CARRIER STATUS
- ☐ OPT IN: PHARMACOGENETIC VARIANTS

COMMENTS:

ADDITIONAL FAMILY MEMBERS (PGxome Family Only)

Please list family members' information. Biological parent samples are recommended.

NAME (LAST, FIRST)	DATE OF BIRTH	SAMPLE TYPE	RELATIONSHIP TO PROBAND	AFFECTED?	REPORT REQUESTED?*
	____ / ____ / ____ MONTH / DAY / YEAR				☐ NO ☐ YES
	____ / ____ / ____ MONTH / DAY / YEAR				☐ NO ☐ YES
	____ / ____ / ____ MONTH / DAY / YEAR				☐ NO ☐ YES

*If family member(s) desire their own test report, please complete either the "PGxome Health Screen Test Requisition Form" or the "PGxome Diagnostic Test Requisition Form," whichever is more appropriate, for each individual. Reports for family member(s) incur an additional $990 charge per family member. If left blank, we will assume no report is requested.

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CLINICAL INFORMATION

A copy of clinic records is required for PGxome®. Clinical information is critical for best interpretation of data. Other relevant medical records, genetic testing results, and/or family medical health history (pedigree) are encouraged to be included.

PRIMARY INDICATION

☐ Developmental Delay ☐ Multiple Congenital Anomalies ☐ Neuromuscular
☐ Dysmorphic Features ☐ Neurological ☐ Other _____

ADDITIONAL CLINICAL INFORMATION *(optional - check all that apply)*

PERINATAL HISTORY
- ☐ Cystic Hygroma
- ☐ Hydrops Fetalis
- ☐ Increased Nuchal Translucency (NT)
- ☐ Intrauterine Growth Restriction (IUGR)
- ☐ Oligohydramnios
- ☐ Polyhydramnios
- ☐ Prematurity
- ☐ Other _____

GROWTH AND DEVELOPMENT
- ☐ Failure to Thrive
- ☐ Fine Motor Delay
- ☐ Gross Motor Delay
- ☐ Overgrowth
- ☐ Short Stature
- ☐ Other _____

COGNITION AND BEHAVIOR
- ☐ ADHD
- ☐ Autism
- ☐ Global Developmental Delay
- ☐ Intellectual Disability
  - ☐ Mild
  - ☐ Moderate
  - ☐ Severe
- ☐ Learning Disability
- ☐ Obsessive-Compulsive Disorder
- ☐ Speech Delay
- ☐ Other _____

MUSCULOSKELETAL
- ☐ Club Foot / Feet
- ☐ Contractures
- ☐ Diaphragmatic Hernia
- ☐ Joint Hypermobility
- ☐ Kyphosis
- ☐ Limb Anomaly
- ☐ Pes Planus
- ☐ Polydactyly
- ☐ Pterygium
- ☐ Scoliosis
- ☐ Syndactyly
- ☐ Vertebral Anomaly
- ☐ Other _____

CARDIOVASCULAR
- ☐ Arrhythmia
- ☐ ASD
- ☐ Cardiomyopathy
- ☐ Coarctation of Aorta
- ☐ Tetralogy of Fallot
- ☐ VSD
- ☐ Other _____

SKIN, HAIR, AND NAILS
- ☐ Hyperpigmentation (describe) _____
- ☐ Hypopigmentation (describe) _____
- ☐ Unusual Scarring
- ☐ Connective Tissue Abnormality (describe) _____
- ☐ Ichthyosis
- ☐ Rash
- ☐ Blistering
- ☐ Lipoma (or other skin tumors)
- ☐ Hair Abnormality (describe) _____
- ☐ Nail Abnormality (describe) _____
- ☐ Other _____

HEMATOLOGIC AND IMMUNOLOGIC
- ☐ Anemia
- ☐ Thrombocytopenia
- ☐ Neutropenia
- ☐ Pancytopenia
- ☐ Immunodeficiency
- ☐ Other _____

NEUROLOGICAL AND MUSCULAR
- ☐ Ataxia
- ☐ Chorea
- ☐ Seizures / Epilepsy
- ☐ Encephalopathy
- ☐ Hypotonia
- ☐ Hypertonia
- ☐ Spasticity
- ☐ Dystonia
- ☐ Muscle Weakness / Atrophy
- ☐ Exercise Intolerance
- ☐ Structural Brain Abnormalities / Abnormal Brain Imaging (describe) _____
- ☐ Other _____

CRANIOFACIAL
INCLUDING HEARING & VISION
- ☐ Cleft Lip
- ☐ Cleft Palate
- ☐ Craniosynostosis
- ☐ Dysmorphic Features (describe) _____
- ☐ Ear Malformation (describe) _____
- ☐ Microcephaly
- ☐ Macrocephaly
- ☐ Cataracts
- ☐ Coloboma (of eye)
- ☐ Chronic Progressive External Ophthalmoplegia
- ☐ Ptosis
- ☐ Abnormal Vision (describe) _____
- ☐ Optic Atrophy
- ☐ Retinitis Pigmentosa
- ☐ Abnormal Eye Movement
- ☐ Abnormal Hearing (describe) _____
- ☐ Other _____

GASTROINTESTINAL
- ☐ Gastroschisis
- ☐ Omphalocele
- ☐ Pyloric Stenosis
- ☐ Anal Atresia
- ☐ Tracheoesophageal Fistula
- ☐ Chronic Diarrhea
- ☐ Chronic Constipation
- ☐ Gastroesophageal Reflux
- ☐ Recurrent Vomiting
- ☐ Hirschsprung Disease
- ☐ Chronic Intestinal Pseudoobstruction
- ☐ Other _____

GENITOURINARY
- ☐ Ambiguous Genitalia
- ☐ Cryptochidism
- ☐ Hydronephrosis
- ☐ Hypospadias
- ☐ Kidney Malformation
- ☐ Renal Agenesis or Dysgenesis
- ☐ Renal Tubulopathy
- ☐ Other _____

ENDOCRINE
- ☐ Diabetes Mellitus
  - ☐ Type I
  - ☐ Type II
- ☐ Hypothyroidism
- ☐ Hyperthyroidism
- ☐ Hypoglycemia
- ☐ Hypoparathyroidism
- ☐ Hyperparathyroidism
- ☐ Other _____

METABOLIC
- ☐ Abnormal Acylcarnitine Profile (describe) _____
- ☐ Abnormal CPK
- ☐ Abnormal Urine Organic Acids (describe) _____
- ☐ Abnormal Plasma Amino Acids (describe) _____
- ☐ Hyperammonemia
- ☐ Ketosis
- ☐ Lactic Acidosis
- ☐ Metabolic Acidemia
- ☐ Other _____

CANCER / TUMORS
- ☐ Tumor (describe) _____
  - ☐ Age of Onset _____
- ☐ Other _____

ADDITIONAL TESTING
ATTACH COPIES OF RESULTS, IF AVAILABLE
- ☐ Chromosomes (Karyotype), result: _____
- ☐ Chromosomal Microarray (CMA), result: _____
- ☐ Newborn Screening, result: _____
- ☐ Other Molecular Studies, results: _____

FAMILY HISTORY
ATTACH PEDIGREE, IF AVAILABLE
- ☐ Consanguinity, degree of relationship: _____

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PREVENTIONGENETICS USE ONLY

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PROVIDER / LABORATORY CONTACT INFORMATION

Our preferred method of report transmission is secure email (via ZixCorp).
Please provide an email address when possible. If you have additional specific reporting requests, indicate them below.

PROVIDER INFORMATION

INSTITUTION

ADDRESS (City, State, Country and Postal Code)

REQUESTING PHYSICIAN (First, Last, Degree)		**REQUESTING GENETIC COUNSELOR OR ALLIED PROVIDER (First, Last, Degree)**	
PHONE NUMBER	**NPI#**	**PHONE NUMBER**	**NPI#**
EMAIL		**EMAIL**	

TEST REPORTING INSTRUCTIONS
Our preferred method of report transmission is email via ZixCorp

SECURE EMAIL VIA ZIXCORP ☐ *Use above email address*

☐ DO NOT USE ZIXCORP. EMAIL RESULTS VIA SHAREFILE.
☐ DO NOT EMAIL RESULTS. Send via fax (provide fax number):

(_____) _____-_____

TEST REPORTING INSTRUCTIONS
Our preferred method of report transmission is email via ZixCorp

SECURE EMAIL VIA ZIXCORP ☐ *Use above email address*

☐ DO NOT USE ZIXCORP. EMAIL RESULTS VIA SHAREFILE.
☐ DO NOT EMAIL RESULTS. Send via fax (provide fax number):

(_____) _____-_____

SENDOUT LABORATORY COMPLETE ONLY IF REPORT IS NEEDED

INSTITUTION / CONTACT

ADDRESS (City, State, Country and Postal Code)

PHONE NUMBER	**NPI# (Where Applicable)**
EMAIL	

TEST REPORTING INSTRUCTIONS
Our preferred method of report transmission is email via ZixCorp

SECURE EMAIL VIA ZIXCORP ☐ *Use above email address*

☐ DO NOT USE ZIXCORP. EMAIL RESULTS VIA SHAREFILE.
☐ DO NOT EMAIL RESULTS. Send via fax (provide fax number):

(_____) _____-_____

OTHER

INSTITUTION / CONTACT

ADDRESS (City, State, Country and Postal Code)

PHONE NUMBER	**NPI# (Where Applicable)**
EMAIL	

TEST REPORTING INSTRUCTIONS
Our preferred method of report transmission is email via ZixCorp

SECURE EMAIL VIA ZIXCORP ☐ *Use above email address*

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☐ DO NOT EMAIL RESULTS. Send via fax (provide fax number):

(_____) _____-_____

As the ordering Healthcare Provider, I confirm I have obtained the patient's informed consent, either verbally or in writing, to perform this test. I further confirm the patient has been appropriately counseled and understands the risks, benefits, and limitations of this genetic testing and the implications of the results.

PREVENTION GENETICS

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PREVENTIONGENETICS USE ONLY

BILLING - PLEASE SELECT INSTITUTIONAL OR SELF-PAY WITH OPTION TO SUBMIT TO INSURANCE

PATIENT TESTING WILL BE DELAYED UNTIL ALL OF THE BILLING REQUIREMENTS HAVE BEEN MET. PLEASE PRINT CLEARLY.

If the patient's specimen is collected in New York, a New York State Non-Permitted Laboratory Test Request approval letter (where applicable) and Genetic Testing Healthcare Provider Statement (see website) must be included before testing will proceed.

INSTITUTIONAL BILLING

BILLING INSTITUTION		PO NUMBER	
CONTACT	PHONE NUMBER	EMAIL	
ADDRESS	CITY	STATE	ZIP
BILLING ACCOUNT NUMBER ☐ UPDATED INFO		COPY OF TEST REPORT(S) FOR BILLING	
EMAIL INVOICE VIA ZIXCORP (PROVIDE EMAIL ADDRESS)		☐ EMAIL (VIA ZIXCORP) _____	
		☐ OTHER (PLEASE SPECIFY) _____	

SELF-PAY **THIS SECTION MUST BE FILLED OUT COMPLETELY**

RESPONSIBLE PARTY'S NAME *(MUST BE 18 YEARS OR OLDER)*	PHONE NUMBER	EMAIL	
ADDRESS	CITY	STATE	ZIP

ACCEPTANCE of financial responsibility for genetic testing ***PREVENTIONGENETICS CANNOT PROCEED WITH TESTING OF THE SPECIMEN WITHOUT A SIGNATURE BELOW.***

My signature indicates that I accept financial responsibility for all fees associated with this genetic testing order.

SIGNATURE OF RESPONSIBLE PARTY _____ PRINTED NAME OF RESPONSIBLE PARTY _____ DATE _____

COMPLETE THE FOLLOWING FOR CREDIT CARD PAYMENT	CREDIT CARD NUMBER (VISA, DISCOVER, OR MASTERCARD ONLY)	EXPIRATION DATE	3-DIGIT SECURITY CODE

My signature authorizes PreventionGenetics to charge my credit card for services for which I am responsible.

SIGNATURE _____ DATE _____

☐ SUBMIT CLAIM TO INSURANCE (OPTIONAL)

POLICYHOLDER'S NAME (REQUIRED)		PLEASE INDICATE THE TYPE OF INSURANCE ☐ PRIVATE ☐ MEDICARE ☐ WI MEDICAID *We only accept WI Medicaid*	
PRIMARY INSURANCE COMPANY NAME (REQUIRED)		☐ *ATTACH A COPY OF INSURANCE CARD both sides*	
INSURANCE COMPANY ADDRESS - CLAIMS	CITY	STATE	ZIP
ICD-10 CODES (REQUIRED)	POLICY ID#	GROUP #	AUTHORIZATION #

PLEASE ATTACH THE FOLLOWING DOCUMENTATION *PreventionGenetics cannot proceed with testing of the specimen until all information is received.*

☐ NPI # of Requesting Physician _____

☐ *MEDICARE – signed ABN Form completed IN FULL*

☐ *AUTHORIZATION NUMBER* or letter of agreement from Insurance Company (if available). If not included, we will routinely perform pre-verification prior to initiating testing and will relay information to ordering provider.

☐ *Relevant Medical Records addressing medical necessity and/or Letter of Medical Necessity*

☐ ***SHARE RESULTS*** of benefits investigation with patient directly via email provided above

or FAX # (_____) _____-_____

AUTHORIZATION to assign benefits and accept financial responsibility for my account

PREVENTIONGENETICS CANNOT PROCEED WITH TESTING OF THE SPECIMEN WITHOUT A SIGNATURE BELOW.

I authorize PreventionGenetics to release information received including, without limitation, medical information, which includes laboratory test results, such as genetic tests results, to my health plan/insurance carrier and its Authorized Representatives. I further authorize insurance payments directly to PreventionGenetics for the services rendered. I understand my health plan/insurance/Medicare/Medicaid carrier may not approve and reimburse my medical genetic services in full due to usual and customary rate limits, benefit exclusions, coverage limits, lack of authorization, medical necessity or otherwise. I understand I am financially responsible for fees not paid in full by my insurer, co-payments, and policy deductibles except where my liability is limited by contract or State and Federal law. I agree to help PreventionGenetics resolve any insurance claim issues. My signature indicates that I accept financial responsibility for all fees associated with this genetic testing order.

SIGNATURE OF RESPONSIBLE PARTY _____ PRINTED NAME OF RESPONSIBLE PARTY _____ DATE _____

PREFERRED SPECIMEN REQUIREMENTS AND TURNAROUND TIMES (TAT)

PLEASE CONTACT US WITH ADDITIONAL SPECIMEN REQUIREMENT QUESTIONS.

STAT TAT (8-10 calendar days) available at a 25% surcharge for Sanger Sequencing and aCGH. Cannot be guaranteed for aCGH.

WHOLE BLOOD

Collect 3 ml - 5 ml of whole blood in EDTA (purple top tube) or ACD (yellow top tube), minimum 1 ml for small infants.

DNA

Send in screw cap tube at least 5 µg -10 µg of purified DNA at a concentration of at least 20 ng/µL for NGS and Sanger tests and at least 5 µg of purified DNA at a concentration of at least 100 ng/µL for gene-centric aCGH, MLPA, and CMA tests, minimum 2 µg for limited specimens. Indicate concentration on tube label. For requests requiring more than one test, send an additional 5 µg DNA per test ordered when possible.

SALIVA

Oragene™ or GeneFiX™ Saliva Collection kit used according to manufacturer instructions.

FETAL (CVS / AMNIOCYTES) AND OTHER CELL CULTURES

Culture and send at least two, T25 flasks of confluent cells. For sequencing or gene-centric aCGH panels, two flasks are often sufficient; however, some panels may require additional flasks (dependent on size of genes, amount of Sanger sequencing required, etc.). Multiple test requests may also require additional flasks. Please contact us for details. We strongly recommend maintaining a back-up culture. Fetal cell cultures are available at PreventionGenetics from direct amniotic fluid, chorionic villi, or products of conception (POC) via Test Code #995 (cost $250). Collect 10 ml - 20 ml of direct amniotic fluid or 5 mg - 10 mg cleaned CVS tissue (~15-20 cleaned villi) or 2mm x 2mm x 2mm fresh tissue. CPT code 88235 for amniotic fluid/chorionic villi or 88233 for POC specimens.

	TEST METHOD	WHOLE BLOOD	DNA	SALIVA	CELL CULTURES	FRESH, FROZEN TISSUE	BUCCAL SWAB	DIRECT AMNIOTIC FLUID/CVS	OTHER	TURN AROUND TIME (TAT)
SEQUENCING	NextGen (NGS)	★	★	★	★	★	■	■ C	–	28 days
	PGxome® / PGxome Custom Panels	★	★ B	★	★ B	★	–	–	–	6 weeks
	Sanger	★	★	★	★	★	■	■ C	Semen D	18 days
DEL / DUP	Gene-centric aCGH	★	★	■	■	★	–	■ C	–	28 days
	MLPA	★	★ A	ONLY TEST #1941	★ A	–	–	–	–	20 days
	Chromosomal Microarray (CMA)	★	★	■	★	★	–	■	–	20 days

EXCEPTIONS

A - Cell cultures and DNA extracted from CVS and amniocytes not accepted for MLPA; DNA extracted from saliva (except test #1941) also not accepted.

B - Cell cultures and DNA extracted from CVS and amniocytes acceptable for PGxome for non-ongoing pregnancies only.

C - Direct prenatal specimen types most appropriate for targeted prenatal familial variant testing (Test Code #990), and strongly discouraged for full gene and panel tests. Back-up culture highly recommended.

D - Semen: Collect 1-2 vials and flash freeze. Vials to be sent frozen (preferably on dry ice). Contact us for details.

KEY

★ PREFERRED

■ ACCEPTED

– NOT ACCEPTED

FRESH, FROZEN TISSUE

Collect 2mm x 2mm x 2mm tissue and flash freeze. Tissue to be sent frozen (preferably dry ice). Contact us for additional details.

BUCCAL SWAB

ORAcollect•Dx (OCD-100) collection kit used according to manufacturer instructions. Buccal swabs are most appropriate for targeted, known variant testing.

DIRECT AMNIOTIC FLUID / CHORIONIC VILLI

Collect 10 ml -20 ml of direct amniotic fluid or 5 mg -10 mg cleaned CVS tissue (~15-20 cleaned villi). We strongly recommend maintaining a local back-up culture. Fetal cell cultures available (Test Code #995, $250).

SHIPPING AND HANDLING INSTRUCTIONS

Please label all specimen containers with the patient's name, date of birth and/or ID number. At least two identifiers should be listed on specimen containers. We accept specimen deliveries Monday-Saturday for all specimen types except cell cultures, direct amniotic fluid, or direct chorionic villi. Cell culture deliveries are routinely accepted Monday-Thursday and require advance notice of arrival. If a Friday or Saturday delivery is necessary, please contact us to make arrangements. Saturday delivery should especially be avoided when possible as prenatal specimens are not processed over the weekend. Holiday schedules will be posted on our home page at least one week prior to major holidays.

BLOOD

DO NOT FREEZE. During hot weather, include a frozen ice pack in the shipping container. Place a paper towel or other thin material between the ice pack and the blood tube. In cold weather include an unfrozen ice pack in the shipping container as insulation. At room temperature, blood specimen is stable for up to 48 hours. If refrigerated, blood specimen is stable for up to one week.

DNA

DNA may be shipped at room temperature. Label the tube with the composition of the solute, DNA concentration as well as the patient's name, date of birth, and/or ID number. We only accept genomic DNA for testing. We do not accept products of whole genome amplification reactions or other amplification reactions.

CELL CULTURES, DIRECT AF/CVS, AND POC

Send specimens overnight in an insulated, shatterproof container. Direct AF/CVS or POC specimens can be sent in saline or culture media at room temperature for culturing at PreventionGenetics (Test Code #995, $250).

PRENATAL TESTING

Please sign Prenatal Healthcare Provider's Statement for ongoing pregnancies and contact us in advance regarding prenatal test requests. When possible, ship prenatal samples to arrive at PreventionGenetics no later than Thursday.

DNA GENOTYPING PANEL

For quality control purposes, the PreventionGenetics DNA Genotyping Panel is performed on all clinical specimens. Genotyping results are not included in test reports.

DNA BANKING

DNA Banking has a reduced price of $98 for patients if clinical testing is also being performed at PreventionGenetics. Visit our website at www.PGDNABank.com for information about the process and forms. For questions related to PGDNABanking, contact our DNA Banking Director at (715) 387-0484, ext. 151, or email: dnabanking@preventiongenetics.com.

CONTACT US

For additional questions or concerns, please contact our Client Service Representatives at (715) 387-0484, ext. 0, or our Genetic Counseling Team at option 2, or email: clinicaldnatesting@preventiongenetics.com.

ADDRESS

PreventionGenetics - Diagnostic Lab
3800 S. Business Park Ave.
Marshfield, Wisconsin 54449
USA

TESTING KITS

Clinical testing kits with prepaid return shipping are available for U.S. Clients. We are able to provide clinical testing kits to International clients without the return postage. To order test kits, submit requests through our electronic order form (see website) or contact our Client Service Representatives at (715) 387-0484, ext. 0.