

# PGxome Test Requisition Form

(revised 3/7/2016)

<b>Person completing form</b>	<b>Contact Information (phone or email)</b>	<b>Date of Request</b>
-------------------------------	---	------------------------

**PGxome Ordering Checklist (required):**

- Proband (patient) specimen
- Family member specimen(s)
- PGxome Healthcare Provider Statement
- Clinic notes, summary, and/ or relevant medical records
- Pedigree with family medical health history
- Previous genetic testing results (where available)

Proband Information				
<b>Patient's Last (Family) Name</b>	<b>First Name</b>	<b>MI</b>	<b>Date of Birth:</b>	<b>Month      Day      Year</b>
<b>Patient ID Code</b>	<b>Date Collected:</b>	<b>Month      Day      Year</b>	<b>Gender:</b> <input type="checkbox"/> Male <input type="checkbox"/> Female <input type="checkbox"/> Other	
<b>Specimen Source:</b> <input type="checkbox"/> Whole blood <input type="checkbox"/> Extracted DNA Source: <input type="checkbox"/> Cultured Cells Source: <input type="checkbox"/> Tissue Source: <input type="checkbox"/> Other:			<b>GeoAncestry/Ethnicity</b>	
<b>Has patient been tested previously at PreventionGenetics?</b> <input type="checkbox"/> Yes <input type="checkbox"/> No <i>If yes, PG ID#:</i>	<b>Has patient's relative been tested at PreventionGenetics?</b> <input type="checkbox"/> Yes <input type="checkbox"/> No <i>If yes, provide name &amp; DOB:</i>	<b>Related to an ongoing pregnancy?</b> <input type="checkbox"/> Yes <input type="checkbox"/> No	<b>Bone marrow transplant or blood transfusion?</b> <input type="checkbox"/> Yes <input type="checkbox"/> No <i>If yes, date:</i>	

Test Selection	Secondary Findings
<input type="checkbox"/> Exome Sequencing only	The American College of Medical Genetics (ACMG) has recommended secondary findings in 56 genes be reported. Pathogenic or likely pathogenic variants in these genes will be reported by default unless a patient desires to "opt-out." <input type="checkbox"/> <b>ACMG 56 OPT OUT</b>  Additional secondary findings can be reported if desired should the patient desire to "opt-in." <input type="checkbox"/> <b>Additional findings OPT IN</b>  <b>Details can be found in the PGxome Healthcare Provider Statement (required).</b>
<input type="checkbox"/> Chromosomal Microarray with reflex to Exome Sequencing	
<input type="checkbox"/> Chromosomal Microarray & Exome Sequencing, concurrent	

Additional Family Members				
<i>For trio testing, please list family members' information. Biological parent samples are required.</i>				
<b>Name (Last, First)</b>	<b>Date of Birth (MM/DD/YY)</b>	<b>Sample Type</b>	<b>Relationship to proband</b>	<b>Affected?</b>
<b>Name (Last, First)</b>	<b>Date of Birth (MM/DD/YY)</b>	<b>Sample Type</b>	<b>Relationship to proband</b>	<b>Affected?</b>
<b>Name (Last, First)</b>	<b>Date of Birth (MM/DD/YY)</b>	<b>Sample Type</b>	<b>Relationship to proband</b>	<b>Affected?</b>

## Clinical Information

### Indication for Testing

A copy of a clinic summary/note is required. Other relevant medical records, genetic testing results, and/or other results are encouraged to be included. Please also include a copy of the pedigree with family medical health history.

**Clinical information is critical for best interpretation of PGxome data.**

**Primary Indication:**

- |  |  |
|--|--|
| <input type="checkbox"/> Developmental Delay           | <input type="checkbox"/> Neurological            |
| <input type="checkbox"/> Dysmorphic Features           | <input type="checkbox"/> Other (provide detail): |
| <input type="checkbox"/> Multiple Congenital Anomalies |  |

### Additional Clinical Information

(optional - check all that apply)

<p><u>Perinatal History</u></p> <input type="checkbox"/> Prematurity <input type="checkbox"/> Intrauterine Growth Restriction (IUGR) <input type="checkbox"/> Oligohydramnios <input type="checkbox"/> Polyhydramnios <input type="checkbox"/> Cystic hygroma <input type="checkbox"/> Increased nuchal translucency (NT) <input type="checkbox"/> Other <p><u>Growth &amp; Development</u></p> <input type="checkbox"/> Failure to thrive <input type="checkbox"/> Overgrowth <input type="checkbox"/> Short stature <input type="checkbox"/> Fine motor delay <input type="checkbox"/> Gross motor delay <input type="checkbox"/> Other <p><u>Cognition &amp; Behavior</u></p> <input type="checkbox"/> Speech delay <input type="checkbox"/> Intellectual disability <ul style="list-style-type: none"> <li><input type="checkbox"/> Mild</li> <li><input type="checkbox"/> Moderate</li> <li><input type="checkbox"/> Severe</li> </ul> <input type="checkbox"/> Learning disability <input type="checkbox"/> Autism spectrum disorder <input type="checkbox"/> ADHD <input type="checkbox"/> Obsessive-compulsive disorder <input type="checkbox"/> Other <p><u>Musculoskeletal</u></p> <input type="checkbox"/> Club foot/feet <input type="checkbox"/> Contractures <input type="checkbox"/> Pterygium <input type="checkbox"/> Diaphragmatic hernia <input type="checkbox"/> Limb anomalies <input type="checkbox"/> Polydactyly <input type="checkbox"/> Syndactyly <input type="checkbox"/> Scoliosis <input type="checkbox"/> Kyphosis <input type="checkbox"/> Vertebral anomaly <input type="checkbox"/> Other	<p><u>Skin, Hair, &amp; Nails</u></p> <input type="checkbox"/> Hyperpigmentation (describe) <input type="checkbox"/> Hypopigmentation (describe) <input type="checkbox"/> Unusual scarring <input type="checkbox"/> Connective tissue abnormality (describe) <input type="checkbox"/> Ichthyosis <input type="checkbox"/> Rash <input type="checkbox"/> Blistering <input type="checkbox"/> Lipoma (or other skin tumors) <input type="checkbox"/> Hair abnormality (describe) <ul style="list-style-type: none"> <li><input type="checkbox"/> Quality</li> <li><input type="checkbox"/> Quantity</li> <li><input type="checkbox"/> Distribution</li> <li><input type="checkbox"/> Pigmentation</li> </ul> <input type="checkbox"/> Nail abnormality (describe) <ul style="list-style-type: none"> <li><input type="checkbox"/> Size</li> <li><input type="checkbox"/> Shape</li> <li><input type="checkbox"/> Texture</li> </ul> <input type="checkbox"/> Other <p><u>Hematologic &amp; Immunologic</u></p> <input type="checkbox"/> Anemia <input type="checkbox"/> Neutropenia <input type="checkbox"/> Pancytopenia <input type="checkbox"/> Immunodeficiency <input type="checkbox"/> Other <p><u>Neurological &amp; Muscular</u></p> <input type="checkbox"/> Ataxia <input type="checkbox"/> Chorea <input type="checkbox"/> Seizures/Epilepsy <input type="checkbox"/> Encephalopathy <input type="checkbox"/> Hypotonia <input type="checkbox"/> Hypertonia <input type="checkbox"/> Spasticity <input type="checkbox"/> Dystonia <input type="checkbox"/> Muscle weakness/atrophy <input type="checkbox"/> Exercise intolerance <input type="checkbox"/> Structural brain abnormalities/abnormal brain imaging (describe) <input type="checkbox"/> Other	<p><u>Craniofacial (including hearing &amp; vision)</u></p> <input type="checkbox"/> Cleft lip <input type="checkbox"/> Cleft palate <input type="checkbox"/> Craniosynostosis <input type="checkbox"/> Dysmorphic features (describe) <input type="checkbox"/> Ear malformation (describe) <input type="checkbox"/> Microcephaly <input type="checkbox"/> Macrocephaly <input type="checkbox"/> Cataracts <input type="checkbox"/> Coloboma (of eye) <input type="checkbox"/> Chronic progressive external ophthalmoplegia <input type="checkbox"/> Ptosis <input type="checkbox"/> Abnormal vision (describe) <input type="checkbox"/> Optic atrophy <input type="checkbox"/> Retinitis pigmentosa <input type="checkbox"/> Abnormal eye movement <input type="checkbox"/> Abnormal hearing (describe) <input type="checkbox"/> Other <p><u>Gastrointestinal</u></p> <input type="checkbox"/> Gastroschisis <input type="checkbox"/> Omphalocele <input type="checkbox"/> Pyloric stenosis <input type="checkbox"/> Anal atresia <input type="checkbox"/> Tracheoesophageal fistula <input type="checkbox"/> Chronic diarrhea <input type="checkbox"/> Chronic constipation <input type="checkbox"/> Gastrointestinal reflux <input type="checkbox"/> Recurrent vomiting <input type="checkbox"/> Hirschsprung disease <input type="checkbox"/> Chronic intestinal pseudo-obstruction <input type="checkbox"/> Other <p><u>Genitourinary</u></p> <input type="checkbox"/> Ambiguous genitalia <input type="checkbox"/> Cryptorchidism <input type="checkbox"/> Hydronephrosis <input type="checkbox"/> Hypospadias <input type="checkbox"/> Kidney malformation	<p><u>Genitourinary (continued)</u></p> <input type="checkbox"/> Renal agenesis or dysgenesis <input type="checkbox"/> Undescended testis <input type="checkbox"/> Renal tubulopathy <input type="checkbox"/> Other <p><u>Endocrine</u></p> <input type="checkbox"/> Diabetes mellitus <ul style="list-style-type: none"> <li><input type="checkbox"/> Type I</li> <li><input type="checkbox"/> Type II</li> </ul> <input type="checkbox"/> Hypothyroidism <input type="checkbox"/> Hyperthyroidism <input type="checkbox"/> Hypoparathyroidism <input type="checkbox"/> Hyperparathyroidism <input type="checkbox"/> Other <p><u>Metabolic</u></p> <input type="checkbox"/> Ketosis <input type="checkbox"/> Lactic acidosis <input type="checkbox"/> Abnormal urine organic acids (describe) <input type="checkbox"/> Abnormal plasma amino acids (describe) <input type="checkbox"/> Abnormal acylcarnitine profile (describe) <input type="checkbox"/> Abnormal CPK <input type="checkbox"/> Other <p><u>Tumors</u></p> <input type="checkbox"/> Tumor (describe) <input type="checkbox"/> Age of onset <input type="checkbox"/> Other <p><u>Additional Testing</u> (please attach copies of results if available)</p> <input type="checkbox"/> Chromosomes (karyotype), result :  <input type="checkbox"/> Chromosomal Microarray (CMA), result :  Other molecular studies, results :
---	---	--	---

Office  
use  
only

## Provider/Laboratory Contact Information

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

Provider Information			
<i>Institution</i>			
<i>Address (please include city, state, country &amp; postal code)</i>			
<i>Requesting Physician (First, Last, Degree)</i>		<i>Requesting Genetic Counselor (First, Last, Degree)</i>	
<i>Phone Number</i>	<i>NPI#:</i>	<i>Phone Number</i>	<i>NPI#</i>
<i>Email</i>		<i>Email</i>	
Test Reporting Instructions		Test Reporting Instructions	
<i>Our preferred method of report transmission is email (via ShareFile)</i>		<i>Our preferred method of report transmission is email (via ShareFile)</i>	
<i>Email (via ShareFile):</i> <input type="checkbox"/> <i>use above</i>		<i>Email (via ShareFile):</i> <input type="checkbox"/> <i>use above</i>	
<input type="checkbox"/> <i>DO NOT email results. Instead, send via fax (provide fax #):</i>		<input type="checkbox"/> <i>DO NOT email results. Instead, send via fax (provide fax #):</i>	

Sendout Laboratory (Complete only if report needed)	Other
<i>Laboratory &amp; Contact Person</i>	<i>Contact Name</i>
<i>Address</i>	<i>Address</i>
<i>Phone Number</i>	<i>Phone Number</i>
<i>Email</i>	<i>Email</i>
Test Reporting Instructions	Test Reporting Instructions
<i>Our preferred method of report transmission is email (via ShareFile)</i>	<i>Our preferred method of report transmission is email (via ShareFile)</i>
<i>Email (via ShareFile):</i> <input type="checkbox"/> <i>use above</i>	<i>Email (via ShareFile):</i> <input type="checkbox"/> <i>use above</i>
<input type="checkbox"/> <i>DO NOT email results. Instead, send via fax (provide fax #):</i>	<input type="checkbox"/> <i>DO NOT email results. Instead, send via fax (provide fax #):</i>

Office  
use  
only

## Billing Instructions

### 1. Please choose one of the three billing options:

- Institutional
- Individual
- Insurance

### 2. Provide all information for the selected option only

Note: Patient testing will be delayed until all of the billing requirements have been met. Please print clearly. If Individual/Insurance billing information is incomplete, the Institution will be billed. Tests that are cancelled while in progress will be billed for the amount of work completed up to that point. If the patient's specimen is collected in New York, a New York State Non-Permitted Laboratory Test Request approval letter must be included before testing will proceed.

<b>1. Institutional Billing (Preferred)</b>			
Billing Institution		PO Number	
Contact	Phone Number(s)	Email	
Address			
City	State	Zip	
Email Invoice	Copy of Test Report(s) for Billing		
Email Address:	<input type="checkbox"/> Email (via ShareFile): <input type="checkbox"/> same as previous <input type="checkbox"/> Other (please specify):		

<b>2. Individual Billing</b>		
Responsible Party's Name <i>(Must be 18 years or older)</i>	Phone Number(s)	Email
Address		
City	State	Zip
<b>ACCEPTANCE OF FINANCIAL RESPONSIBILITY FOR GENETIC TESTING</b>		
<i>Note: PreventionGenetics cannot proceed with testing of the specimen without a signature below.</i>		
My signature below 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
<b>COMPLETE THE FOLLOWING FOR CREDIT CARD PAYMENT</b>		
Credit Card # / <i>(VISA, Discover, or Mastercard only)</i>	Expiration Date	3-Digit Security Code
My signature below authorizes PreventionGenetics to charge my credit card for services for which I am responsible.		
Signature:	Date:	

Office  
use  
only

3800 S. Business Park Ave  
Marshfield, WI 54449  
Phone: 715-387-0484  
Fax: 715-384-3661

**Billing Instructions**

<h3>3. Insurance Billing</h3>			
<p>We will file an insurance claim on behalf of the patient with any commercial insurance company. However, the claim will be submitted as an “out of network” service provider. We are in network (contracted provider) with a limited number of insurance plans (see website). The patient is responsible for any portion of the test fee not covered by the insurance company for any reason including, but not limited to, co-payments, co-insurance, unmet deductibles, or non-covered services.</p>			
Responsible Party's Name <i>(Must be 18)</i>		Phone Number(s)	Email
Responsible Party Address			
City	State	Zip	
Policyholder Name <i>(Required)</i>	Please indicate the type of insurance: <i>(Circle One)</i> Private / Medicare / WI Medicaid		Primary Insurance Company Name <i>(Required)</i>
Insurance Company Address- Claims			
City	State	Zip	
ICD-10 Codes <i>(Required)</i>	Policy ID#	Group #	Authorization #
<p><b>Please attach the following:</b>                  Note: PreventionGenetics cannot proceed with testing of the specimen until all information is received. There is a \$100 DNA extraction fee if testing is not performed (does not apply if DNA is sent).</p> <p> <input type="checkbox"/> NPI # of Requesting Physician _____  <input type="checkbox"/> Medicare – signed ABN Form <u>completed IN FULL</u>  <input type="checkbox"/> Copy of both sides of Insurance Card  <input type="checkbox"/> Authorization number or letter of agreement from insurance company (if available). If not included, we will routinely perform pre-verification prior to initiating testing &amp; will relay information to ordering provider.                 </p> <p> <input type="checkbox"/> Letter of Medical Necessity  <input type="checkbox"/> Relevant Medical Records  <input type="checkbox"/> NY Non-permitted lab approval letter (if specimen collected in NY)                 </p>			
<b>AUTHORIZATION TO ASSIGN BENEFITS AND ACCEPT FINANCIAL RESPONSIBILITY FOR MY ACCOUNT</b>			
<p>Note: PreventionGenetics cannot proceed with testing of the specimen without a signature below.</p> <p>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.</p>			
Signature of Patient or Guardian		Printed Name of Patient or Guardian	Date
Credit Card # / <i>(VISA, Discover, or Mastercard only)</i>	Expiration Date	3- Digit Security Code	
<p>My signature below authorizes PreventionGenetics to charge my credit card for services for which I am responsible upon completion of insurance processing.</p>			
Signature:			Date:

Office  
use  
only

## Specimen and Shipping Instructions

**Specimen Requirements**

**WHOLE BLOOD:** Collect 3-5 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 10 µg of purified DNA at a concentration of at least 100 µg/ml (indicate concentration on tube label) for exome only.

**SALIVA, CELL CULTURE, & FRESH, FROZEN TISSUE:** Please contact us.

**Shipping/Handling Instructions**

Please label all specimen containers with the patient 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. Cell culture deliveries are routinely accepted Monday-Thursday and require advance notice of arrival. If a Friday delivery is necessary please contact us to make arrangements. 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 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:** We are NOT able to culture cells. Send confluent flasks of cultured cells in insulated, shatterproof container overnight.

Address	Testing Kits
Diagnostic Lab PreventionGenetics 3800 S. Business Park Ave. Marshfield, WI 54449 USA	Clinical testing kits with prepaid return shipping are now available for our U.S. clients. We are able to provide Clinical Testing Kits to our international clients without the return postage at this time. To order kits, submit requests through our Electronic Order Form or contact our Client Service Representatives at 715-387-0484, ext. 0.

**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 \$69 for patients if clinical testing is also being performed with us. For DNA Banking, see our DNA Banking Process and DNA Banking Forms. For questions related to DNA Banking, contact our DNA Banking Director at 715-387-0484, ext. 151 or email [dnabanking@preventiongenetics.com](mailto: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 ext. 208 or [clinicaldnatesting@preventiongenetics.com](mailto:clinicaldnatesting@preventiongenetics.com).

**PGxome (Whole Exome Sequencing) Version 1.0  
Healthcare Provider Statement\***

November 5, 2015

\* Note: This Statement is required, and applies to all cases of Whole Exome Sequencing.

Patient's Name: \_\_\_\_\_ Date of Birth: \_\_\_\_\_  
Family Member's Name: \_\_\_\_\_ Relationship: \_\_\_\_\_  
Family Member's Name: \_\_\_\_\_ Relationship: \_\_\_\_\_  
Family Member's Name: \_\_\_\_\_ Relationship: \_\_\_\_\_

The following information should be used to as a guide to provide informed consent to the patient and/or patient's family. We require that the consenting healthcare provider sign below.

**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**

- This test involves the sequencing of thousands of genes at the same time, whereas many other genetic tests look at one gene or a cluster of genes. The way we test the exome is through a procedure called Next Generation Sequencing (NGS) technology. We confirm important results with another type of sequencing, called Sanger sequencing.
- We will need about one teaspoon of blood (3-5 mL of whole blood or DNA extracted from blood) from each family member to perform testing. In rare instances, a second specimen may be requested.
- Based on the information given by the healthcare provider(s) about symptoms and family history, our computer programs will help us filter the results. Our team of experts will then study the results and create an individualized report that will be given to the patient's healthcare provider(s).

**Family Testing**

- Testing of family members is vital for interpretation of results. We require testing of the patient and both parents (called a trio). Trio testing increases the chance of getting a conclusive result. An additional family member, usually an affected or unaffected sib, may also be included in the test.
- It is very important that family genetic relationships are correctly stated because issues such as undisclosed adoption or uncertain paternity can confuse test results. If you are aware of any such issues in the family, they should be discussed confidentially with your genetic counselor or ordering physician.
- Separate reports will not be issued for family members.

**Limitations**

- Roughly 25% of patients will receive a diagnosis or suspected diagnosis from this testing (Yang et al. N Engl J Med. 2013 Oct 17;369(16):1502-11).
- This test targets most, but not all, of the coding part of the genes (called exons). All of the exons together is called the exome. The exome only covers approximately 1% of all the genetic material.
- Testing will detect single base pair changes or small deletions or duplications, but we are generally not able to detect other types of genetic changes (e.g. large deletions and duplications, rearrangements, inversions, deep intronic variants, methylation abnormalities, or repetitive sequence changes). We generally do not have the ability to detect large deletions; however, we can detect gross homozygous or hemizygous deletions if the deletion spans 3 or more coding exons.
- This test may not provide detection of certain genes or specific exons of genes due to complicated technicalities (such as sequence characteristics or interfering pseudogenes).

- Because of the technicalities of WES, this test is not 100% sensitive and may not identify a disease-causing genetic variant associated with the patient's symptoms.
- 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 that your healthcare provider(s) provide us 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.

### Report Information

- We will generally only report results that may explain the patient's clinical features.
- In genes that are believed to be associated with the patient's clinical features, pathogenic variants (known to cause disease), likely pathogenic variants (probably cause disease), and variants of uncertain significance (unknown if they cause disease) will be reported.
- We may report other findings (aka "secondary findings" - see below) depending on the patient's preference. These secondary findings may have an important impact on health but are often unrelated to the patient's clinical features. Some secondary findings for which medical treatment may prevent or minimize serious health problems are provided in the initial report unless the patient opts out.
- Family member information (i.e. parental genotype information) that helps us interpret the patient's result will be included in the patient's report.
- We recommend that the patient stay in touch with their healthcare provider(s) to discuss any updated information regarding results and our interpretation. An ordering healthcare provider can request a re-interpretation from us by contacting our laboratory.

### Issuing the Report

- Results will be sent to the ordering healthcare provider(s) and NOT to the patient/family directly.
- We strongly recommend genetic counseling and/or clinical genetics consultation before and after testing is completed.

### Secondary Findings

- Testing might reveal information unrelated to the patient's clinical features. These are termed secondary findings. The patient undergoing testing may or may not wish to be informed of these potential secondary findings.
- The patient and/or patient's family will have a choice on which types of secondary findings are reported. *Please consider the following carefully.*
  - We follow recommendations by the American College of Medical Genetics and Genomics, who recommend that all labs that perform WES report pathogenic variants in 56 genes that cause certain inherited disorders (Green et al. 2013. Genet Med 15(7):565-574). These disorders may cause serious health problems that are treatable 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. We will standardly report pathogenic or likely pathogenic variants in these genes unless you OPT OUT. These findings will be included in the patient's report.
  - Some genetic conditions are associated with a known disease which may be serious (leading to disability or death) but are not included on the list of 56 genes. Some people may want to know about these genes for planning purposes while others may prefer not to know. Since many of these conditions have adult onset, testing for children is usually delayed until they can make their own decision. Pathogenic or likely pathogenic variants for additional secondary findings will only be reported if you OPT IN.
  - Genetic variants related to recessive carrier status, complex disease, pharmacogenetics, and mitochondrial disorders (excluding nuclear genes) will not be reported.
  - Genetic variants in genes not currently known to be associated with human disease 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**

- Upon request, PreventionGenetics will provide additional WES data (such as complete lists of sequence variants, OMIM list of genes analyzed, and files with exome coverage information). This data will be provided once testing is completed and a final report has been released. PreventionGenetics does not supply software for data review and interpretation.

**Risks**

- Blood draws can have risks associated including bruising and bleeding. There is also a small chance that 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 Genetic Information Non-discrimination Act (GINA) prohibits the use of genetic information for discrimination in health insurance and employment.

**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.

**Turn Around Time (TAT)**

- Our maximum TAT for a report is 4 months. Most reports will be completed in 2-3 months.

I have provided informed consent to my patient and/or patient’s family using the above consent form. My patient and/or patient’s family has had the opportunity to ask questions. Please indicate family preferences for secondary findings on page one of the PGxome Test Requisition Form. Please initial lines below and provide signature.

\_\_\_\_\_ I have explained to my patient and/or patient’s family that the laboratory’s policy is to report on ACMG’s 56 genes that are medically actionable unless the family wishes to OPT OUT.

\_\_\_\_\_ I have explained to my patient and/or patient’s family that the laboratory’s policy is to not standardly report other secondary findings; however, the family can OPT IN if desired.

**Healthcare Provider’s Name:** \_\_\_\_\_

**Healthcare Provider’s Signature:** \_\_\_\_\_ **Date:** \_\_\_\_\_