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.

STANDARD TEST REQUISITION

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)

ADDRESS	CITY	STATE/PROVIDENCE	ZIP / POSTAL CODE

EMAIL	PHONE NUMBER	GEOANCESTRY / ETHNICITY

MEDICAL RECORD NUMBER (MRN)

BIOLOGICAL SEX
☐ Male ☐ Female ☐ Other, specify karyotype _____

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

ONGOING PREGNANCY
☐ NO ☐ YES
For testing on a prenatal specimen from an ongoing pregnancy complete the Prenatal Test Requisition Form.

HAS PATIENT BEEN TESTED PREVIOUSLY AT PreventionGenetics?
☐ No
☐ Yes, PG ID# _____

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

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

HAS PATIENT'S RELATIVE BEEN TESTED? ☐ NO ☐ YES - at PreventionGenetics, include:

RELATIVE'S NAME AND/OR PreventionGenetics ID NUMBER _____ DATE OF BIRTH (MM/DD/YYYY) _____ RELATIONSHIP TO PATIENT _____

ICD-10 CODES
(REQUIRED FOR INSURANCE BILLING) 1 PRIMARY _____ 2 _____ 3 _____

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.

SPECIMEN INFORMATION

SPECIMEN SOURCE
☐ Whole Blood ☐ Direct CVS ☐ Tissue, Source _____
☐ Saliva ☐ Direct Amniotic Fluid ☐ Extracted DNA, Source _____
☐ Buccal ☐ Cultured Cells, Source _____ ☐ Other _____

SPECIMEN COLLECTION DATE (MM/DD/YYYY)
If no collection date is provided, date of receipt will be used.

☐ **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.

TEST SELECTION

Test Codes, Test Names and Turnaround Times (TAT) are available at PreventionGenetics.com. Include any special test instructions in the comments section.
The tests will be performed concurrently (in tandem). Include any special test instructions in the comments section.

TEST CODE	TEST NAME	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.

COMMENTS

ADDITIONAL COMPARATORS Complete for Comparator

Comparator clinical information is required for accurate interpretation. For full analysis of the comparator data for an additional charge, please submit separate orders.
FAMILY - DUO, TRIO, ETC, CHECKLIST: ☐ Patient's Form ☐ Include family/comparator demographics (name, DOB, ID#, and relationship) on proband report.

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

**If YES, must include clinical info.

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 the Test Methods page on our website under Resources.

☐ **STAT TESTING**
STAT surcharge adds 25% to price. STAT surcharge will not apply if report is delivered after 16 days and blood sample was submitted.

☐ **HOLD TESTING - PENDING**
☐ Funding approval (e.g. MOH)
☐ Other:

☐ **REFLEXIVE TESTING**
Checking this box indicates you would like all ordered testing to be performed in the order listed.

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

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

PHONE NUMBER | **NPI#** (US only) | **PHONE NUMBER** | **NPI#** (US only)

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 3 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

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)**

TESTING WILL PROCEED UNLESS:
- We (or you) are working on a required Pre-Authorization.
- No insurance coverage is available. We will work with you or your patient to determine payment options.

OR PLEASE PROVIDE YOUR PREFERENCES BELOW:

☐ **HOLD TESTING** for benefit investigation / pre-authorization and share results with patient directly via email provided.

☐ **PROCEED WITH TESTING:** patient accepts financial responsibility for test; regardless of insurance coverage.
(All tests with an in-network insurance are held for benefits investigation, regardless of selected option, except for prenatal and Rapid NICU tests.)

☐ **OTHER:** _____

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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RESET FORM

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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.