PREVENTION GENETICS part of EXACT SCIENCES

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

PREVENTIONGENETICS USE ONLY

**All testing must be ordered by a qualified Healthcare Provider**

**THIS FORM MUST ACCOMPANY ALL SPECIMENS**

WHOLE EXOME SEQUENCING

# PGxome® Prenatal Diagnostic

HEALTHCARE PROVIDERS STATEMENT

## THIS STATEMENT IS REQUIRED, AND APPLIES TO WHOLE EXOME SEQUENCING TESTS FOR PRENATAL DIAGNOSTIC PURPOSES.

*NOTE: This statement must be signed by the ordering Healthcare Provider indicating the following informed consent has been provided to the patient.*

**Visit our Prenatal Testing web page for details and limitations regarding prenatal testing.**

## MOTHER'S INFORMATION

| LAST (FAMILY) NAME | FIRST NAME | MI | DATE OF BIRTH (MM/DD/YYYY) |
|---|---|---|---|
| | | | |

**TEST(S) REQUESTED**

***This statement is required and applies to all cases of ongoing pregnancy.***

### My signature below indicates all of the following:

- I understand at least one parental specimen is required for any prenatal test for QA purposes.
- I understand that a back up cell culture is required for NGS and strongly recommended for other prenatal tests.
- I understand that prenatal testing will proceed when all test requirements are received and regardless of insurance coverage given the time-sensitive nature of many of these tests. Holds for benefit investigation can be requested (see page 8 of the PGxome Prenatal Diagnostic Test Requisition)
- I have explained the purpose of the prenatal testing I have requested, and I have provided appropriate genetic counseling to my patient.
- I have given the opportunity for the patient to ask questions.
- I am responsible for obtaining written or verbal informed consent (ensuring my patient understands risks, benefits and limitations of the testing and the implications of the results).

**HEALTHCARE PROVIDER SIGNATURE** | **PRINTED NAME** | **DATE**

### Retention of Unused DNA Statement for New York State Specimens

PreventionGenetics' general policy is to retain all excess DNA from patient testing indefinitely. This allows for easier ordering of additional testing in the future and saves considerable phlebotomy and shipping costs to the patient and healthcare system. Excess DNA specimens can also be used for quality control measures or for research on genetic variants associated with the diseases or conditions I was tested for, and any related diseases or conditions, which may include further testing of my retained samples, subject to approval by an Institutional Review Board or as otherwise permitted under applicable law. New York (NY) law requires patient consent in order to retain excess DNA beyond 60 days. If patient specimen was collected in NY and this statement is not signed, excess DNA will be discarded 30 days after testing is completed.

I authorize PreventionGenetics to retain unused DNA for potential future testing ordered by my Healthcare Provider and for the purposes described above.

**PATIENT OR LEGAL REPRESENTATIVE SIGNATURE** | **PRINTED NAME** | **DATE**

The following information should be used as a guide to provide informed consent to the patient and/or patient's family. The term "patient" in this consent can refer to the fetus, mother/person carrying the fetus, biological or intended parents. Testing must be ordered by a qualified Healthcare Provider.

**Purpose**
- The purpose of this test is to find the underlying genetic cause for the prenatal findings using Whole Exome Sequencing (WES).

**About PGxome Testing**
- This test involves the sequencing of thousands of genes at the same time, whereas many other genetic tests look at only one gene or small group of genes. Exome testing is performed via Next Generation Sequencing (NGS). Both sequence variants (SVs) and copy number variants (CNVs), also known as deletions/duplications are detected from NGS data. Variants that require further clarification will be confirmed with another technology such as Sanger sequencing, aCGH, MLPA, or PCR.
- An accepted specimen type (see PreventionGenetics.com) is required for each individual to perform testing. In rare instances a second specimen may be required.
- 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 PGxome Prenatal test description and Prenatal Testing Guidelines www.preventiongenetics.com/ClinicalTesting/TestCategory/PGxome.

**Family Testing**
- Testing of family members is invaluable 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, issues such as an undisclosed adoption, gamete donor, our uncertain paternity can interfere with accurate result interpretation. Inaccurate biological relationships are potentially identifiable with genetic testing. 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 interpret the patient's results and will be included in the patient's report. All sequence variants reported will include parental status. While large CNVs identified in the proband may include parental inheritance information, confirmation using an additional method will not be performed on parental specimens. 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".
- If family member(s) tested as part of PGxome Family desire their own PGxome analysis and test report, a separate completed diagnostic or health screen test requisition must be submitted. Full PGxome reports for family member(s) incur an additional charge per family member.

**Report Information**
- Genetic variants are defined as the differences between the patient's DNA and the human reference DNA.
- Generally only results that may explain the patient's clinical features are reported.
- In genes believed to be associated or possibly associated with the patient's clinical features, all Pathogenic, Likely Pathogenic, and Variants of Uncertain Significance (unknown if they cause disease) are reported.
- Other findings (aka "Secondary Findings" - see below) may be reported depending on the family'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 the 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.
- genetic counseling and/or clinical genetics consultation before and after testing is completed is recommended.
- 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, but not directly related to the reason testing was ordered. These are termed secondary findings. The patient may or may not wish to be informed of secondary findings.
- For PGxome Prenatal the patient and/or patient's family will have a choice if secondary findings are reported (see Test Selection section of the Test Requisition Form). Please consider the following carefully. Variants described in this section will only be reported if the patient OPTS IN.
  - Guideline Recommended Genes: The American College of Medical Genetics and Genomics recommends all labs performing WES report pathogenic variants in specific genes that cause certain, mostly dominantly inherited disorders (Version 3.2, Miller et al 2023. PubMed ID:37347242). 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.
  - Childhood Onset Disorders: The American College of Medical Genetics and Genomics recommends all labs performing prenatal WES report pathogenic or likely pathogenic variants detected in genes unrelated to the fetal clinical features, but known to cause moderate to severe childhood onset disorders (Monaghan et al. 2020. PubMed ID: 31911674). Many of these disorders, especially those associated with nonsyndromic intellectual disability/neurodevelopmental disorders and metabolic conditions, are not detectable with fetal imaging.
- Genetic variants related to complex disease, and mitochondrial disorders (excluding nuclear genes) will not be reported at this time. Heterozygous carrier variants for autosomal recessive or X-linked recessive disorders (in females) unrelated to the phenotype are not reported.
- Genetic variants in genes not currently known to be clinically relevant will not be reported.
- If testing reveals the 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 three 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. PreventionGenetics will provide WES data to an ordering provider upon request. PreventionGenetics does not supply software for data review and interpretation.

**Risks**
- 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 is generally unable 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 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**
- Confidentiality and patient privacy are taken very seriously. The laboratory is CAP and CLIA certified, and adheres to confidentiality laws related to protected health information.



**3800 S. Business Park Ave., Marshfield, WI 54449 USA • PreventionGenetics.com**

Phone: (715) 387-0484 · General Fax: (715) 406-4175 · Billing Fax: (715) 207-6602 · Email: support@preventiongenetics.com

CLIA 52D2065132 · CAP 7185561 · NPI 1114140571 · ISO 15189:2012 #3950.01

PREVENTION GENETICS part of EXACT SCIENCES

Test information is available on our website:
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PREVENTIONGENETICS USE ONLY

RESET FORM

**All testing must be ordered by a qualified Healthcare Provider**

A completed online order **OR** paper TRF and labeled specimen is required to initiate testing.

WHOLE EXOME SEQUENCING

# PGxome® PRENATAL DIAGNOSTIC REQUISITION

**The primary purpose of this test is for prenatal diagnosis. For WES in the case of fetal demise or pregnancy termination, our standard Diagnostic PGxome codes and forms can be used.**

## ORDERING CHECKLIST

Specimen(s): ☐ Fetal ☐ Biological Parent(s)
☐ Prenatal Healthcare Provider Statement (included)
☐ Clinical Features Checklist / Clinical Records

| PERSON COMPLETING FORM | CONTACT (DIRECT PHONE OR EMAIL) | DATE OF REQUEST (MM/DD/YYYY) |
|---|---|---|
| | | |

## FETAL AND MATERNAL INFORMATION

| LAST (FAMILY) NAME | MOTHER'S FIRST NAME (FETUS OF) | MI | MOTHER'S DATE OF BIRTH (MM/DD/YYYY) |
|---|---|---|---|
| | | | |

**MATERNAL ID CODE**

**FETAL SAMPLE TIME AND DATE COLLECTED***
_____ TIME ☐ AM ☐ PM _____ DATE (MM/DD/YYYY)

**FETAL SEX**
☐ Male ☐ Female ☐ Unknown ☐ Ambiguous
BASED ON: _____

**PRENATAL SPECIMEN SOURCE**
☐ Cell Culture, Source _____
☐ Direct Amniotic Fluid
☐ Fetal Blood (PUBS)
☐ Extracted DNA, Source _____
☐ Direct CVS
☐ Other, Source _____

☐ **SPECIMEN COLLECTED IN NEW YORK STATE**
Include New York State Genetic Testing Healthcare Provider Statement and New York State Non-Permitted Laboratory Test Request approval letter if test is not NY state approved. For a list of NY state approved tests, see website.

**WILL A BACK-UP SAMPLE/CELL CULTURE BE MAINTAINED AT ANOTHER LOCATION?**
☐ Yes ☐ No, *A back-up cell culture is required for NGS and strongly recommended for other prenatal tests. Please include cell culture with your order if needed.*

**ICD-10 CODES**
(required for insurance billing)
1 _____ PRIMARY
2 _____
3 _____

## ADDITIONAL MATERNAL INFORMATION

**If a maternal or gestational carrier specimen is received, maternal cell contamination (MCC) testing (Test Code #800) will always be performed.**

**MATERNAL SPECIMEN SOURCE**
☐ Whole Blood 5mL EDTA - Preferred
☐ Extracted DNA, Source _____
☐ Other, Source _____
☐ Saliva
☐ Buccal

☐ **SPECIMEN COLLECTED IN NEW YORK STATE**
Include New York State Genetic Testing Healthcare Provider Statement and New York State Non-Permitted Laboratory Test Request approval letter if test is not NY state approved. For a list of NY state approved tests, see website.

**DATE COLLECTED***
_____ TIME ☐ AM ☐ PM _____ DATE (MM/DD/YYYY)

**BLOOD TRANSFUSION**
☐ NO ☐ Within last 6 weeks,
DATE (MM/DD/YYYY) _____
TYPE _____

**BONE MARROW TRANSPLANT**
☐ NO ☐ YES, include date
DATE (MM/DD/YYYY) _____

**CLINICAL FEATURES**
☐ Unaffected ☐ Unknown ☐ Affected, features _____

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

**GEOANCESTRY / ETHNICITY**

## PREGNANCY HISTORY

**GESTATIONAL AGE AT SAMPLE COLLECTION**
_____ ☐ by U/S ☐ by LMP

**IS THIS AN ONGOING PREGNANCY?**
☐ No ☐ Yes

**DONOR PREGNANCY**
☐ No
☐ Yes _____

**MULTIPLE GESTATION PREGNANCY?**
☐ Twins ☐ Triplets ☐ Other _____
Chorionicity _____ Amnionicity _____

## PATERNAL INFORMATION

**Only required if specimen is sent for Prenatal PGxome Duo or Trio testing.**

| LAST (FAMILY) NAME | FIRST NAME | MI | DATE OF BIRTH (MM/DD/YYYY) |
|---|---|---|---|
| | | | |

**PATERNAL SPECIMEN SOURCE**
☐ Whole Blood 5mL EDTA - Preferred
☐ Extracted DNA, Source _____
☐ Other, Source _____
☐ Saliva
☐ Buccal

☐ **SPECIMEN COLLECTED IN NEW YORK STATE**
Include New York State Genetic Testing Healthcare Provider Statement and New York State Non-Permitted Laboratory Test Request approval letter if test is not NY state approved. For a list of NY state approved tests, see website.

**DATE COLLECTED (MM/DD/YYYY)***

**PATIENT ID CODE**

**BLOOD TRANSFUSION**
☐ NO ☐ Within last 6 weeks,
DATE (MM/DD/YYYY) _____
TYPE _____

**BONE MARROW TRANSPLANT**
☐ NO ☐ YES, include date
DATE (MM/DD/YYYY) _____

**CLINICAL FEATURES**
☐ Unaffected ☐ Unknown ☐ Affected, features _____

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

**GEOANCESTRY / ETHNICITY**

**If no collection date is provided, date of receipt will be used.*

## ADDITIONAL COMPARATORS

**Complete for PGxome Family Duo or Trio orders**

Please submit a separate completed diagnostic or health screen test requisition to request a full analysis of the comparator data for an additional charge, if desired.

| NAME (LAST, FIRST) | DATE OF BIRTH (MM/DD/YYYY) | SAMPLE TYPE | RELATIONSHIP TO PROBAND | AFFECTED?* |
|---|---|---|---|---|
| | | | | ☐ NO ☐ YES |
| | | | | ☐ NO ☐ YES |
| | | | | ☐ NO ☐ YES |

*If YES, must include clinical info.

## CLINICAL INFORMATION (REQUIRED)

**CLINICAL INDICATION**

☐ Abnormal fetal ultrasound (specify, attach report if available)
☐ Fetal loss / stillbirth / POC
☐ Abnormal amniotic fluid AFP: specify _____
☐ Other, specify, please call to discuss prior to submission _____

### ADDITIONAL CLINICAL INFORMATION

**RELEVANT CLINICAL INFORMATION.** We require the inclusion of detailed clinical notes/completion of the Clinical Features Checklist and a pedigree. The ability to interpret variants directly correlates with the quality of clinical information provided. ☐ Clinical records attached.

## PRENATAL PGxome TEST SELECTION

Include any special instructions in the comments section. See Prenatal Guidelines for more information. If a maternal specimen is received, maternal cell contamination (MCC) testing (Test Code #800) will always be performed. For WES in the case of fetal demise or pregnancy termination, our standard Diagnostic PGxome codes and forms can be used.

### PRENATAL PGxome

☐ **PATIENT ONLY** Test Code 14010
Rapid WES of fetus.

☐ **FAMILY DUO** Test Code 14012
Rapid WES of fetus and one comparator

☐ **FAMILY TRIO** Test Code 14013
Rapid WES of fetus and two comparator

☐ **FAMILY QUAD OR MORE** Test Code 14014
Rapid WES of fetus and three or more comparator

☐ Include family/comparator demographics (name, DOB, ID#, and relationship) on the proband report.

☐ **FETAL CELL CULTURE** Test Code 995
(only available for testing performed at PreventionGenetics)

### SECONDARY (ADDITIONAL) FINDINGS

**Details can be found in the PGxome Healthcare Provider Statement. Options for reporting of Secondary Findings are to be marked below.**

☐ OPT IN: GUIDELINE RECOMMENDED GENES
☐ OPT IN: CHILDHOOD ONSET DISORDERS

### SPECIAL INSTRUCTIONS

**All prenatal testing will be run at a STAT priority unless otherwise noted. Please indicate below if testing should be run at a standard TAT. Note, this results in a 25% cost reduction.**

☐ Run testing at standard priority.

COMMENTS:

PREVENTION GENETICS part of EXACT SCIENCES

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PREVENTIONGENETICS USE ONLY

**PATIENT**

**LAST NAME**

**FIRST NAME** | **MI**

## PROVIDER / LABORATORY CONTACT AND REPORTING

***Our preferred method of report transmission is uploading to our secure web portal, myPrevent.***
**Please provide an email address, when possible. If you have additional specific reporting requests, indicate them BELOW.**

### PROVIDER INFORMATION

**INSTITUTION**

**ADDRESS** | **CITY** | **STATE** | **ZIP**

**REQUESTING PHYSICIAN** (First, Last, Degree)

**EMAIL ADDRESS** (For report access via myPrevent)

**PHONE NUMBER** | **NPI#**

**REQUESTING GENETIC COUNSELOR OR ALLIED PROVIDER** (First, Last, Degree)

**EMAIL ADDRESS** (For report access via myPrevent)

**PHONE NUMBER** | **NPI#**

**IF YOU REQUIRE REPORTS TO BE TRANSMITTED VIA ANOTHER SECURE METHOD, SPECIFY HERE.**

As the ordering Healthcare Provider, I certify that: (1) I have obtained the patient's informed consent and family member's informed consent (as applicable) to perform this test as documented on a signed consent form that complies with applicable law and is consistent, in all material respects, with PreventionGenetics' Informed Consent form (available at https://assets.preventiongenetics.com/documents/patient-informed-consent.pdf), which I will maintain on file and make available to PreventionGenetics upon request; (2) The patient and their family member (as applicable) have been appropriately counseled and understand the risks, benefits, and limitations of this genetic testing and the implications of the results; and (3) I have received the patient's and family member's (as applicable) consent for PreventionGenetics to use and disclose information, test results, and sample as described in the consent form.

### SEND OUT LABORATORY

***COMPLETE ONLY IF REPORT IS NEEDED***

**INSTITUTION / CONTACT**

**ADDRESS** | **CITY** | **STATE** | **ZIP**

**EMAIL ADDRESS** (For report access via myPrevent)

**PHONE NUMBER**

**NPI#** (where applicable)

**IF YOU REQUIRE REPORTS TO BE TRANSMITTED VIA ANOTHER SECURE METHOD, SPECIFY HERE.**

**ADDITIONAL ACCESS TO REPORTS** List additional Healthcare Providers and their emails to allow access to reports

## INSTITUTION BILLING

**PATIENT TESTING WILL PROCEED WHEN ALL BILLING INFORMATION HAS BEEN RECEIVED.**

***IF INSTITUTIONAL BILLING IS SELECTED, PAGE 4 IS NOT REQUIRED.***

☐ Send invoice to the contact information above. Please provide PO number below if applicable.

**BILLING INSTITUTION** | **PO NUMBER**

**CONTACT** | **PHONE NUMBER** | **EMAIL**

**ADDRESS** | **CITY** | **STATE** | **ZIP**

**BILLING ACCOUNT NUMBER** ☐ **UPDATED INFO**

**EMAIL INVOICE VIA SECURE EMAIL** (provide email address

**ACCESS TO TEST REPORT(S) FOR BILLING**

☐ **EMAIL ADDRESS**
(For report access via myPrevent) _____

☐ **OTHER** (specify) _____

PREVENTION GENETICS part of EXACT SCIENCES

Test information is available on our website:
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PREVENTIONGENETICS USE ONLY

**PATIENT**

**LAST NAME**

**FIRST NAME** | **MI**

## COMPLETE THIS FORM FOR PATIENT PAY AND/OR INSURANCE BILLING

***PATIENT TESTING WILL PROCEED WHEN ALL BILLING INFORMATION HAS BEEN RECEIVED.***

### ** THIS SECTION MUST BE FILLED OUT COMPLETELY **

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

### ACCEPTANCE of financial responsibility for genetic testing

***SIGNATURE REQUIRED BELOW TO PROCEED WITH TESTING.***

**MY SIGNATURE INDICATES I ACCEPT FINANCIAL RESPONSIBILITY FOR ALL FEES ASSOCIATED WITH THIS GENETIC TESTING ORDER.** If applicable, 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. I understand my out-of-network benefits may apply. PreventionGenetics may contact me to resolve any billing-related issues and to request payment.

**SIGN HERE:** Required to process form

_____ _____ _____

**PATIENT / RESPONSIBLE PARTY SIGNATURE** | **PRINTED NAME OF RESPONSIBLE PARTY** | **DATE**

## CREDIT CARD PAYMENT

- **PATIENT PROMPT PAY (excludes insurance billing)**
  Card information provided below will be charged when specimen arrives. The 10% Patient Prompt Pay discount will apply.
- **PATIENT PAY - INSURANCE BILLING**
  Card information provided below will be charged when the claim is processed. The 10% Patient Prompt Pay discount **WILL NOT** apply.

**CREDIT CARD INFORMATION**

| 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.**

**SIGN HERE:** Required to process credit card

_____ _____

**CREDIT CARD HOLDERS SIGNATURE** | **DATE**

## INSURANCE INFORMATION - IF APPLICABLE

**INDICATE THE TYPE OF INSURANCE** ☐ **Attach a copy of Insurance Card (both sides)**

☐ **PRIVATE** ☐ **TRICARE** *include signed Tricare waiver* ☐ **MEDICARE** *include signed ABN form* ☐ **MEDICAID** *Visit PreventionGenetics.com for in-network Medicaid plans.*

| POLICY HOLDER NAME | DATE OF BIRTH (MM/DD/YYYY) | RELATIONSHIP TO PATIENT |
|---|---|---|
| | | |

| PRIMARY INSURANCE COMPANY NAME (REQUIRED) | PHONE NUMBER |
|---|---|
| | |

| POLICY ID# | GROUP # | AUTHORIZATION # ☐ *Attach copy of authorization, PreventionGenetics must be listed as servicing provider.* |
|---|---|---|
| | | |

**SECONDARY INSURANCE** ☐ **Attach a copy of Insurance Card (both sides)**

**Prenatal testing will proceed when all test requirements are received and regardless of insurance coverage given the time-sensitive nature of many of these tests. Testing will be held for the following situation only:**

- To obtain required in-network pre-authorization.

Indicate if testing should be held for the following:

☐ For benefit investigation / pre-authorization and share results with patient directly via email provided.

☐ Other: _____.

**Note: holds placed on testing will extend overall TAT.**

**NOTE:** Prenatal CMA, re-analysis, and cell cultures cannot be canceled once a sample is received. Testing placed on hold will extend overall TAT.

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PREVENTIONGENETICS USE ONLY

RESET FORM

**All testing must be ordered by a qualified Healthcare Provider**

A completed online order **OR** paper TRF and labeled specimen is required to initiate testing.

## CLINICAL INFORMATION IS REQUIRED for PGnome®, PGxome®, and PGmax™ panels.

Orders **MUST** include the completed clinical features checklist (preferred) or clinical notes/records. Completion of the checklist is strongly encouraged for all panel testing. The ability to interpret variants directly correlates with the quality of clinical information provided. Also include family medical history/pedigree, if available.

# CLINICAL FEATURES

| PERSON COMPLETING FORM | CONTACT (DIRECT PHONE OR EMAIL) | DATE OF REQUEST (MM/DD/YYYY) |
|---|---|---|
| | | |

## PATIENT INFORMATION

| LAST (FAMILY) NAME | FIRST NAME | MI | DATE OF BIRTH (MM/DD/YYYY) |
|---|---|---|---|
| | | | |

| PATIENT ID | HAS PATIENT BEEN TESTED PREVIOUSLY AT PREVENTIONGENETICS? | BIOLOGICAL SEX |
|---|---|---|
| | ☐ NO<br>☐ YES, PG ID# _____ | ☐ Male ☐ Female ☐ Other<br>SPECIFY KARYOTYPE _____ |

## CLINICAL INFORMATION (CHECK ALL THAT APPLY)

### PRE/PERINATAL

- ☐ Abnormality of septum pellucidum
  - ☐ Absent septum pellucidum
  - ☐ Cavum septum pellucidum
- ☐ Choroid plexus cyst (CPC)
- ☐ Absent nasal bone
- ☐ Congenital heart defect
- ☐ Intracardiac echogenic focus (IEF)
- ☐ Cystic hygroma
- ☐ Increased nuchal translucency, Size (mm): _____
- ☐ Pleural effusion
- ☐ Pericardial effusion
- ☐ Generalized edema
- ☐ Fetal ascites
- ☐ Hydrops fetalis
- ☐ Diaphragmatic hernia
- ☐ Absent stomach bubble
- ☐ Omphalocele
- ☐ Gastroschisis
- ☐ Echogenic bowel
- ☐ Fetal pyelectasis/hydronephrosis
- ☐ Decreased fetal movement
- ☐ Encephalocele
- ☐ Myelomeningocele/Spina bifida
- ☐ Sacrococcygeal teratoma
- ☐ Intrauterine growth retardation (IUGR)
- ☐ Small for gestational age (SGA)
- ☐ Oligohydramnios
- ☐ Polyhydramnios
- ☐ Short long bones
- ☐ Small thorax
- ☐ Fetal demise
- ☐ Prematurity, Gestational Age: _____
- ☐ Other: _____

### STRUCTURAL BRAIN ABNORMALITIES / IMAGING

- ☐ Abnormal/delayed myelination
- ☐ Abnormality of basal ganglia
- ☐ Abnormality of brainstem
- ☐ Abnormality of white matter:
  - ☐ Periventricular
  - ☐ Other: _____
- ☐ Abnormality of cerebral ventricles:
  - ☐ Colpocephaly
  - ☐ Hydrocephalus
  - ☐ Ventriculomegaly
- ☐ Abnormality of corpus callosum morphology:
  - ☐ Agenesis
    - ☐ Complete
    - ☐ Partial
  - ☐ Aplasia/hypoplasia
- ☐ Aplasia/hypoplasia of cerebellar vermis
- ☐ Aplasia/hypoplasia of cerebellum
- ☐ Arnold-Chiari malformation:
  - ☐ Type I
- ☐ Cerebral atrophy/hypoplasia
- ☐ Cerebral calcification
- ☐ Holoprosencephaly
- ☐ Intraventricular hemorrhage
  - ☐ Preterm Intraventricular hemorrhage
- ☐ Iron deposition
- ☐ Leukodystrophy
- ☐ Neuronal migration abnormality
  - ☐ Cortical gyration
  - ☐ Gray matter heterotopia
- ☐ Other: _____

### DEVELOPMENTAL/ BEHAVIORAL

- ☐ Aggressive/violent behavior
- ☐ Anxiety
- ☐ Attention-deficit hyperactivity disorder
- ☐ Autistic behavior
- ☐ Autism/autism spectrum disorder
- ☐ Cognitive impairment
- ☐ Delayed fine motor development
- ☐ Delayed gross motor development
- ☐ Developmental regression
- ☐ Gait disturbance Specify: _____
- ☐ Global developmental delay
- ☐ Hyperactivity
- ☐ Incoordination
- ☐ Intellectual disability
  - ☐ Mild
  - ☐ Moderate
  - ☐ Severe/profound
- ☐ Learning disability
- ☐ Language impairment
  - ☐ Absent speech
  - ☐ Apraxia
  - ☐ Articulation difficulties
  - ☐ Delayed speech and language development
    - ☐ Expressive
    - ☐ Receptive
  - ☐ Dysarthria
  - ☐ Echolalia
  - ☐ Loss of speech
- ☐ Memory impairment
- ☐ Obsessive-compulsive behavior
- ☐ Self-injurious behavior:
  - ☐ Biting
  - ☐ Head-banging
  - ☐ Skin picking
- ☐ Sensory processing disorder/ neurodevelopmental abnormality
- ☐ Sleep disturbance
- ☐ Stereotypy
  - ☐ Recurrent hand flapping
  - ☐ Stereotypical hand wringing
- ☐ Other: _____

### NEUROLOGICAL

- ☐ Abnormality of nervous system
- ☐ Ataxia
- ☐ Athetosis
- ☐ Bradykinesia
- ☐ Cerebral palsy
- ☐ Chorea
- ☐ Cortical visual impairment
- ☐ Dementia
- ☐ Dysarthria
- ☐ Dyskinesia
- ☐ Dysphagia
- ☐ Dystonia
- ☐ Encephalopathy
- ☐ Gait disturbance, Specify: _____
- ☐ Headache
- ☐ Hemiplegia
- ☐ Hypotonia
- ☐ Hypertonia
- ☐ Infantile spasms
- ☐ Migraine
- ☐ Myoclonus
- ☐ Neuropathy
  - ☐ Peripheral
  - ☐ Sensory
- ☐ Parkinsonism/Parkinson Disease
- ☐ Seizures, Type: _____
- ☐ Spasticity
- ☐ Syncope
- ☐ Tremors
- ☐ Vertigo
- ☐ Other: _____

### CRANIOFACIAL/ DYSMORPHISM

- ☐ Abnormal facial shape, Specify: _____
- ☐ Abnormality of incisors, Specify: _____
- ☐ Ala nasi
  - ☐ Cleft
  - ☐ Thick
  - ☐ Underdeveloped
- ☐ Anteverted nares
- ☐ Brachycephaly

- [ ] Chin abnormality, Specify: _____
- [ ] Cleft lip:
  - [ ] Unilateral
  - [ ] Bilateral
  - [ ] Midline
- [ ] Cleft palate:
  - [ ] Unilateral
  - [ ] Bilateral
  - [ ] Midline
  - [ ] Submucous cleft
- [ ] Cloverleaf skull
- [ ] Columella abnormality:
  - [ ] Broad
  - [ ] High insertion
  - [ ] Low hanging
  - [ ] Low insertion
  - [ ] Short
- [ ] Craniosynostosis:
  - [ ] Coronal
  - [ ] Lambdoidal
  - [ ] Metopic
  - [ ] Orbital
  - [ ] Sagittal
- [ ] Dolichocephaly
- [ ] Face abnormality:
  - [ ] Broad
  - [ ] Coarse facial features
  - [ ] Flat
  - [ ] Long
  - [ ] Narrow
  - [ ] Round
  - [ ] Short
  - [ ] Square
  - [ ] Triangular
- [ ] Forehead abnormality:
  - [ ] Broad
  - [ ] Narrow
  - [ ] Prominent
  - [ ] Sloping
  - [ ] Creases
- [ ] Frontal bossing
- [ ] Jaw abnormality:
  - [ ] Broad
  - [ ] Narrow
- [ ] Lip vermilion abnormality
- [ ] Lip abnormality:
  - [ ] Pit
  - [ ] Thin
  - [ ] Thick
  - [ ] Tented
  - [ ] Exaggerated cupid's bow
  - [ ] Absent cupid's bow
- [ ] Malar abnormality:
  - [ ] Flattening
  - [ ] Prominence
- [ ] Midface abnormality:
  - [ ] Flat
  - [ ] Prominence
  - [ ] Retrusion
- [ ] Macrocephaly:
  - [ ] Relative
  - [ ] True
- [ ] Metopic suture abnormality:
  - [ ] Depression
  - [ ] Ridge
- [ ] Microcephaly
- [ ] Micrognathia
- [ ] Nasal base abnormality:
  - [ ] Narrow
  - [ ] Wide
- [ ] Nasal bridge abnormality:
  - [ ] Depressed
  - [ ] Narrow
  - [ ] Prominent
  - [ ] Short
  - [ ] Wide
- [ ] Nasal cartilage, absent
- [ ] Nasal ridge abnormality:
  - [ ] Depressed
  - [ ] Narrow
  - [ ] Wide
- [ ] Nasal tip abnormality:
  - [ ] Bifid
  - [ ] Broad
  - [ ] Depressed
  - [ ] Deviated
  - [ ] Narrow
  - [ ] Overhanging
- [ ] Nasolabial fold abnormality:
  - [ ] Prominent
  - [ ] Underdeveloped
- [ ] Neck abnormality:
  - [ ] Broad
  - [ ] Long
  - [ ] Webbed
  - [ ] Short
  - [ ] Redundant nuchal skin
- [ ] Nose abnormality:
  - [ ] Absent
  - [ ] Bifid
  - [ ] Long
  - [ ] Narrow
  - [ ] Prominent
  - [ ] Short
  - [ ] Wide
- [ ] Occiput abnormality:
  - [ ] Flat
  - [ ] Prominent
- [ ] Plagiocephaly
- [ ] Philtrum abnormality:
  - [ ] Broad
  - [ ] Deep
  - [ ] Hypoplastic
  - [ ] Long
  - [ ] Narrow
  - [ ] Smooth
  - [ ] Short
  - [ ] Tented
- [ ] Proboscis
- [ ] Prognathism
- [ ] Retrognathia
- [ ] Scaphocephaly
- [ ] Supraorbital ridge abnormality:
  - [ ] Prominent
  - [ ] Underdeveloped
- [ ] Trigonocephaly
- [ ] Turricephaly
- [ ] Other:_____

## EYES/VISION

Age of onset of vision issues: _____

- [ ] Abnormality of eye movement
  - [ ] Esotropia
  - [ ] Exotropia
  - [ ] Nystagmus
  - [ ] Smooth pursuit
  - [ ] Strabismus
  - [ ] Other: _____
- [ ] Abnormality of vision, Specify: _____
- [ ] Abnormal anterior eye segment morphology
- [ ] Ablepharon
- [ ] Achromatopsia
- [ ] Aniridia
- [ ] Ankyloblepharon
- [ ] Anophthalmia
- [ ] Blepharochalasis
- [ ] Blepharophimosis
- [ ] Cataracts
- [ ] Cataracts, congenital
- [ ] Coloboma
- [ ] Corneal opacity
- [ ] Corneal dystrophy
- [ ] Cone/cone-rod dystrophy
- [ ] Congenital stationary night blindness
- [ ] Cryptophthalmos
- [ ] Deeply set eyes
- [ ] Distichiasis
- [ ] Dyschromatopsia (color blindness)
- [ ] Ectopia lentis
- [ ] Ectropion
- [ ] Entropion
- [ ] Epiblepharon
- [ ] Epicanthus/epicanthal folds
- [ ] Epicanthus inversus
- [ ] Eyebrow abnormality:
  - [ ] Broad
  - [ ] Highly arched
  - [ ] Horizontal
  - [ ] Sparse
  - [ ] Thick
- [ ] Eyelash abnormality:
  - [ ] Absent
  - [ ] Long
  - [ ] Prominent
  - [ ] Sparse
- [ ] Eyelid cleft
- [ ] External ophthalmoplegia
  - [ ] Progressive
- [ ] Glaucoma
- [ ] Infraorbital abnormality:
  - [ ] Crease
  - [ ] Fold
- [ ] Iris abnormality, Specify: _____
- [ ] Lagophthalmos
- [ ] Leber optic atrophy
- [ ] Lens subluxation
- [ ] Macular abnormality, Specify: _____
- [ ] Macular dystrophy
- [ ] Microphthalmia
- [ ] Myopia
- [ ] Ocular albinism
- [ ] Optic atrophy
- [ ] Optic neuropathy
- [ ] Palpebral fissure abnormality:
  - [ ] Downslanted
  - [ ] Upslanted
  - [ ] Long
  - [ ] Short
  - [ ] Almond-shaped
- [ ] Proptosis
- [ ] Ptosis
- [ ] Retinal flecks
- [ ] Retinal detachment
- [ ] Retinitis pigmentosa
- [ ] Synophrys
- [ ] Telecanthus
- [ ] Other:_____

## EARS/HEARING

Age of onset of hearing loss:_____

- [ ] Hearing impairment
  - [ ] Sensorineural
    - [ ] Congenital
    - [ ] Bilateral
    - [ ] Progressive
  - [ ] Conductive
    - [ ] Congential
    - [ ] Bilateral
    - [ ] Progressive
  - [ ] Mixed
- [ ] Anotia
- [ ] Abnormal newborn screen, Specify:_____
- [ ] Antihelix abnormality:
  - [ ] Absent
  - [ ] Additional crus
  - [ ] Angulated
  - [ ] Inferior crus broad
  - [ ] Inferior crus prominent
  - [ ] Inferior crus underdeveloped
  - [ ] Superior crus prominent
  - [ ] Superior crus underdeveloped
- [ ] Antitragus abnormality:
  - [ ] Absent
  - [ ] Bifid
  - [ ] Everted
  - [ ] Prominent
  - [ ] Underdeveloped
- [ ] Ear abnormality:
  - [ ] Abnormality of the tragus
  - [ ] Auricular pit
  - [ ] Crumpled
  - [ ] Cupped
  - [ ] Long
  - [ ] Low-set
  - [ ] Posteriorly rotated
  - [ ] Preauricular pit
  - [ ] Protruding
  - [ ] Short
  - [ ] Satyr
  - [ ] Tag

- ☐ Helix abnormality:
  - ☐ Cleft / Notching
  - ☐ Crimped
  - ☐ Darwin notch
  - ☐ Darwin tubercle
  - ☐ Notching
  - ☐ Overfolded
  - ☐ Prominent
  - ☐ Thin
- ☐ Lobe abnormality:
  - ☐ Cleft
  - ☐ Forward-facing
  - ☐ Large
  - ☐ Small
  - ☐ Uplifted
- ☐ Macrotia
- ☐ Other:_____

**ENDOCRINE**

- ☐ Adrenal insufficiency (Addison)
- ☐ Androgen excess
- ☐ Androgen insensitivity
- ☐ Congenital adrenal hypoplasia
- ☐ Congenital adrenal hyperplasia
- ☐ Delayed bone age
- ☐ Delayed puberty
- ☐ Diabetes insipidus
- ☐ Diabetes Mellitus
- ☐ Hyperandrogenism
- ☐ Hyperglycemia
- ☐ Hyperphosphatemia
- ☐ Hyperthyroidism
- ☐ Hypoglycemia
- ☐ Hypophosphatemia
- ☐ Hypothyroidism
- ☐ Increased cortisol level (Cushing)
- ☐ Maturity-onset diabetes of the young
- ☐ Precocious puberty
- ☐ Rickets
- ☐ Other:_____

**RESPIRATORY**

- ☐ Asthma
- ☐ Bronchiectasis
- ☐ Bronchomalacia
- ☐ Hyperventilation
- ☐ Hypoventilation
- ☐ Laryngomalacia
- ☐ Laryngeal cleft
- ☐ Pneumothorax
- ☐ Pulmonary fibrosis
- ☐ Respiratory insufficiency
- ☐ Tracheomalacia
- ☐ Tracheoesophageal fistula
- ☐ Other:_____

**HEMATOLOGIC/IMMUNOLOGIC**

- ☐ Agammaglobulinemia
- ☐ Allergic rhinitis
- ☐ Anemia
- ☐ Hemolytic anemia
- ☐ Immunodeficiency, Specify:_____
- ☐ Lymphopenia
- ☐ Neutropenia
- ☐ Pancytopenia
- ☐ Recurrent infections
- ☐ Severe combined immunodeficiency
- ☐ Thrombocytopenia
- ☐ Other: _____

**SKIN/HAIR**

- ☐ Abnormal blistering of the skin, Specify:_____
- ☐ Abnormality of nail:
  - ☐ Broad
  - ☐ Deep-set
  - ☐ Pits
- ☐ Albinism
- ☐ Alopecia
- ☐ Anhidrosis
- ☐ Cafe-au-lait spot:
  - ☐ Single
  - ☐ Multiple
- ☐ Coarse hair
- ☐ Collodion baby
- ☐ Cutaneous photosensitivity
- ☐ Cutis laxa
- ☐ Dry skin
- ☐ Eczema
- ☐ Erythematous skin
- ☐ Hemangioma
- ☐ Hairline:
  - ☐ Anterior
    - ☐ Low
    - ☐ High
  - ☐ Posterior
    - ☐ Low
    - ☐ High
- ☐ Hyperextensible skin
- ☐ Hyperpigmentation of the skin
- ☐ Hypopigmentation of the skin
- ☐ Hypohidrosis
- ☐ Ichthyosis
- ☐ Jaundice
- ☐ Lipoma
- ☐ Lymphedema
- ☐ Palmoplantar keratoderma
- ☐ Scarring of skin
- ☐ Skin rash
- ☐ Sparse hair
- ☐ Telangiectasia
- ☐ Vascular skin abnormality
- ☐ Velvety skin
- ☐ Other:_____

**CARDIAC**

- ☐ Amyloidosis
- ☐ Aortic root dilatation
- ☐ Arrhythmia
- ☐ Atrial septal defect
- ☐ Atrioventricular canal defect
- ☐ Arrhythmogenic right ventricular dysplasia
- ☐ Bicuspid aortic valve
- ☐ Bradycardia
- ☐ Coarctation of the aorta
- ☐ Congenital heart defect
- ☐ Dilated cardiomyopathy
- ☐ Double outlet right ventricle
- ☐ Ebstein anomaly
- ☐ Heterotaxy
- ☐ Hypertension
- ☐ Hypertrophic cardiomyopathy
- ☐ Mitral valve prolapse
- ☐ Noncompaction cardiomyopathy
- ☐ Patent ductus arteriosus
- ☐ Patent foramen ovale
- ☐ Prolonged QTc interval
- ☐ Pulmonary hypertension
  - ☐ Arteria
  - ☐ Vascular
- ☐ Sudden death
- ☐ Tetralogy of Fallot
- ☐ Transposition of the great vessels
- ☐ Truncus arteriosus
- ☐ Ventricular septal defect
- ☐ Ventricular tachycardia
- ☐ Other:_____

**GASTROINTESTINAL**

- ☐ Biliary atresia
- ☐ Cholestasis
- ☐ Constipation:
  - ☐ Acute
  - ☐ Chronic
- ☐ Diarrhea
- ☐ Diaphragmatic hernia
- ☐ Duodenal stenosis/atresia
- ☐ Esophageal stenosis/atresia
- ☐ Exocrine pancreatic insufficiency
- ☐ Failure to thrive
- ☐ Feeding difficulties
- ☐ Gastroesophageal reflux
- ☐ Gastroschisis
- ☐ Hepatomegaly
- ☐ Hepatosplenomegaly
- ☐ Inflammatory bowel disease
- ☐ Jaundice
- ☐ Liver disease
- ☐ Liver failure
- ☐ Nausea
- ☐ Omphalecele
- ☐ Pancreatitis
- ☐ Pyloric stenosis
- ☐ Splenomegaly
- ☐ Tracheoesophageal fistula
- ☐ Tube feeding
  - ☐ Nasogastric
  - ☐ Gastrostomy
  - ☐ Gastrojejunal
- ☐ Umbilical hernia
- ☐ Vomiting
- ☐ Other:_____

**GENITOURINARY**

- ☐ Abnormality of the uterus, Specify: _____
- ☐ Ambiguous genitalia
- ☐ Chordee
- ☐ Cryptorchidism
- ☐ Duplicated collecting system
- ☐ Horseshoe kidney
- ☐ Hydronephrosis
- ☐ Hypospadias/epispadias
- ☐ Inguinal hernia
- ☐ Micropenis
- ☐ Multicystic kidney dysplasia
- ☐ Nephrolithiasis
- ☐ Polycystic kidney disease
- ☐ Renal agenesis/hypoplasia
  - ☐ Unilateral agneisis
  - ☐ Bilateral ageneisis
  - ☐ Unilateral hypoplasia
  - ☐ Blateral hypoplasia
- ☐ Sex reversal
- ☐ Vesicoureteral reflux
- ☐ Other:_____

**MUSCULOSKELETAL**

- ☐ Abnormal connective tissue
- ☐ Abnormal digit morphology
  - ☐ Broad
  - ☐ Short
  - ☐ Clinodactyly
  - ☐ Ectrodatyly
  - ☐ Oligodactyly
  - ☐ Polydactyly
    - ☐ Postaxial
    - ☐ Preaxial
  - ☐ Syndactyly
- ☐ Arachnodactyly
- ☐ Arthralgia
- ☐ Arthrogryposis
- ☐ Bruising susceptibility
- ☐ Chest abnormality:
  - ☐ Small chest
  - ☐ Barrel-shaped
  - ☐ Bell-shaped thorax
  - ☐ Pectus carinatum
  - ☐ Pectus excavatum
- ☐ Contractures of joint(s)
- ☐ Decreased muscle mass
- ☐ Delayed bone age
- ☐ Dolichostenomelia
- ☐ Exercise intolerance
- ☐ Fatigue
- ☐ Fracture(s)
- ☐ Hemihypertrophy
- ☐ Hypertonia
- ☐ Hypotonia
- ☐ Joint hypermobility
- ☐ Kyphosis
- ☐ Limb shortening:
  - ☐ Mesomelic
  - ☐ Micromelic
  - ☐ Rhizomelic
- ☐ Metaphyseal abnormalities:
  - ☐ Dumbbell

- ☐ Flared
- ☐ Muscle weakness
- ☐ Myalgia
- ☐ Myopathic facies
- ☐ Myopathy
- ☐ Myelomeningocele/Spina Bifida/ Neural Tube Defect
- ☐ Osteoarthritis
- ☐ Osteoporosis
- ☐ Osteopenia
- ☐ Pain:
  - ☐ Absent/decreased
  - ☐ Abnormal sensation
  - ☐ Episodic
  - ☐ Limb
  - ☐ Muscle
- ☐ Platyspondyly
- ☐ Recurrent fractures
- ☐ Rhabdomyolysis
- ☐ Rib abnormality:
  - ☐ Cupped
  - ☐ Fused
  - ☐ Supernumerary
  - ☐ Missing
  - ☐ Short
  - ☐ Spatulate
- ☐ Other:
- ☐ Rickets
- ☐ Scoliosis
- ☐ Short stature
- ☐ Skeletal dysplasia
- ☐ Talipes
  - ☐ Equinovarus
  - ☐ Other:
- ☐ Tall stature
- ☐ Thoracic dysplasia
- ☐ Thumb abnormality:
  - ☐ Adducted
  - ☐ Broad
  - ☐ Triphalangeal
- ☐ Vertebral bodies, abnormal form
  - ☐ Aplasia/hypoplasia
  - ☐ Butterfly
  - ☐ Fusion
  - ☐ Hemivertebrae
- ☐ Other:

**VASCULAR SYSTEM**

- ☐ Aneurysm
- ☐ Aortic:
  - ☐ Abdominal
  - ☐ Dissecting
  - ☐ Thoracic
- ☐ Cerebral
  - ☐ Other:
- ☐ Arterial calcification
- ☐ Arterial dissection
- ☐ Arterial tortuosity
- ☐ Arteriovenous malformation
- ☐ Epistaxis
- ☐ Lymphedema
- ☐ Pulmonary hypertension:
  - ☐ Arterial
  - ☐ Vascular
- ☐ Stroke
- ☐ Other:

**OTHER TESTING**
***Provide copy of report(s)***

Echocardiogram: _____
EEG: _____
EMG/NCV: _____
Biopsy: _____
Gene testing: _____
Results: _____

***If you would like us to comment on the presence/ absence of previously identified variants, provide a copy of the original report.***

Chromosomal Microarray (CMA): _____
MRI brain: _____
MRI (other): _____
CT brain: _____
CT (other): _____
Muscle biopsy: _____
Ultrasound: _____
X-Ray: _____

## METABOLIC FINDINGS • *Attach relevant lab reports and values.*

- ☐ Abnormal newborn screen
  Specify: _____

### Abnormal metabolic profile
***(please check each metabolite outside normal limits)***

- ☐ Acylcarnitine _____
- ☐ Acylglycines _____
- ☐ Amino Acids _____
- ☐ Amylase _____
- ☐ Biotindase _____
- ☐ Carnitine _____
- ☐ Cerebrospinal fluid _____
- ☐ Coenyzme/enzyme activity _____
- ☐ Creatine phosphokinase (CPK) _____
- ☐ Essential fatty acids _____
- ☐ Folate _____
- ☐ Hepatic Transaminase _____
- ☐ Homocysteine _____
- ☐ Hormones _____
- ☐ Ketones _____
- ☐ Lactic acidosis _____
- ☐ Lipase _____
- ☐ Lipoproteins _____
- ☐ Lysosomal enzymes _____
- ☐ Mucopolysaccharides _____
- ☐ Oligosaccharides _____
- ☐ Porphyrin _____
- ☐ Pterins _____
- ☐ Purines _____
- ☐ Pyrimidine _____
- ☐ Pyruvate _____
- ☐ Serum alpha fetoprotein (AFP) _____
- ☐ Sterols/Oxysterols _____
- ☐ Transferrin _____
- ☐ Uric acid _____
- ☐ Very long chain fatty acids (VLCFA) _____

### Abnormal vitamin levels
***(please check each vitamin measuring outside normal limits)***

- ☐ Copper _____
- ☐ Magnesium _____
- ☐ Manganese _____
- ☐ Vitamin B6 _____
- ☐ Vitamin B12 _____
- ☐ Vitamin D _____
- ☐ Zinc _____
- ☐ Other _____

### Other metabolic features

- ☐ Abnormal cerebrospinal fluid (CSF) studies _____
- ☐ Abnormal glycosylation _____
- ☐ Abnormal mitochondrial respiratory chain activity _____
- ☐ Hyperammonemia _____
- ☐ Hyperbilirubinemia _____
- ☐ Hyperglycemia _____
- ☐ Hyperlipidemia _____
- ☐ Hypoglycemia _____
- ☐ Hypolipidemia _____
  - ☐ Plasma _____
  - ☐ Urine _____
- ☐ Lactic Acidosis _____
- ☐ Metabolic Acidosis _____
- ☐ Methylmalonic aciduria _____
- ☐ Methylmalonic acidemia _____

PREVENTION GENETICS part of EXACT SCIENCES

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

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**PATIENT**

LAST NAME

FIRST NAME | MI

# CANCER HISTORY

## Patient Information

☐ **No personal history of cancer**

☐ **Breast**
Age of diagnosis: _____
- ☐ Triple-Negative (ER, PR, Her2 negative)
- ☐ DCIS (Ductal Carcinoma In Situ)
- ☐ DC (Invasive Ductal Carcinoma)
- ☐ ILC (Invasive Lobular Carcinoma)
- ☐ Bilateral / >1 Primary

☐ **Ovarian/Fallopian Tube / Primary Peritoneal**
Age of diagnosis: _____

☐ **Colorectal**
Age of diagnosis: _____
MSI/IHC results: _____

☐ **Endometrial / Uterine**
Age of diagnosis: _____
MSI/IHC results: _____

☐ **Pancreatic**
Age of diagnosis: _____

☐ **Prostate**
Age of diagnosis: _____
Metastatic ☐ Yes ☐ No ☐ Unknown
Gleason Score _____

**Polyps**
Age of diagnosis: _____
Number of polyps: _____
Pathology details: _____

**Other**
Age of diagnosis: _____
Details: _____

## Family History of Cancer or Include Pedigree

☐ **No known family history of cancer**

☐ **Limited Family Structure** Limited family history available such as fewer than two female first or second-degree maternal or paternal relatives having lived beyond age 45

**Ashkenazi Jewish** ☐ NO ☐ YES, Maternal ☐ Yes, Paternal ☐ Unknown

| RELATION TO PATIENT | SELECT | CANCER / POLYP TYPE / GLEASON SCORE | AGE OF DIAGNOSIS | UNAVAILABLE FOR TESTING | RELATIVE IS DECEASED | PATIENT HAS NO CONTACT WITH WITH RELATIVE | RELATIVE DECLINES TESTING |
|---|---|---|---|---|---|---|---|
| | ☐ Maternal ☐ Paternal | | | ☐ | ☐ | ☐ | ☐ |
| | ☐ Maternal ☐ Paternal | | | ☐ | ☐ | ☐ | ☐ |
| | ☐ Maternal ☐ Paternal | | | ☐ | ☐ | ☐ | ☐ |
| | ☐ Maternal ☐ Paternal | | | ☐ | ☐ | ☐ | ☐ |
| | ☐ Maternal ☐ Paternal | | | ☐ | ☐ | ☐ | ☐ |

**PAST FAMILY GENETIC TESTING** ☐ NO previous testing in family. ☐ YES, *Include Germline, Somatic or Tumor testing results.* ***Describe or attach copies of report.***

**KNOWN FAMILIAL VARIANT:** GENE _____ VARIANT _____

# PEDIGREE

Use this area to include a pedigree and/or additional relevant medical/family history.