

Testing can also be ordered via online portal – please scan or click on QR code.
Please complete every field and tick box clearly.



STEP 1: PATIENT INFORMATION

Patient's First Name Middle Initial Patient's Last Name

Patient's Date of Birth Patient ID/MR Number/External Sample Number

Biological Sex: Male Female Unknown
 Gender Identity (if different from above):

Patient's Street Address City / Town

State Zip Code Country Patient's Preferred Phone Patient's Email

Ethnicity (check all that apply): African-American Asian (China, Japan, Korea) Caucasian/N. European/S. European Finnish French Canadian Hispanic
 Jewish - Ashkenazi Jewish - Sephardic Mediterranean Middle Eastern (Saudi Arabia, Qatar, Iraq, Turkey) Native American E. Indian
 Southeast Asian (Vietnam, Cambodia, Thailand) South Asian (India, Pakistan) Other (specify)

PATIENT SAMPLE INFORMATION

SAMPLE TYPE: Whole Blood Saliva Swab Dried Blood Spots DNA, Source: Other:
 Collection Date: Was this sample collected in the State of NV, NY or OR?: Yes No (If yes, separate consent is required. See forms section of website.)

INDICATION FOR TESTING

Clinical Diagnosis: (medical records/clinical notes are required.) Age at Initial Presentation:

STEP 2: ORDERING PROVIDER AND REPORTING PREFERENCES

Provider's First and Last Name NPI

Clinic/Hospital/Institution Name Provider's Email

Provider's Street Address City / Town State Zip Code Country

Provider's Phone Provider's Fax

How would you like to receive the report?:
 Fax Email Portal

SEND ADDITIONAL COPY OF RESULTS TO (If applicable)

Name Role with patient/Job title Clinic/Hospital/Institution Name

Phone Number Fax Number Email Address

How would you like to receive the report?:
 Fax Email Portal

STEP 3: BILLING INFORMATION

INSTITUTIONAL BILLING

Institution/Organization Name Billing Account ID P.O. Number (if applicable)

Contact Name Contact Phone

PATIENT (SELF) PAYMENT

By providing payment information, you are authorizing Revvity Omics to process payment at the associated charge for tests ordered. Test cost is available on our website, or may be confirmed by calling 877-475-4436. Payment is required prior to test initiation. The patient's sample will be placed on hold (for up to 30 days) until payment is secured. If the patient does not provide payment to Revvity Omics within 30 days, the test order may be canceled. Please note that failure by the patient to respond in a timely fashion to Revvity Omics attempts to obtain payment may cause a delay in the receipt of the results report.

CREDIT CARD (Please fill out all information below) **CHECK:** \$ Amount Enclosed (Please make checks payable to: Revvity Omics, Inc.)

Credit Card Number Exp. Date CVV Cardholder Printed Name as Appears on Card Amount

Credit Card Billing Street Address City / Town State Zip Code

Cardholder Signature Cardholder Phone

CONTACT FOR PAYMENT INFORMATION

Name Phone Email Address

FOR INTERNAL USE ONLY

Date Rec'd	Rec'd	TEMP	SPEC	COL	#TUBES	VOL
R/C/F						
R/C/F						
R/C/F						

Clinical Genomics Test Requisition Form

STEP 4: TEST MENU

WHOLE GENOME SEQUENCING*

UltraRapid TAT

- D2010 Ultrarapid Genomic Testing, Proband Only
- D2330F Ultrarapid Genomic Testing, DUO
- D2310 Ultrarapid Genomic Testing, TRIO
- D2340F Ultrarapid Genomic Testing, QUAD

Standard TAT

- D2000 Whole Genome Sequencing, Proband Only
- D2330 Whole Genome Sequencing, DUO
- D2300 Whole Genome Sequencing, TRIO
- D2340 Whole Genome Sequencing, QUAD

WHOLE EXOME SEQUENCING*

Rapid TAT

- D1010 Rapid Whole Exome Sequencing, Proband Only
- D1330F Rapid Whole Exome Sequencing, DUO
- D1310 Rapid Whole Exome Sequencing, TRIO
- D1340F Rapid Whole Exome Sequencing, QUAD

Standard TAT

- D1000 Whole Exome Sequencing, Proband Only
- D1330 Whole Exome Sequencing, DUO
- D1300 Whole Exome Sequencing, TRIO
- D1340 Whole Exome Sequencing, QUAD

CNGNOME®

- D0900 CNGnome® NGS Array Test

REANALYSIS OPTIONS

- D0580 First Reanalysis on Previous Revvity Whole Exome Sequencing Test⁺
- D0590 First Reanalysis on Previous Revvity Whole Genome Sequencing Test⁺
- D0510 Reanalysis and Interpretation of Previous Revvity Whole Exome Proband
- D0520 Reanalysis and Interpretation of Previous Revvity Whole Genome Proband
- D0598 Add Familial Report to Previous WES TRIO
- D0599 Add Familial Report to Previous WGS TRIO
- D0055 Reanalysis of Secondary Findings of Previous RevvityTest
- D5242 Reflex to Whole Genome Sequencing from Genome Panel

Information from Previous Testing (REQUIRED)[^]

	MM/DD/YYYY	MM/DD/YYYY
Original Proband Accession ID Number	Original Report Date	Proband DOB

OTHER

Other Test Code: _____

Test Name: _____

* For prenatal testing options and requisition forms, please visit our website.

⁺ Complimentary test selection will be reviewed for qualification prior to initiating testing. Billing method is still required for processing

[^] Can be found at the top of the first page of original Revvity test report.

STEP 5: FAMILIAL INFORMATION (Required with DUO, TRIO and QUAD orders)

FAMILY MEMBER 1

Last name, First name

Relationship to Patient

Date of Birth: MM/DD/YYYY Symptomatic (clinically affected): Yes No Sample Included - Collection Date MM/DD/YYYY To be sent later

FAMILY MEMBER 2

Last name, First name

Relationship to Patient

Date of Birth: MM/DD/YYYY Symptomatic (clinically affected): Yes No Sample Included - Collection Date MM/DD/YYYY To be sent later

FAMILY MEMBER 3

Last name, First name

Relationship to Patient

Date of Birth: MM/DD/YYYY Symptomatic (clinically affected): Yes No Sample Included - Collection Date MM/DD/YYYY To be sent later

Clinical Genomics Test Requisition Form

STEP 6: SECONDARY FINDINGS AND ENHANCEMENTS

SECONDARY FINDINGS ^{^+}

- ACMG
- Carrier Status
- Pharmacogenetics
- Other Pathogenic Findings

For minors, check if you want **only** pediatric-onset findings reported for these selected categories (if not checked, both adult-onset and pediatric-onset conditions will be reported):

- ACMG
- Other Pathogenic Findings

[^] Secondary findings are only provided with Whole exome testing options and standard turn-around Whole genome. Secondary findings are not available with other tests, including CNGnome, prenatal testing of ongoing pregnancies, or reanalysis tests. If Secondary findings are desired along with ultrarapid WGS or a reanalysis order, D0055 needs to also be ordered.

⁺ For more information on the secondary finding categories, please reference clinical informed consent form.

ULTRARAPID WGS OPTIONAL ENHANCEMENTS*

- StepOne Comprehensive Biochemical Profile** - a newborn screening test that can detect more than 70 disorders.
- Metagenomic Clinical Research Report** – The patient consents to participate in a metagenomic screening clinical research study conducted by Revvity. In this study, whole genome sequencing data will be used to screen for pathogenic microorganisms in the patient’s sample (at the time of specimen collection) to assist in unbiased culture-independent detection. A research-use-only report will be provided with screening results on potentially disease-causing microbes in the patient’s sample. IRB: 1281574
- Congenital Cytomegalovirus (cCMV) Screening** – Not valid after 21 days of age.

* **StepOne and cCMV screening are only available with the submission of a DBS sample. Metagenomics research is available on DBS or whole blood.**

STEP 7: DATA PERMISSIONS*

1. Check here if you would like to opt out of anonymized sample retention. Note, if not checked, this is interpreted as “consent given.”
2. Check here if you would like to opt out of anonymized data retention. Note, if not checked, this is interpreted as “consent given.”
3. Check here if you would like to opt out of your contact information being shared by Revvity to external researchers for direct communication regarding their studies.
Note, if not checked, this is interpreted as “consent given.”
4. Check here if sample(s) was or will be collected in the state of NV NY or OR. Additional consent must be completed, which can be found on the Revvity Omics website.

* For more information on data permissions options, please reference Page 5.

STEP 8: PHYSICIAN CONFIRMATION OF INFORMED CONSENT AND MEDICAL NECESSITY

The undersigned person (or designated representative thereof) certifies that: (a) he/she is a licensed medical professional authorized to order the testing ordered herein; (b) he/she fully complies with all applicable federal, state, and local laws, regulations, and rules, including but not limited to those governing genetic testing, informed consent, and patient consent and authorization requirements for the test(s) ordered; (c) he/she will obtain informed consent of the patient in compliance with all applicable laws and regulations, which shall include, to the extent applicable: (i) a statement of the purpose of the test(s) ordered; (ii) a statement that prior to signing the consent form, the consenting person discussed with the medical practitioner ordering the test the reliability of positive or negative test results and the level of certainty that a positive test result for that disease or condition serves as a predictor of such disease; (iii) a statement that the consenting person was informed about the availability and importance of genetic counseling and provided with written information identifying a genetic counselor or medical geneticist from whom the consenting person might obtain such counseling; (iv) a general description of each disease or condition tested for; and (v) the person or persons to whom the test results may be disclosed; (d) he/she will maintain, as part of the patient’s record, documentation of the patient’s informed consent and authorization for the test(s) ordered that complies with applicable laws and regulations, and will make such documentation available to Revvity upon request; (e) tests ordered are medically necessary and results may impact medical management for the patient; and (f) the information provided on this Clinical Genomics Test Requisition Form is complete, true, and accurate to the best of his/her knowledge.

Signature _____ Date _____

Clinical Genomics Test Requisition Form

DETAILED MEDICAL RECORDS, PREVIOUS TEST RESULTS AND FAMILY HISTORY MUST BE ATTACHED FOR ALL CASES.
CLINICAL INFORMATION IS CRUCIAL FOR ACCURATE INTERPRETATION OF RESULTS.

ADDITIONAL PHENOTYPE / PATIENT HISTORY SECTION (Check all that apply)

Clinical diagnosis: _____ Age of manifestation: _____

NEUROLOGY

1. Neurodevelopmental abnormality

- 1.1 Autism
- 1.2 Attention deficit disorder
- 1.3 Global developmental delay
- 1.4 Delayed motor development
- 1.5 Delayed language development
- 1.6 Developmental regression
- 1.7 Intellectual disability

2. Brain imaging

- 2.1 Abnormal myelination
- 2.2 Agenesis of corpus callosum
- 2.3 Brain atrophy
- 2.4 Cerebellar hypoplasia
- 2.5 Heterotopia
- 2.6 Holoprosencephaly
- 2.7 Hydrocephalus
- 2.8 Leukodystrophy
- 2.9 Lissencephaly

3. Movement abnormality

- 3.1 Ataxia
- 3.2 Chorea
- 3.3 Dystonia
- 3.4 Parkinsonism

4. Neuromuscular abnormality

- 4.1 Muscular hypotonia
- 4.2 Muscular hypertonia
- 4.3 Hyperreflexia
- 4.4 Spasticity

5. Seizures

- 5.1 Febrile seizures
- 5.2 Focal seizures
- 5.3 Generalized seizures

6. Others

- 6.1 Craniosynostosis
- 6.2 Dementia
- 6.3 Encephalopathy
- 6.4 Headache / Migraine
- 6.5 Macrocephaly
- 6.6 Microcephaly
- 6.7 Neuropathy
- 6.8 Stroke

METABOLISM

- 1. Elevated creatine kinase
- 2. Decreased plasma carnitine
- 3. Hyperalaninemia
- 4. Hypoglycemia
- 5. Increased CSF lactate
- 6. Increased serum pyruvate
- 7. Ketosis
- 8. Lactic acidosis
- 9. Organic aciduria

EYE

- 1. Blepharospasm
- 2. Cataract
- 3. Coloboma
- 4. Glaucoma
- 5. Microphthalmos
- 6. Nystagmus
- 7. Ophthalmoplegia
- 8. Optic atrophy
- 9. Ptosis
- 10. Retinitis pigmentosa
- 11. Retinoblastoma
- 12. Strabismus
- 13. Visual impairment

MOUTH, THROAT AND EAR

- 1. Abnormality of dental color
- 2. Cleft lip / palate
- 3. Conductive hearing impairment
- 4. External ear malformation
- 5. Hypodontia
- 6. Sensorineural hearing impairment

SKIN, INTEGUMENT AND SKELETAL

1. Skeletal

- 1.1 Abnormal limb morphology
- 1.2 Abnormal vertebral column
- 1.3 Joint hypermobility
- 1.4 Multiple joint contractures
- 1.5 Polydactyly
- 1.6 Scoliosis
- 1.7 Syndactyly
- 1.8 Talipes equinovarus

2. Skin and integument

- 2.1 Abnormal skin pigmentation
- 2.2 Abnormal hair
- 2.3 Abnormal nail
- 2.4 Hyperextensible skin
- 2.5 Ichthyosis

CARDIOVASCULAR

- 1. Angioedema
- 2. Aortic dilatation
- 3. Arrhythmia
- 4. Coarctation of aorta
- 5. Defect of atrial septum
- 6. Defect of ventricular septum
- 7. Dilated cardiomyopathy
- 8. Hypertrophic cardiomyopathy
- 9. Lymphedema
- 10. Malf. of heart and great vessels
- 11. Myocardial infarction
- 12. Tetralogy of Fallot

GASTROINTESTINAL, GENITOURINARY, ENDOCRINE

1. Gastrointestinal

- 1.1 Aganglionic megacolon
- 1.2 Constipation
- 1.3 Diarrhea
- 1.4 High hepatic transaminases
- 1.5 Gastroschisis
- 1.6 Hepatic failure
- 1.7 Hepatomegaly
- 1.8 Obesity
- 1.9 Pyloric stenosis
- 1.10 Vomiting

2. Genitourinary

- 2.1 Hydronephrosis
- 2.2 Renal agenesis /hypoplasia
- 2.3 Renal cyst
- 2.4 Renal tubular dysfunction

3. Endocrine

- 3.1 Diabetes mellitus
- 3.2 Hypothyroidism
- 3.3 Hyperparathyroidism
- 3.4 Hypoparathyroidism
- 3.5 Hyperthyroidism

REPRODUCTION

- 1. Abnormal external genitalia
- 2. Abnormal internal genitalia
- 3. Hypogonadism
- 4. Hypospadias
- 5. Infertility

ONCOLOGY

- 1. Adenomatous polyposis
- 2. Breast carcinoma
- 3. Colorectal carcinoma
- 4. Leukemia
- 5. Myelofibrosis
- 6. Neoplasm of the lung
- 7. Neoplasm of the skin
- 8. Paraganglioma
- 9. Pheochromocytoma

HEMATOLOGY AND IMMUNOLOGY

- 1. Abnormality of coagulation
- 2. Anemia
- 3. Immunodeficiency
- 4. Neutropenia
- 5. Pancytopenia
- 6. Abnormal hemoglobin
- 7. Splenomegaly
- 8. Thrombocytopenia

PRENATAL AND DEVELOPMENT

- 1. Failure to thrive
- 2. Hemihypertrophy
- 3. Hydrops fetalis
- 4. IUGR
- 5. Oligohydramnios
- 6. Overgrowth
- 7. Polyhydramnios
- 8. Premature birth
- 9. Disproportionate short stature
- 10. Proportionate short stature
- 11. Tall stature

OTHER (INCLUDING DYSMORPHIC FACIAL FEATURES AND OTHER DESCRIPTORS):

The purpose of this Informed Consent Form (ICF) is to provide you with a description of the Test ordered, known risks and benefits of the Test, anonymization of personal health information ("PHI"), sample and data retention, research opportunities, and the reporting of secondary findings, if applicable.

TEST INFORMATION

Your healthcare provider ("HCP") has recommended that you, or your dependent, receive clinical testing ("Test") indicated on the submitted Test Requisition Form ("Requisition"). For more information on the reasons your HCP has ordered the Test, and the disorders your HCP is testing for, please consult with your HCP. Providing a Sample and undergoing the Test is voluntary.

Enzyme/Biomarker Test: This type of test measures the presence or absence of enzymes/biomarkers and/or their level of activity in an individual. Only the enzymes/biomarkers identified on the requisition will be tested. Results from this type of Test may indicate the presence of a specific condition or conditions, and follow-up confirmatory testing may be recommended.

Genetic/Genomic Test: This type of Test analyzes one or more segments of your DNA depending on the assay requested. This Test is used to identify what, if any, DNA variant(s) you or your dependent possesses that is causing the specific disease, condition or risk you are being tested for. Identifying the mutation may be useful for diagnostic and treatment purposes, and allows at-risk family members to be tested. In some cases, we may not be able to determine with certainty which gene is actually causing a disease.

TEST METHOD

If you consent to the Test, your HCP will take a sample of your and/or your dependent's blood, saliva, body fluid, tissue or other sample type. The Sample will be sent to Revvity's laboratories in the United States for the Test; the majority of testing will be performed at our laboratory headquarters in Pittsburgh, PA.

Under some circumstances, including inadequate or poor quality sample, an additional Sample may be required for Tests to be performed.

TEST RESULTS

Your treating HCP has sole responsibility for all decisions concerning the possible management of your diagnosis and disease; Revvity will not provide a diagnosis. Revvity will report Test results via secure email, a secure internet portal, or fax. Your HCP is responsible for communicating with you regarding the results of the Test and may refer you or your dependent to a specialist for further clinical evaluation and confirmation of diagnosis, if applicable. Possible results for Genetic/Genomic Tests include:

1. *Positive:* A positive genetic test result may indicate that you are a carrier of, predisposed to, or have the specific disease or condition being tested for.
2. *Negative:* A negative result indicates that no disease-causing variant was identified in the Test performed. No Test can rule out all genetic diseases or conditions. A negative result does not guarantee that you are free from genetic disorders or other medical conditions.
3. *Inconclusive/Variant of Uncertain Significance:* A variant of uncertain significance (VOUS) result indicates that a DNA change was detected, but it is currently unknown if the variant is associated with a genetic disorder. A VOUS is not the same as a positive result and does not clarify whether there is an increased risk to develop a genetic disorder. The variant could be a benign change or it could be indicative of disease/disease-causing.
4. *Unexpected Results:* This Test may reveal an important genetic change that is not directly related to the reason for ordering this test. This information would be disclosed to your HCP if it potentially impacts medical care, and you have consented to receive this type of result.

TEST REPORT

Variants are described as pathogenic, likely pathogenic, or variant of uncertain significance in genes interpreted to be responsible for, or potentially contributing to, a disease or condition. For testing performed on prenatal samples, or for screening of apparently healthy individuals, only variants classified as pathogenic or likely pathogenic will be reported.

When Whole Exome Sequencing (WES) or Whole Genome Sequencing (WGS) tests are ordered by your HCP, you may have the option to receive some findings not directly related to the reason for ordering the Test called "Secondary Findings". When Secondary Findings are requested, only Pathogenic or Likely Pathogenic findings will be reported, where applicable. Please read the Secondary Findings sections on page 3 and/or 4 of this consent form for more information. Secondary findings are not available for all Tests.

INFORMATION ABOUT PARENTAL AND FAMILIAL SAMPLES

In some circumstances, it may be helpful for additional family members to undergo testing in order to provide information that can aid in the interpretation of the Test results. These Tests could be part of a WES/WGS TRIO Test or as stand-alone targeted testing. If the HCP recommends testing for additional family members, family members may have the option to receive information about secondary findings either as a part of the proband report or as a standalone parental report. A full analysis of the parental samples for secondary findings will only be completed if standalone reports are ordered (for an additional charge). In conjunction with proband testing, any variants reported in the proband will include inheritance information from family members included in the study.

TEST LIMITATIONS

Due to current limitations in technology and incomplete knowledge of diseases and genes, some variants may not be detected by the Test ordered. There is a possibility that the Test result is uninterpretable or deemed of unknown significance. Further testing may be required when more information is gained. In rare circumstances, Test results may be suggestive of a condition different from that which was originally considered for the purpose of consenting to this Test. The Test may also find variants or genes that lead to conditions for which you currently do not have symptoms or may not be related to your current condition.

TEST RISKS

Patients and family members may experience anxiety before, during, and/or after testing. Testing multiple family members may reveal that familial relationships are not biologically what they were assumed to be. For example, the Test may indicate non-paternity (the stated father of an individual is not the biological father) or consanguinity (the parents of an individual are closely related by blood). These biological relationships may need to be reported to the HCP who ordered the test.

Taking a blood or tissue sample from you and/or your child may lead to mild pain, bruising, swelling, redness, and a slight risk of infection. Light-headedness, fainting or nausea may occur if your HCP collects blood or tissue samples. These side-effects are typically brief and transient, but you should contact your HCP if you and/or your child require treatment. Under some circumstances an additional sample may be required for Tests to be performed.

A positive test result may limit your access to health insurance or life assurance coverage; for example, a life insurance company might ask you to provide genetic information indicating a disorder if this information is available to you. Please refer to information on the Genetic Information Nondiscrimination Act (GINA) and applicable local laws for more information.

CONFIDENTIALITY

You have the right to confidential treatment of the Sample and your PHI. Your HCP will provide Revvity with Personal Health Information (“PHI”) such as your name, date of birth, gender and clinical symptoms to help track your sample and report results. To maintain confidentiality, the test results will only be released to the referring health care provider, to others designated by the referring health care provider as being involved in your care, to the ordering laboratory, to the patient/guardian, to other health care providers involved in your diagnosis and treatment, or as otherwise required by law or regulation. Unless required by law, Revvity will not disclose your PHI to any person or entity except with your written consent.

You and your HCP can control how your Sample and PHI are processed. You have the right to request access to your PHI, request corrections of any errors in recorded PHI, or where PHI may be missing or incomplete ask that it be completed. You also have the right to ask that your PHI be erased, subject to law or regulation. You can contact your HCP for such requests and your HCP will contact Revvity, or you can contact Revvity directly by visiting www.revvity.com. If requests for access, correction, completion, or erasure cannot be fulfilled, you will be informed and provided with the reasons why your requests cannot be fulfilled.

SAMPLE AND DATA RETENTION

Pursuant to laboratory best practices, your DNA sample will be retained by Revvity for a minimum of two years and then destroyed. Additionally, your PHI, the data from the Tests (including those performed before any withdrawal of consent) and the related reports will be retained by Revvity indefinitely, unless otherwise noted. In some instances, it may be beneficial to you for Revvity to retain your sample for a longer period of time in order to conduct additional testing, and Revvity will do so with appropriate documentation from you or your HCP.

Revvity is requesting consent to keep your and/or your child’s anonymized sample and data indefinitely for ongoing test development, scientific research, and/or other activities. Consent is optional, and the Test will be performed whether or not you provide consent to the following, as selected by your HCP on page 2 of the Test Requisition Form:

- Revvity will anonymize and retain your Sample indefinitely for internal quality control, test validation, assay development and improvement. By allowing Revvity to retain your Sample, you understand and agree that you give up any property rights you may have in the Sample and are donating it to Revvity Omics, Inc. If you withdraw your consent, no additional tests or anonymization will be carried out on your Sample; no results will be reported and your sample, reports and data that have not been anonymized will be destroyed.
- Revvity will anonymize your data and retain the anonymized data and related anonymized reports from your Tests indefinitely for internal statistical, quality analysis, research, scientific and technical development, and market research.

RESEARCH OPTIONS

Revvity may collaborate with scientists, researchers and drug developers to advance knowledge of genetic diseases. If there are opportunities to participate in future research relevant to the disease in you and/or your child, Revvity may contact you or your HCP about the development of new testing, drug development, or other treatments. Revvity may also work with scientists or researchers from academic or commercial institutions who have received the necessary approvals to conduct a research study. In some instances, these scientists or researchers may like to contact you directly about your interest in participating in a specific research study. You may opt-out of research on page 2 of the Test Requisition Form.

WITHDRAWAL OF CONSENT

I understand this consent is voluntary and is valid until I withdraw my consent. I understand I may withdraw my consent to sample and data retention, and to the Test at any time, that Revvity will not perform the Test unless consent has been obtained by the HCP. If I withdraw any consent, it will not affect actions taken before I withdrew my consent, including any anonymization of data or of my Sample. I understand that if I wish to withdraw my consent I should contact Revvity via email at: genomics@revvity.com or toll-free by telephone +1-866-354-2910 to request withdrawal.

SECONDARY FINDINGS: APPLIES ONLY TO WES/WGS

- 1. ACMG Recommended Secondary Findings:** The American College of Medical Genetics and Genomics (ACMG), has recommended that secondary findings should be offered for a specific subset of highly penetrant and medically actionable genes associated with various inherited disorders for all individuals undergoing WGS or WES. Please refer to the latest version of the ACMG Recommendations for Reporting of Secondary Findings in Clinical Exome and Genome Sequencing for complete details of genes and conditions at www.acmg.net. Medically-actionable conditions are those for which there is currently recommended treatment or preventative actions that can be taken to reduce the risk of developing the disease. An example would be hereditary cancer syndromes such as Lynch syndrome. Please note that only Pathogenic or Likely Pathogenic variants will be reported if this category of Secondary Findings is selected. We are unable to guarantee that the Test will find all medically-actionable conditions for which you have a pathogenic or likely pathogenic variant. You may have a pathogenic or likely pathogenic variant for a condition in which there was little or no coverage in the Test and therefore will not be detected. Additional testing for health purposes should be discussed with your doctor or genetic counselor.
- 2. Pharmacogenetic variants:** This category of Secondary Findings will include changes in the DNA that do not cause a disease but may be related to how your body processes certain medications, such as chemotherapy drugs, antipyretics, antidepressants, anticoagulants, and others. These variants may tell you how well medications will work or if you will have side effects if you do take the medications now or in the future.
- 3. Carrier status (ex. cystic fibrosis):** This category of Secondary Findings will include carrier findings for a select list of autosomal recessive conditions. The list of genes included in the carrier reporting is available upon request. A recessive condition is one in which two disease-causing variants in the same gene are typically required in order to show symptoms of the disease (one variant is inherited from each parent). Someone who has only one disease-causing variant is called a carrier. Please note that only Pathogenic or Likely Pathogenic variants will be reported if this category of Secondary Findings is selected. Further testing may be necessary to look for a second disease-causing variant in that gene. The Test is not designed to be a comprehensive carrier test. We are unable to guarantee that all conditions for which you are a carrier will be determined by the Test. You may be a carrier for a condition in which there was little or no coverage in the Testing and therefore will not be detected. Additional carrier testing for reproductive purposes should be discussed with your doctor or genetic counselor.
- 4. Diagnostic findings in all other disease-causing genes not related to your clinical features:** This category of Secondary Findings will include conditions that are medically-actionable but not included in the ACMG-recommended list, as well as conditions that are not medically-actionable (do not have recommended treatment or preventative measures). An example would be Alzheimer’s disease. Please note that only Pathogenic or Likely Pathogenic variants will be reported if this category of Secondary Findings is selected. Furthermore, we are unable to guarantee that the Test will find all disease-causing variants in all disease-causing genes. You may have a disease-causing variant for a condition in which there was little or no coverage in the Test and therefore will not be detected. Additional testing for health purposes should be discussed with your doctor or genetic counselor.