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

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

WHOLE GENOME SEQUENCING
PGnome® TEST REQUISITION

ORDERING CHECKLIST
- ☐ Patient and comparator (if provided) specimens
- ☐ Healthcare Provider Statement (required for specimens collected in NY)
- ☐ Clinical Features Checklist (last 5 pages of this pdf) and/or relevant medical records and family health history (i.e. clinic notes, prior genetic testing, pedigree) **(required for Diagnostic and Rapid PGnome)**

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_____

SPECIMEN INFORMATION

SPECIMEN SOURCE
☐ Whole Blood
☐ Extracted DNA, Whole Blood

SPECIMEN COLLECTION DATE
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.

PATIENT TEST SELECTION

STANDARD DIAGNOSTIC PGnome

☐ **PATIENT ONLY** Test Code 7000

FAMILY
☐ **DUO** Test Code 8000
☐ **TRIO** Test Code 8001
☐ **OTHER** *Specify* _____
Complete a Comparator Test Requisition form for each family member.
Clinical information is REQUIRED for each comparator for accurate interpretation.
☐ Include family/comparator demographics (name, DOB, ID#, and relationship) on the proband report.

SECONDARY (ADDITIONAL) FINDINGS
Details can be found in the PGnome Healthcare Provider Statement. Options for reporting of Secondary Findings are to be marked below.
☐ OPT IN: GUIDELINE RECOMMENDED GENES

TRIO (WITH PARENTS) ONLY
☐ OPT IN: PG DISCOVERY

OPTIONAL SECONDARY FINDINGS ADD-ON
Details can be found in the PGxome Healthcare Provider Statement. Marking one or both of the options below will add an additional $590 USD to your order.
☐ OPT IN: OTHER PREDISPOSITIONS / DIAGNOSES
☐ OPT IN: CARRIER STATUS

RAPID DIAGNOSTIC PGnome

☐ **PATIENT ONLY** Test Code 14000
☐ **FAMILY - DUO, TRIO, ETC** Codes Duo-14001/Trio-14002
Complete a Comparator Test Requisition form for each family member.
Clinical information is REQUIRED for each comparator for accurate interpretation.

NO SECONDARY FINDINGS AVAILABLE FOR RAPID PGNOME
☐ Include family/comparator demographics (name, DOB, ID#, and relationship) on the proband report.

HEALTH SCREEN PGnome

☐ **PATIENT ONLY** Test Code 9000
Includes carrier status.

SECONDARY (ADDITIONAL) FINDINGS
Details can be found in the PGnome Healthcare Provider Statement. Options for reporting of Secondary Findings are to be marked below.
☐ OPT IN: GUIDELINE RECOMMENDED GENES
☐ OPT IN: OTHER PREDISPOSITIONS / DIAGNOSES

COMMENTS OR SPECIAL INSTRUCTIONS:

☐ **RE-ANALYSIS**
Re-analysis will be completed with original secondary finding selections unless otherwise specified. Changes to secondary finding opt ins or structure of the analysis (additional comparators) may result in additional charges. See our website for full re-analysis policy.

Test Code _____

Desired secondary findings (choose opt ins at left)

ADDITIONAL COMPARATORS Complete for PGxome Family Duo or Trio orders

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

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

*If YES, must include clinical info.

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 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:
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PREVENTIONGENETICS USE ONLY

PATIENT

LAST NAME

FIRST NAME | MI

COMPLETE THIS FORM FOR PATIENT PAY AND/OR INSURANCE BILLING

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

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

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

ACCEPTANCE of financial responsibility for genetic testing

SIGNATURE REQUIRED BELOW TO PROCEED WITH TESTING.

MY SIGNATURE INDICATES I ACCEPT FINANCIAL RESPONSIBILITY FOR ALL FEES ASSOCIATED WITH THIS GENETIC TESTING ORDER.
If applicable, I authorize PreventionGenetics to release information received including, without limitation, medical information, which includes laboratory test results, such as genetic tests results, to my health plan / insurance carrier and its Authorized Representatives. I further authorize insurance payments directly to PreventionGenetics for the services rendered. I understand my Health Plan / Insurance / Medicare / Medicaid carrier may not approve and reimburse my medical genetic services in full due to usual and customary rate limits, benefit exclusions, coverage limits, lack of authorization, medical necessity or otherwise. **I understand I am financially responsible for fees not paid in full by my insurer,** co-payments, and policy deductibles except where my liability is limited by contract or State and Federal law. I agree to help PreventionGenetics resolve any insurance claim issues. I understand my out-of-network benefits may apply. PreventionGenetics may contact me to resolve any billing-related issues and to request payment.

SIGN HERE: Required to process form

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

CREDIT CARD PAYMENT

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

CREDIT CARD INFORMATION

CREDIT CARD NUMBER (VISA, DISCOVER, OR MASTERCARD ONLY)	EXPIRATION DATE	3-DIGIT SECURITY CODE

My signature authorizes PreventionGenetics to charge my credit card for services for which I am responsible.

SIGN HERE: Required to process credit card

CREDIT CARD HOLDERS SIGNATURE | DATE

INSURANCE INFORMATION - IF APPLICABLE

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

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

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

PRIMARY INSURANCE COMPANY NAME (REQUIRED)	PHONE NUMBER

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

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

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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PREVENTIONGENETICS USE ONLY

All testing must be ordered by a qualified Healthcare Provider

THIS FORM MUST ACCOMPANY ALL SPECIMENS

PGnome® - Whole Genome Sequencing

HEALTHCARE PROVIDER STATEMENT

This statement is required for patient specimens collected in NY and recommended for others, and applies to Whole Genome Sequencing.

PATIENT INFORMATION

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

FAMILY MEMBERS

If a Family (duo, trio, etc.) is being tested, please provide family member information:

FAMILY MEMBER'S NAME	RELATIONSHIP
FAMILY MEMBER'S NAME	**RELATIONSHIP**
FAMILY MEMBER'S NAME	**RELATIONSHIP**

I have provided informed consent to my patient. My patient has had the opportunity to ask questions. Please indicate patient preferences for secondary findings on the PGnome® Test Requisition Form.

HEALTHCARE PROVIDER SIGNATURE _____ PRINTED NAME _____ DATE _____

Retention of Unused DNA Statement for New York State Specimens

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

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

PATIENT OR LEGAL REPRESENTATIVE SIGNATURE _____ PRINTED NAME _____ DATE _____

The following information should be used as a guide to provide informed consent to the patient and/or patient's family. Testing must be ordered by a qualified Healthcare Provider.

PURPOSE

- **Diagnostic PGnome:** The purpose of this test is to find the underlying genetic cause for the patient's health condition using Whole Genome Sequencing (WGS).
- **Health Screen PGnome**: The purpose of this test is to provide pan-ethnic carrier screening using a Whole Genome Sequencing (WGS) test. Variants in any gene that relate to an autosomal recessive or X-linked recessive disorder (in females) would be reported (regardless of the incidence of the condition). In addition, patients have the option of also receiving genetic variants that predispose to autosomal or X-linked dominant disorders or X-linked recessive disorders (in males).

ABOUT PGNOME TEST

- This test involves the sequencing of thousands of genes at the same time, whereas many other genetic tests look at only one gene or a small group of genes. The way we perform the genome test is through a procedure called Next Generation Sequencing (NGS). We confirm important results with another type of sequencing called Sanger sequencing. Copy number variants (CNVs), also known as deletions/duplications, and are detected from NGS data. Most reported CNVs are confirmed using another technology such as aCGH, MLPA, or PCR.
- We will need about one teaspoon of blood (3-5 ml of whole blood or DNA extracted from blood) from each individual to perform testing. In rare instances, a second specimen may be requested.
- Results of the test will be presented in an individualized, written report transmitted to the patient's Healthcare Provider(s).
- For additional information about this test, see the PGnome test description on the PreventionGenetics website (https://www.preventiongenetics.com/ClinicalTesting/TestCategory/PGnome.php).

FAMILY TESTING
(Diagnostic PGnome Only)

- Testing of family members is very helpful for interpretation of results. When possible, testing of the patient and two other family members (called a trio), preferably biological parents, should be performed. If one or both biological parents are unavailable, sometimes siblings or other close relatives can be tested. Family testing increases the chance of getting a conclusive result.
- It is very important family genetic relationships are correctly stated because issues such as an undisclosed adoption or uncertain paternity can cause confusion. If you are aware of any such issues in the family, they should be discussed confidentially with your Genetic Counselor or Ordering Physician.
- Family member information (i.e. parental genotype information) helps us interpret the patient's results and will be included in the patient's report. All sequence variants reported will include parental status. While large CNVs identified in the proband may include parental inheritance information, confirmation using an additional method will not be performed on parental specimens. If parental status for variants in the patient's report is not desired (for primary and/or secondary findings), please make note of this under "Patient Test Selection".
- If family member(s) tested as part of PGnome Family desire their own PGnome analysis and test report, their healthcare

(continued to page 9)

provider should complete a PGnome Diagnostic or PGnome Health Screen test requisition form. Full PGnome reports for family member(s) incur an additional charge per family member.

REPORT INFORMATION

- **Diagnostic PGnome:**
  - Testing searches for variants in both the nuclear and mitochondrial genome.
    - Genetic variants are defined as the differences between the patient's DNA and the human reference DNA. Generally only results that may explain the patient's clinical features will be reported..
    - Expansions in *C9orf72, ATXN2, PABPN1, PHOX2B* and *FMR1* can be detected by PGnome. These findings will be reported if relevant to the described phenotype or with certain secondary finding selections
    - In genes believed to be associated or possibly associated with the patient's clinical features, all Pathogenic, Likely Pathogenic, and Variants of Uncertain Significance (unknown if they cause disease) will be reported.

- **Health Screen PGnome:**
  - Testing will only include analysis of variants located in the nuclear genome.'
  - Pathogenic and Likely Pathogenic sequence variants (Richards et al. 2015 Genet Med 17:405-424) within genes currently known to be clinically relevant for carrier status in autosomal recessive disorders or X-linked recessive disorders (in females) will be reported. Variants in genes not currently known to be clinically relevant will not be reported.
  - Some individuals may have two Pathogenic or Likely Pathogenic genetic variants (compound heterozygous or homozygous) in a gene that causes an autosomal recessive disorder. Even if the patient may not be obviously affected by the disorder, this finding could lead to a diagnosis. If identified this information will be included in the patient's test report as it also indicates a positive carrier status.
  - The patient will very likely have many recessive Variants of Uncertain Significance. These variants will not be included in the report, but the laboratory will retain this data.

- Other findings (aka "Secondary Findings" - see below) may be reported depending on the patient's preferences. These Secondary Findings may have an important impact on health.
- New research results are continually improving the ability to interpret the WGS results. An ordering Healthcare Provider can request a re-interpretation from us.

ISSUING THE REPORT

- Results will be sent directly to the ordering Healthcare Provider(s) and NOT to the patient.
- Genetic counseling and/or clinical genetics consultation before and after testing is recommended.
- Patients have the right to receive a copy of their test report. They may obtain a copy from their Healthcare Provider(s) or if a signed patient authorization (form available upon request) is received, from PreventionGenetics.

SECONDARY FINDINGS

- In many patients, WGS will reveal one or more additional genetic variants which could be important to the patient's health. These include for example variants predisposing the patient to cancer or heart disease, or variants relevant to reproductive planning. These are termed secondary findings. The patient may or may not wish to be informed of secondary findings.
- Secondary findings are not available for Rapid PGnome.
- Carrier Status is always reported for Health Screen PGnome. The patient will have a choice about what other secondary findings are reported.
- For Standard Diagnostic PGnome the patient and/or patient's family will have a choice on which types of secondary findings are reported.
- Please consider the following carefully. Variants described in these sections will only be reported if the patient OPTS IN.

SECONDARY FINDINGS NO CHARGE OPT INS

- **Guideline Recommended Genes:** The American College of Medical Genetics and Genomics recommends all labs performing WGS report pathogenic variants in specific genes that cause certain, mostly dominantly inherited disorders (Version 3.3, Lee et al. 2025, PubMed ID 40568962). These disorders are treatable and/or preventable. Included on this list are some cancer predisposition conditions, heart conditions associated with sudden death, and conditions that could result in severe health consequences if surgery is performed with certain anesthetics.
- **PG Discovery (candidate genes, available for diagnostic PGnome trios with proband and biological parents only):** WGS provides the opportunity to identify rare variants in candidate genes for which there is limited available evidence. Relevant rare homozygous, hemizygous, compound heterozygous, and/or de novo variants are reported. These genes and variants reported within them will be classified as uncertain significance, and the variants will not be confirmed by a second method. Any literature, such as limited animal studies, etc., is referenced where available. Further research is required to understand if any human disease association exists. PreventionGenetics may reach out to request consent for submission of these variants to research programs and databases like GeneMatcher (https://genematcher.org/).

ADD ON SECONDARY FINDINGS

(additional charge)

- Please consider the following carefully. Variants described in these sections will only be reported if the patient OPTS IN.
  - **Other Predispositions/Diagnoses:** This secondary finding option refers to a very broad range of disorders beyond the Guideline Recommended Genes. Examples vary widely and include adult onset neurological conditions such as Alzheimer's disease, Parkinson disease, amyotrophic lateral sclerosis (ALS), and small vessel disease, as well as cancer predispositions, and renal conditions, among others. Some of these disorders are very serious, leading to death. While treatment or prevention will be effective for some of these disorders but not others, knowledge of these predispositions may be useful for the patient and their family. (Amendola et al. 2015. Genome Res 25(3):305- 315; Dorschner et al. 2013. Am J Hum Genet 93(4):631-640). Some people may want to know about these disorders while others may prefer not to know. If this option is selected, all pathogenic and likely pathogenic variants in genes that are likely to result in a Mendelian (single gene) disorder (i.e., one variant in a dominant gene or X-linked gene or two variants in a recessive gene) will be reported. Individuals will be screened for expansions in ***C9orf72, FMR1, PABPN1, PHOX2B*** and ***ATXN2***. Variants in the mitochondrial genome will not be screened in this analysis.

    Many of these conditions have adult onset, reviewing professional guidelines before discussing these options with minors and their families is recommended. (Borry et al. 2006 Clin Genet 70(5):374-81; Lucassen et al. 2010 British Society for Human Genetics; Fallat et al. 2013 Pediatrics 131(3): 620–2; NSGC Position Statement 2017. For minors, predictive testing should be postponed until they have reached an age capable of true informed consent (ability to understand the risks, benefits, and implications of results). Consideration of testing in minors should ideally include genetic counseling, the parents, and assent of the child.
  - **Carrier Status** (included n/c in Health Screen orders)**:** WGS can also provide panethnic carrier screening. For carrier status, variants in any gene that relate to an autosomal recessive or X-linked recessive disorder (in females) would be reported if this option is selected (regardless of the incidence of the condition). Such single recessive, pathogenic variants usually don't appreciably affect a patient's health, but may be useful in reproductive planning. In accordance with current professional guidelines (Borry et al. 2006. Eur J Hum Genet 14(2):133-8; NSGC Position Statement 2012; Ross et al. 2013 Genet Med 15(3):234-245). For minors, predictive testing should be postponed until they have reached an age capable of true informed consent (ability to understand the risks, benefits, and implications of results). Consideration of testing in minors should ideally include genetic counseling, the parents, and assent of the child.
- Genetic variants related to complex disease, will not be reported at this time.
- Genetic variants in genes not currently known to be clinically relevant will not be reported with this add-on analysis.
- If testing reveals the family relationships are not as expected (for example, non-paternity), this information will be relayed to the healthcare provider(s) for discussion, but will not be included in the patient's report.

DATA

- PreventionGenetics will store the patient's sequence data. This will permit reanalysis and reinterpretation of the data in the future. Upon a physician's request, PreventionGenetics will perform, without additional charge, one reanalysis and reinterpretation of the data within three years of the date on the original test report. Thereafter, reanalysis and reinterpretation may be requested, but a fee will be charged for this service.
- PreventionGenetics recommends DNA sequence information from this test also be stored in the patient's electronic medical record. This will best benefit the patient and family members. PreventionGenetics will provide WGS data upon request. PreventionGenetics does not supply software for data review and interpretation..

RISKS

- Blood draw risks include bruising and bleeding. There is also a small chance the patient may get an infection, have excess bleeding, become dizzy, or faint from the blood draw.
- Learning about test results can be stressful and upsetting.
- The patient and/or patient's family may have concerns about genetic discrimination, including health insurance, life insurance, employment and long-term disability. These should be addressed according to federal and state laws. The Federal Genetic Information Non-discrimination Act (GINA) prohibits the use of genetic information for discrimination in health insurance and employment.

LIMITATIONS

- While WGS captures nearly all regions of the genome, this test primarily reports on most of the coding parts of our genes (called exons). All of the exons together is called the exome. The exome only covers approximately 1.5% of all the genetic material. However, testing the exome covers the vast majority of genetic variants which cause single gene (or Mendelian) disorders.
- Interpretation of the test results is limited by the information currently available. Better interpretation could be possible in the future as more data and knowledge about human genetics are accumulated.
- Testing will detect single base pair changes and Structural Variants (SVs), such as small and large deletions or duplications, but we are generally not able to detect or currently analyze and report other types of genetic changes (e.g. blanced translocations, deep intronic variants, methylation abnormalities, or some repetitive sequence changes).
- This test will not provide detection of certain genes or specific exons of genes due to complicated technicalities (such as sequence characteristics, interfering pseudogenes, or inadequate coverage). In the case of deletions/duplications, most will be detected including intragenic CNVs and large cytogenetic events. At this time, SV is limited to deletions larger than 1 kb in size, duplications (no size limit) and inversions. For these SVs, the overall sensitivity during validation was 86%. Sensitivity for ≥ 1 kb deletions alone is > 95%. Sensitivity may vary from gene-to-gene based on size, depth of coverage, and characteristics of the region. Because of these technicalities, this test is not 100% sensitive and will not identify all disease-causing genetic variants.
- Even if a disease-causing genetic variant associated with the patient's symptoms is identified, it may not allow for predictions regarding severity of the disease or prognosis.
- It is very important to provide an accurate family history and clinical information as that information is critical for result interpretation. Detailed clinical information (such as clinical features, a family pedigree, and results of prior testing) is required for testing to proceed.
- Additional limitations to this test will be provided in the Supplementary material included with the report.

CONFIDENTIALITY

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

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