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

Prenatal - Healthcare Providers Statement

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.
- For prenatal targeted testing (Test Code #990), familial positive control specimens are required.
- 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 4 of the Prenatal 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** _____

Test Description, Methods, and Limitations

See specific full test description at PreventionGenetics.com for information about clinical features, genetics, indications for testing, test procedure, test description, clinical sensitivity, analytical validity, analytical limitations, and turnaround times. **Please visit our Prenatal testing web page for details and limitations regarding prenatal testing**.

Purpose of Testing (mark all that apply)

☐ Diagnosis

☐ Other (please describe):

_____

_____

_____

Results

- Positive genetic testing results may mean a person carries or has the condition or disease being tested. Often this means Pathogenic or Likely Pathogenic genetic variation(s) has/have been identified. Consulting with a Physician or Genetic Counselor prior to and after completion of testing is recommended to learn the full meaning of the results and their implications.
- Negative results may mean, within limitations of the test, no Pathogenic or Likely Pathogenic genetic variations were identified. However, consultation with a Medical Geneticist, Genetic Counselor or Specialty Provider is recommended should the patient or Ordering Provider have additional questions or concerns.
- Uncertain results may mean a Variant(s) of Uncertain Significance (VUS) was/were identified. It is not clear if these variants are linked to the patient's phenotype or are associated with disease.
- Pathogenic variants, Likely Pathogenic variants, and Variants of Uncertain Significance in genes thought to be associated with the clinical phenotype will always be reported.
- We recommend the patient stay in touch with their Healthcare Provider(s) to discuss any updated information regarding results and our interpretation(s). An ordering Healthcare Provider can request a re-interpretation from us by contacting our laboratory.
- Upon request, PreventionGenetics will provide raw data for sequencing tests to authorized recipients. The data will be provided once testing is complete and final reports have been released. PreventionGenetics does not supply software for data review and interpretation.

Incidental Findings

- Testing could reveal information unrelated to the patient's clinical features. If we learn of information which could be medically actionable, we will relay this information to the Healthcare Provider(s) for discussion.
- If we learn family relationships are not as expected (for example, due to possible specimen mix-up or possible non-paternity), this information will be relayed to the Healthcare Provider(s) for discussion, but will not be included in the patient's report.

Who Has Access to Test Results?

- The patient tested or his/her Authorized Representative (Preventions Genetics requires a signed patient authorization form which is available upon request).
- Any person specifically authorized in writing by the patient tested or his/her Authorized Representative.
- A researcher for medical research or public health purposes if the research is done under federal or state law governing clinical and biological research, or if the identity of the individual is not disclosed.
- The ordering Healthcare Provider or an Authorized Agent or employee of the Healthcare Provider, if they are authorized to obtain the test results, provide patient care, treatment, or counseling, and need to know the information to perform or improve the patient care, treatment, or counseling.
- The hospital or Healthcare Provider for purposes of quality assurance.
- Federal, state, or county health agencies, as they may be authorized.

Confidentiality

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

Risks

- Learning about test results can be stressful and upsetting for the patient and their family.
- 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. We recommend patients discuss specific concerns with their Healthcare Provider.
- As genetic knowledge and understanding changes over time, it is possible a patient's result may be reclassified. This could lead to changes in medical management recommendations or care of family members.

Right to Genetic Counseling

The patient has the right to genetic counseling prior to having testing and again when results have been issued.

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:
PreventionGenetics.com

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.

PRENATAL TEST REQUISITION

ORDERING CHECKLIST
- ☐ Fetal specimen
- ☐ Biological parent specimen(s)
- ☐ Positive control samples (required for targeted tests)
- ☐ Prenatal Healthcare Provider Statement (included)
- ☐ Clinical Feature Checklist / Clinical Records

INSTRUCTIONS
- All testing must be ordered by a qualified Healthcare Provider.
- See Prenatal Guidelines for further ordering details

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 _____ MM/DD/YYYY

FETAL SEX
☐ Male ☐ Female ☐ Unknown ☐ Ambiguous
Based On: _____

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

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

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

☐ **SPECIMEN COLLECTED IN NEW YORK STATE** Inclued 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.

CLINICAL FEATURES
☐ Unaffected ☐ Unknown
☐ Affected, features _____

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

GEOANCESTRY / ETHNICITY

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

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

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 (Targeted Prenatal Testing Only, if needed)

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

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

DATE COLLECTED (MM/DD/YYYY)*

PATIENT ID CODE

☐ **SPECIMEN COLLECTED IN NEW YORK STATE** Inclued 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.

CLINICAL FEATURES
☐ Unaffected ☐ Unknown ☐ Affected, features _____

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

GEOANCESTRY / ETHNICITY

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

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

ADDITIONAL FAMILY MEMBER INFORMATION (Targeted Prenatal Testing Only, if needed)

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

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

DATE COLLECTED (MM/DD/YYYY)*

PATIENT ID CODE

CLINICAL FEATURES
☐ Unaffected ☐ Unknown
☐ Affected, features _____

RELATIONSHIP TO FETUS

GEOANCESTRY / ETHNICITY

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

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

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

BIOLOGICAL SEX
☐ Male ☐ Female
☐ Other _____ SPECIFY KARYOTYPE

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

PREVENTION GENETICS part of EXACT SCIENCES

Test information is available on our website:
PreventionGenetics.com

PREVENTIONGENETICS USE ONLY

PATIENT

LAST NAME

FIRST NAME | MI

CLINICAL INFORMATION (REQUIRED)

CLINICAL INDICATION

- ☐ Abnormal fetal ultrasound (specify, attach report if available)
- ☐ Family history: known familial variant testing (provide relationship, copy of variant report, clinical details).
- ☐ Advanced maternalage (CMA only)
- ☐ Fetal loss / stillbirth / POC
- ☐ Abnormal cell-free fetal DNA test (specify)_____
- ☐ Abnormal amniotic fluid AFP (specify level) _____
- ☐ Other, specify, please call to discuss prior to submission

_____

ADDITIONAL CLINICAL INFORMATION (REQUIRED)

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.

TEST SELECTION

FETAL

Please include any special instructions in the comments section. The tests will be performed concurrently unless otherwise specified.
If targeted testing, please include details. For other tests, the Test Numbers and Names can be obtained from our web site. We require at least one parental specimen be sent for prenatal testing. SEE PRENATAL GUIDELINES FOR MORE INFORMATION.

SELECT TEST OF INTEREST OR FILL IN DESIRED TEST CODE

FOR PRENATAL EXOME TESTING COMPLETE PGXOME PRENATAL DIAGNOSTIC REQUISITION (CLICK HERE FOR FORM).

	TEST CODE	TEST NAME	GENE(S)	VARIANT(S)
☐	990	**Targeted Prenatal Testing for Known Familial Variants** Includes STAT turnaround time (~2 weeks); positive control(s) required.		
☐	3780	**Prenatal Rapid CMA with Cell Culture***		
☐	995	**Fetal Cell Culture*** (only available for testing performed at PreventionGenetics)		
☐			**ORDER OPTIONS** ☐ Patient Only ☐ Family - Duo ☐ Family - Trio ☐ Include family/comparator demographics (name, DOB, ID#, and relationship) on the proband report.	
☐			☐ Patient Only ☐ Family - Duo ☐ Family - Trio ☐ Include family/comparator demographics (name, DOB, ID#, and relationship) on the proband report.	

ADDITIONAL ORDER INFORMATION

SPECIAL INSTRUCTIONS

☐ **ADD EXOME-WIDE CNV ANALYSIS**
$250, CPT CODE 81479
With an order for any PGxome-based or custom panel, exome-wide CNV analysis is available as an add on. To confirm if this is an option for your order, visit the panel-specific description on our website. Unavailable for PG-Select panels, Sanger sequencing, and other test methods. To learn more, visit our website.

STAT TESTING
Prenatal testing is always 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% reduction in price.
☐ **No STAT, test at a standard TAT**

TESTING WILL BE RUN CONCURRENTLY UNLESS OTHERWISE NOTED
☐ Sequential testing, specify order above.
☐ **POSITIVE CONTROL ONLY**
No charge / No report

MATERNAL

Targeted Prenatal Testing (Test Code 990), positive controls from parents and/or proband are required. Maternal Cell Contamination (MCC) Studies (Test Code 800, CPT Code 81265) are strongly recommended for any fetal testing and offered at no additional charge.

	TEST	GENE(S)	VARIANT(S)	REPORT WANTED?
☐	**Positive Control for Variant(s)** Test Code 100, 200, or 300 - no charge			If fetal targeted sequencing, a targeted report for positive controls can be issued upon request. ☐ **YES** ☐ **NO**
☐	**Maternal Cell Contamination (MCC) Study** Test Code 800 - no charge			

PATERNAL

For Targeted Prenatal Testing (Test Code 990), positive controls from parents and/or proband are required.

	TEST	GENE(S)	VARIANT(S)	REPORT WANTED?
☐	**Positive Control for Variant(s)** Test Code 100, 200, or 300 - no charge			If fetal targeted sequencing, a targeted report for positive controls can be issued upon request. ☐ **YES** ☐ **NO**

ADDITIONAL FAMILY MEMBER

For Targeted Prenatal Testing (Test Code 990), positive controls from parents and/or proband are required.

	TEST	GENE(S)	VARIANT(S)	REPORT WANTED?
☐	**Positive Control for Variant(s)** Test Code 100, 200, or 300 - no charge			If fetal targeted sequencing, a targeted report for positive controls can be issued upon request. ☐ **YES** ☐ **NO**

* Cannot be cancelled once a specimen is received.

PREVENTION GENETICS part of EXACT SCIENCES

Test information is available on our website:
PreventionGenetics.com

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:
PreventionGenetics.com

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: Prenatal CMA and cell cultures cannot be canceled once a sample is received. Testing placed on hold will extend overall TAT.

PREVENTION GENETICS part of EXACT SCIENCES

Test information is available on our website:
PreventionGenetics.com

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 ☐ YES, PG ID# _____	☐ Male ☐ Female ☐ Other 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

PREVENTIONGENETICS USE ONLY

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.