PRAXIS GENOMICS

All testing requests must be submitted by a qualified Healthcare Provider

THIS FORM MUST ACCOMPANY ALL SAMPLES

LAB USE ONLY

6115 Peachtree Dunwoody Rd, Suite 220 | Atlanta, GA 30328 | Phone: 678-837-4022 | Testing information available at www.praxisgenomics.com

SOMATIC TEST REQUEST FORM

Patient Information

Last (Family) Name	First Name	MI	Date of Birth
			_____ /_____ /_____ MONTH DAY YEAR

Patient ID / Medical Record Number	Shipping Condition	Specimen Collection Dates	Sex
	☐ Room Temp ☐ Frozen	Normal _____ /_____ /_____ MONTH DAY YEAR Malignant _____ /_____ /_____ MONTH DAY YEAR	☐ Male ☐ Female

Has Patient been previously tested at this facility?

☐ Yes, Praxis ID: _____

Have any Relatives been previously tested at this facility?

Praxis ID: _____ Relation to patient: _____

Date of Birth _____ /_____ /_____ MONTH DAY YEAR

Praxis ID: _____ Relation to patient: _____

Date of Birth _____ /_____ /_____ MONTH DAY YEAR

Specimen Source/s

- ☐ Whole Blood
- ☐ Cultured Cells, Type: _____
- ☐ Tissue, Type: _____
- ☐ Saliva
- ☐ Buccal Swab
- ☐ Sorted Cells, Type: _____
- ☐ Extracted DNA
- ☐ FFPE Block, Quantity: _____
- ☐ FFPE Slide, Quantity: _____
- ☐ Bone Marrow Aspirate

See website for specimen requirements by test

Collection Notes

ICD-10 Diagnosis Codes

1:_____ 2:_____
3:_____ 4:_____
5:_____ 6:_____
7:_____ 8:_____

Blood Transfusion / Marrow Transplant

☐ No ☐ Yes, Specify

_____ /_____ /_____ MONTH DAY YEAR

Type: _____

Test Selection

Whole Genome Sequencing (WGS)

☐ (SRG601) Somatic Genome One Normal and One Malignant Specimen
☐ (SRG602) Somatic Genome Add-On, Additional Specimen
Quantity: _____

Combined WGS & OGM

☐ (PRX601) Somatic OGM+WGS, One Normal and One Malignant Specimen
☐ (PRX602) Somatic OGM+WGS Add-On, Additional Specimen
Quantity: _____

Optical Genome Mapping (OGM)

☐ (OGM601) Somatic OGM, One Normal and One Malignant Specimen
☐ (OGM602) Somatic OGM Add-On, Additional Specimen
Quantity: _____

Transcriptome Sequencing

☐ (SRT701) Somatic Transcriptome, One Normal and One Malignant Specimen
☐ (SRT702) Somatic Transcriptome Add-On, Additional Specimen
Quantity: _____

STAT Testing: ☐ **2W**
Expedited Turnaround time *2 Week TAT*

Submitting Provider Information

Institution		Invoice ID (if known)	
Address	City	State	ZIP
Requesting Provider (First, Last, Degree)		Additional Provider (First, Last, Degree)	
Primary Email Address for Report Delivery		Secondary Email Address for Report Delivery	
Phone Number	NPI#	Additional Emails for Report Delivery	
Email Address for Invoice Delivery			

Billing Information Notice

Payment for testing is handled via invoice system. The invoice is generated following consultation about the appropriate test to be ordered. Please include the unique Invoice ID in the **Submitting Provider Information** section above. Samples sent without prior consultation must include an address for invoice delivery. The invoice must be paid in full before samples are processed by Praxis Genomics. Samples received with incorrect or missing invoice or provider information will be delayed or possibly rejected depending on sample collection date.

For payment options visit our website at: www.praxisgenomics.com/billing or contact us at 678-837-4022 or support@praxisgenomics.com.

Clinical Information - Please fill out separate sheet for each individual

Behavioral/Psychiatric
- ☐ Anxiety
- ☐ Attention Deficit Hyperactivity Disorder
- ☐ Autism
- ☐ Behavioral/Psychiatric Abnormality
- ☐ Dementia
- ☐ Pervasive Developmental Delay

Cardiac
- ☐ Aortic Atresia
- ☐ ASD
- ☐ Atrial Septal Defect
- ☐ AV Canal Defect
- ☐ Coarctation of the Aorta
- ☐ Congestive Heart Failure
- ☐ Dextrocardia and Situs Inversus
- ☐ Double Outlet Right Ventricle
- ☐ Ebstein's Anomaly
- ☐ Echogenic Intracardiac Focus
- ☐ Hypoplastic Left Heart
- ☐ Pulmonary Valve Atresia
- ☐ Tetralogy of Fallot
- ☐ Transposition of the Great Vessels
- ☐ Truncus Arteriosus
- ☐ Ventricular Septal Defect

Cognitive/Developmental
- ☐ Developmental Delay
- ☐ Fine Motor Delay
- ☐ Gross Motor Delay
- ☐ Intellectual Disability/MR
- ☐ Learning Disability
- ☐ Speech Delay

Craniofacial
- ☐ Cleft Lip +/- Cleft Palate
- ☐ Dysmorphic Facial Features
- ☐ Ear Malformation
- ☐ Hyper/Hypotelorism
- ☐ Macrocephaly
- ☐ Microcephaly

Cutaneous
- ☐ Acne
- ☐ Alopecia
- ☐ Axillary/Inguinal Freckling
- ☐ Blisters
- ☐ Café-Au-Lait Spots
- ☐ Congenital Nevus
- ☐ Ectodermal Dysplasia
- ☐ Ichthyosis
- ☐ Loose Skin
- ☐ Papules
- ☐ Psoriasis
- ☐ Striae
- ☐ Urticaria

Endocrine/Exocrine
- ☐ Adrenal
- ☐ Bone
- ☐ Diabetes Mellitus
- ☐ Hypothalamus
- ☐ Pancreas
- ☐ Parathyroid
- ☐ Pituitary
- ☐ Reproductive
- ☐ Thyroid

Gastrointestinal
- ☐ Absent Stomach
- ☐ Echogenic Focus
- ☐ Gastrointestinal Pseudo Obstruction
- ☐ Gastroschisis
- ☐ Meconium Ileus / Anal Atresia
- ☐ Omphalocele
- ☐ Pyloric Stenosis
- ☐ Tracheoesophageal Fistula

Genitourinary
- ☐ Ambiguous Genitalia
- ☐ Cryptorchidism
- ☐ Hydronephrosis
- ☐ Hypoparathyroidism
- ☐ Hypospadias
- ☐ Hypothyroidism
- ☐ Kidney Malformation
- ☐ Megacystis (incl. posterior valves)
- ☐ Myoglobinuria
- ☐ Polycystic Kidneys
- ☐ Renal Agenesis
- ☐ Urethra/Ureter Obstruction

Growth
- ☐ Failure to Thrive
- ☐ Overgrowth
- ☐ Short Stature

Hearing/Vision
- ☐ Abnormality of Eye Movement
- ☐ Abnormality of Vision
- ☐ Hearing Loss
- ☐ Migraine Headache
- ☐ Optic Atrophy
- ☐ Ptosis
- ☐ Retinopathy

Hematology
- ☐ Anemia
- ☐ Clotting Disorder
- ☐ Hemophilia
- ☐ Neutropenia
- ☐ Thrombocytopenia

Immune System
- ☐ Autoimmune Disorders
- ☐ Immunodeficiency

Metabolic/Laboratory Abnormalities
- ☐ Biliary Tract
- ☐ Blood Gases
- ☐ Electrolytes
- ☐ Gastrointestinal
- ☐ Glycoproteins
- ☐ Kidney
- ☐ Lipids
- ☐ Liver
- ☐ Mitochondrial

Musculoskeletal
- ☐ Acromelia
- ☐ Clenched Hands
- ☐ Club Foot (bilateral)
- ☐ Contractures (arthrogryposis)
- ☐ Cramps After Exercise
- ☐ Diaphragmatic Hernia
- ☐ Exercise Intolerance
- ☐ Joint Laxity
- ☐ Limb Anomaly
- ☐ Mesomelia
- ☐ Micromelia
- ☐ Myoclonus
- ☐ Ophthalmoplegia
- ☐ Polydactyly
- ☐ Proximal Limb Weakness
- ☐ Scoliosis
- ☐ Skeletal Dysplasia
- ☐ Syndactyly
- ☐ Vertebral Anomaly

Neoplasia
- ☐ Bone and Joints
- ☐ Breast
- ☐ Cancer Syndromes
- ☐ CNS
- ☐ Endocrine
- ☐ GI
- ☐ GU
- ☐ Hamartoma
- ☐ Head and Neck
- ☐ Hematopoietic / Lymphatic
- ☐ Intrathoracic Malignancies
- ☐ Neurofibroma
- ☐ Optic Gliomas
- ☐ Skin
- ☐ Soft Tissue

Neurological
- ☐ Abnormal Gyri (Lissencephaly)
- ☐ Agenesis of the Corpus Callosum
- ☐ Asthenia
- ☐ Cerebellar Hypoplasia
- ☐ Cerebellar Signs
- ☐ Cerebral Blindness
- ☐ Cerebral Palsy
- ☐ Dandy Walker (posterior fossa abnormality)
- ☐ Encephalopathy
- ☐ Headache
- ☐ Holoproscencephaly
- ☐ Hypertonia
- ☐ Hypotonia
- ☐ Macrocephaly
- ☐ Microcephaly
- ☐ Neural Tube Defect
- ☐ Neuropathy
- ☐ Seizures
- ☐ Stroke
- ☐ Structural Brain Anomaly
- ☐ Ventriculomegaly/Hydrocephaly

Prenatal/Perinatal History
- ☐ 2 Vessel Cord
- ☐ Decreased Fetal Movement
- ☐ Echogenic Focus
- ☐ Floppy Baby
- ☐ Increased Nuchal Translucency
- ☐ IUGR
- ☐ Non-Immune Hydrops Fetalis
- ☐ Oligohydraminos
- ☐ Perinatal Insult
- ☐ Polyhydramnios
- ☐ Prematurity

Pulmonary
- ☐ CCAM/Small Thoracic Cavity
- ☐ Diaphragmatic Hernia
- ☐ Eventration of Diaphragm
- ☐ Pleural Effusion
- ☐ Pulmonary Sequestration
- ☐ Respiratory Insufficiency

Pedigrees may be attached or illustrated on the back of this page. Detail any findings of chromosomal abnormalities in the comments below. Prior pregnancy risk, multiple gestation evidence and maternal age/weight must be disclosed below if applicable.

Diagnosis
- ☐ Colorectal Carcinoma
- ☐ Breast
- ☐ Prostate
- ☐ Melanoma
- ☐ Ovarian
- ☐ NSCLC
- ☐ Other: _____

Disease Status
- ☐ Metastatic
- ☐ Refractory
- ☐ None
- ☐ Recurrent
- ☐ Relapse
- ☐ Progression

Family History / Ethnicity / IVF Details

Prior Genetic Testing

Electrophysiology / Imaging (describe findings)

Labs (describe findings)

Attachments
- ☐ Copy of recent pathology/cytology reports including (if available), CBC/differential, BMA differential, FAB classification.
- ☐ Test results from all other Molecular Diagnostic Assays by FISH, IHC, or other genetic assays, e.g., ER, PR, HER2, EGFR, KRAS, etc.

Please attach any additional sheets or information you think is pertinent to the patient's condition.

Diagnostic Testing Informed Consent Overview

Notice to Health Care Practitioner

This document is a consent form for clinical Whole Genome Sequencing (WGS), external whole genome data analysis (EWGDA), Transcriptome (RNAseq) and/or Optical Genome Mapping (OGM) tests. The laboratory will only accept such test requests after the patient/parent or legal guardian/next of kin has received genetic counseling from a healthcare provider with experience in counseling patients for such a test. Please be aware of any applicable state laws in regards to counseling needs related to the current condition, the possibilities of detecting unsuspected conditions, incidental findings, as well as other issues related to health insurance, and possible effects on life insurance. Please explain this consent to the patient, or authorized representative/guardian, and obtain an informed consent. Alternatively, contact Praxis Genomics for consenting services available by appointment.

Whole Genome Sequencing (WGS), External Whole Genome Data Analysis (EWGDA), Transcriptome (RNAseq) Optical Genome Mapping (OGM) Informed Consent

Because OGM, EWGDA, RNAseq and WGS results have potential consequences for the patient's family, we recommend that the consenting and ordering process be performed with the assistance of a genetic counselor and/or the ordering physician.

About the tests:
Praxis Genomics offers the most comprehensive diagnostic testing currently available anywhere in the world. We use OGM and/or WGS data to analyze the customers' genetic composition at a previously unavailable resolution. The two methods complement each other and detect different types of variants. RNAseq is used to evaluate the functional consequence of variants in coding regions. OGM focuses on detecting structural variants in the genome, such as deletions, insertions, inversions, and translocations that may have pathogenic associations. WGS is primarily used for detection of small variants, such as single nucleotide variants (SNVs) or small insertions and deletions (indels) less than 50bp long. WGS can also detect repeat expansions associated with several neurological conditions and is useful for confirmation of structural variants detected by OGM. Combination of the two methods will significantly increase diagnostic sensitivity and specificity and is the best available test for those who failed to obtain a genetic diagnosis with prior microarray, panel, exome, genome or cytogenetic studies.

Sample requirements:
OGM, WGS and RNAseq can be performed on multiple sample types. For detail, see description of individual tests on the website. For EWGDA arrangements have to be made with the laboratory that has previously performed WGS to have the data securely transferred.

Who should get tested?
All patients who have a constitutional (inherited) or somatic genetic condition (malignancy) but have obtained no genetic diagnosis for this condition after prior microarray, panel, exome, genome or cytogenetic testing. To help rule out rare family specific variants as cause of disease, the parents should be tested along with the patient if possible; additional family members such as healthy siblings, or similarly affected distant relatives can help to rule out or prove the pathogenic role of variants detected.

What kind of results are reported?

1. In a positive report we will list variants that are known pathogenic or are deemed likely pathogenic based on well-established scientific evidence (ACMG) and cause a severe medical condition in the individual tested.
2. In an indeterminate report, we will list variants that we cannot exclude as the cause the patient's condition based on the current state of medical knowledge
3. In a negative report we will state the absence of variants that could be associated with the patient's condition.

What implications do the results have?
When a known or predicted disease causing variant is detected, the test result is highly accurate. A positive result will help the clinician to treat/manage the patient's condition better and prepare for or potentially prevent further manifestations of the medical condition. An indeterminate result can guide the physician in further testing to confirm or rule out the pathogenic role of the reported variant(s). A negative result does not guarantee the absence of a genetic cause for the patient's condition, since it is based on current medical and scientific knowledge which is incomplete.

Are there results that will not be reported?

1. Variants in genes that affect susceptibility to a condition, but do not definitively cause it, will not be reported.
2. There might be disease causing variants identified in the patient that are unrelated to the patient's condition for which testing was ordered. These are referred to as "Secondary Findings" and indicate the presence of previously undiagnosed, potentially serious conditions that can be prevented or treated if diagnosed. A list of such conditions based on the recommendation of the American College of Medical Genetics (ACMG) is provided on the ACMG website. Such findings are not automatically reported. Please initial below whether or not you want secondary findings reported. ***Leaving these lines blank is interpreted as refusing secondary findings.***

_____ **Yes, I want secondary findings reported.**
Initial

_____ **No, I do not want secondary findings reported.**
Initial

3. Results from the patient's relatives tested will not be reported, unless they are directly relevant to the patient's condition. However, the patient's genetic results may have implications for their relatives, and it is important that these implications are discussed with a genetic counselor.
4. Carrier status for recessive disorders that do not overlap with the patient's condition will not be reported. Most people carry variants that are not disease causing unless they are inherited from both parents. Having such variants is referred to as being a "carrier" for a condition. If you are concerned about carrier status for conditions that might run in your family, a carrier test should be ordered
5. Non-paternity (when the reported father of the child is not the biological father) or half sibships (when siblings do not share the same father AND mother) would be detected. We do not report these findings unless they have direct clinical significance.

Are there limitations to WGS, RNAseq, EWGDA and OGM Testing?
There is a possibility that genetic variants that cause a condition are not identified by testing either because of the technical limitations of the assay or because of incomplete understanding of the significance of variants detected. EWGDA is sensitive to the quality of the data received. RNAseq is sensitive to the sample source's gene expression levels.

Who will have access to the results?
Test results are maintained electronically by the laboratory. The results are provided to the ordering physician and/or health care facility that ordered the test. Results may also be made available to individuals/organizations with a legal right of access under applicable Federal and/or State law, or as authorized by the patient or the patient's representative. Patient privacy is of utmost concern to us, and we adhere to HIPAA privacy and security requirements.

How long are testing results kept in the testing lab?
The laboratory may keep the raw data obtained during testing in the lab indefinitely. This helps us improve our diagnostic capabilities. To advance the understanding of genetic disorders, your results might be analyzed and published in scientific articles in a de-identified manner consistent with HIPAA guidelines.

What will happen to my DNA/RNA sample?
No additional tests will be performed on these samples without specific signed authorization by the individual(s) tested. However, the samples will be kept indefinitely for possible additional testing unless you instruct us to destroy the samples. In this case the samples will be destroyed within 60 days from the test order date.

Circle Yes/No. Leaving these lines blank is interpreted as agreement.

Y / N _____ Initial — **I agree that my sample may be kept beyond 60 days for future testing or research participation. I understand that my sample may be used for quality control purposes. I understand that there is no guarantee of availability for my sample beyond 60 days.**

Y / N _____ Initial — **I agree that my data may be retained indefinitely to help with improving Praxis Genomics' diagnostic capabilities.**

Y / N _____ Initial — **I agree that my diagnostic testing data may be used for scientific contributions in a de-identified manner.**

Diagnostic Testing Informed Consent

What are the risks of testing?

1. Testing may identify carrier status for serious and/or untreatable genetic conditions. It can result in unexpected psychological trauma, both for you and your family. The detection of such a condition or conditions could also affect the health or health care needs of your siblings, children, or other close relatives.

2. Although testing is highly accurate, the interpretation of the report is based on current medical knowledge which is incomplete. This may result in failure to correctly diagnose or predict the manifestation or severity of certain conditions that are not yet described or not well characterized. We do not automatically report out changes in interpretation of variants due to scientific advances relating to the variant detected, but we do have mechanisms to issue an updated report if requested by the patient's physician.

3. Genetic non-discrimination law prevents insurance companies from using your genetic information to deny health insurance coverage, but the law does not cover life insurance, disability insurance or long- term care insurance. Please be aware of any applicable State laws and applicable terms of any active insurance policies.

Consent for Diagnostic Testing

All of the above has been explained to me to my satisfaction. My signature below confirms this. I understand this testing is voluntary, and I have had the opportunity to ask questions about alternative testing. I authorize my Health Care Provider to have access to my results.

Individual Signature _____ Individual Printed Name _____

Date of Birth _____ Signature Date _____

I have provided genetic counseling and have explained the risks, benefits and limitations of Diagnostic Testing to the individual.

Provider Signature _____ Provider Printed Name _____

NPI# _____ Signature Date _____

Informed Consent of Family Member/s

I understand that my sample is being submitted in the efforts to evaluate or diagnose the proband, and that any results obtained from my sample will be used for this purpose alone. I will not be informed of any test results for my sample. If I request any results, I must be tested separately.

Family Member 1	Family Member 2	Family Member 3
Signature	Signature	Signature
Print Name	Print Name	Print Name
Relationship to Proband	Relationship to Proband	Relationship to Proband
Signature Date	Signature Date	Signature Date

Circle Yes/No

Y / N	Initial 1	**Y / N**	Initial 2	**Y / N**	Initial 3
Y / N	Initial 1	**Y / N**	Initial 2	**Y / N**	Initial 3
Y / N	Initial 1	**Y / N**	Initial 2	**Y / N**	Initial 3

Leaving these lines blank is interpreted as agreement.

We agree that our data may be retained indefinitely to help with improving Praxis Genomics' diagnostic capabilities.

We agree that our data may be used for scientific contributions in a de-identified manner.

We agree that our sample may be kept beyond 60 days. We understand no additional tests will be performed without our specific consent, but our sample may be used for quality control purposes. We understand there is no guarantee of availability for our sample beyond 60 days.