Complete, print and submit.

Reset Form

MAYO CLINIC LABORATORIES

Whole Exome Sequencing Plus Pharmacogenomics: Ordering Checklist

Complete Paperwork (required)

- ☐ Whole Exome Sequencing: Patient Information
- ☐ Whole Exome Sequencing: Informed Consent

Attach Clinical Documentation (required)

- ☐ Clinic notes from specialists relevant to patient's clinical features
- ☐ Pedigree

Required Samples **Do not put the patient's name/label on the parent specimens.**

- ☐ Patient's Sample
- ☐ Mother's Sample
- ☐ Father's Sample

Instructions

Send required paperwork and clinical documentation with the samples. Fax a copy of paperwork to the Molecular Genetics Laboratory, Attn: WES Genetic Counselors at 507-284-0670.

Questions: Call with any questions and ask to speak to a WES genetic counselor: 507-293-7299.

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Whole Exome Sequencing Plus Pharmacogenomics: Patient Information

Instructions: Provide the requested information below for appropriate interpretation of the Whole Exome Sequencing Plus Pharmacogenomics (WESSP) test result. **In addition, submit relevant clinic notes and pedigree.**

Patient Information (required)

Patient Name *(Last, First, Middle)*	Medical Record No.	Birth Date *(mm-dd-yyyy)*	Gender ☐ Male ☐ Female
Referring Provider *(Last, First)*		Phone	Fax*
Other Contact/Geneticist/Genetic Counselor *(Last, First)*		Phone	Fax*

**Fax number given must be from a fax machine that complies with applicable HIPAA regulations.*

Ethnic/Ancestral Background

☐ Caucasian ☐ Eastern European ☐ Native American ☐ Pacific Islander
☐ African American ☐ Northern European ☐ Hispanic ☐ Asian
☐ Ashkenazi Jewish ☐ Western European ☐ Middle Eastern ☐ Other: _____
History of consanguinity: ☐ No ☐ Yes, provide relationship details: _____

Biological Family Member Information

Samples from both biological parents are required for WESSP testing. Order WESSP on the patient and both parents; label the parental samples with full name and birth date. **Do not label the parental samples with the child's name.**

Mother's Information

Mother Name *(Last, First, Middle)*	Medical Record No.	Birth Date *(mm-dd-yyyy)*

Does this relative share any relevant clinical features or clinical history with the patient? ☐ Yes ☐ No If yes, describe:

Father's Information

Father Name *(Last, First, Middle)*	Medical Record No.	Birth Date *(mm-dd-yyyy)*

Does this relative share any relevant clinical features or clinical history with the patient? ☐ Yes ☐ No If yes, describe:

Patient Name *(Last, First, Middle)*	Birth Date *(mm-dd-yyyy)*

Previous Clinical Evaluations Performed Indicate the previous tests and evaluations performed for this patient and provide details regarding the specific tests and pertinent results below. It will be assumed that any evaluations left blank were not performed or are unknown.

Karyotype	☐ Not performed	☐ Normal	☐ Abnormal: _____
Chromosomal Microarray	☐ Not performed	☐ Normal	☐ Abnormal: _____
Gene Sequencing/Panel*	☐ Not performed	☐ Normal	☐ Abnormal: _____
Methylation/UPD*	☐ Not performed	☐ Normal	☐ Abnormal: _____
Mitochondrial DNA*	☐ Not performed	☐ Normal	☐ Abnormal: _____
Metabolic Work-up*	☐ Not performed	☐ Normal	☐ Abnormal: _____
Brain MRI	☐ Not performed	☐ Normal	☐ Abnormal: _____
Brain Spectroscopy	☐ Not performed	☐ Normal	☐ Abnormal: _____
Electroencephalogram (EEG)	☐ Not performed	☐ Normal	☐ Abnormal: _____
Echocardiogram	☐ Not performed	☐ Normal	☐ Abnormal: _____
Electrocardiogram (ECG/EKG)	☐ Not performed	☐ Normal	☐ Abnormal: _____
Skeletal Survey	☐ Not performed	☐ Normal	☐ Abnormal: _____
Renal Imaging	☐ Not performed	☐ Normal	☐ Abnormal: _____
Muscle Biopsy	☐ Not performed	☐ Normal	☐ Abnormal: _____
Electromyogram (EMG)	☐ Not performed	☐ Normal	☐ Abnormal: _____
Ophthalmology Exam	☐ Not performed	☐ Normal	☐ Abnormal: _____
Audiology Evaluation	☐ Not performed	☐ Normal	☐ Abnormal: _____

*Describe Details

Suspected Diagnoses List any suspected diagnoses that you would like considered for this evaluation.

Patient Name *(Last, First, Middle)*	Birth Date *(mm-dd-yyyy)*

Patient Clinical Features Check all that apply and provide additional descriptions if available.

Perinatal History
- ☐ Prematurity
- ☐ Increased NT/cystic hygroma
- ☐ IUGR
- ☐ Oligohydramnios
- ☐ Polyhydramnios

Description:

Growth
- ☐ Asymmetric growth
- ☐ Failure to thrive
- ☐ Obesity
- ☐ Overgrowth
- ☐ Short stature
- ☐ Tall stature

Description:

Behavioral/Psychiatric
- ☐ ADHD
- ☐ Autism spectrum disorder
- ☐ Oppositional-defiant disorder
- ☐ Obsessive-compulsive disorder
- ☐ Psychiatric diagnosis:

Description:

Cognitive/Developmental
- ☐ Intellectual disability/MR
  - ☐ Mild
  - ☐ Moderate
  - ☐ Severe
- ☐ Motor delay
- ☐ Speech delay
- ☐ Developmental regression

Description:

Craniofacial
- ☐ Cleft lip
- ☐ Cleft palate
- ☐ Craniosynostosis
- ☐ Dysmorphic features
- ☐ Ear malformation
- ☐ Macrocephaly
- ☐ Microcephaly
- ☐ Synophrys

Description:

Gastrointestinal
- ☐ Anal atresia
- ☐ Gastroschisis
- ☐ Hirschsprung disease
- ☐ Liver failure
- ☐ Omphalocele
- ☐ Pyloric stenosis
- ☐ Tracheoesophageal fistula

Description:

Genitourinary
- ☐ Ambiguous genitalia
- ☐ Cliteromegaly
- ☐ Cryptorchidism
- ☐ Hydronephrosis
- ☐ Hypogonadism
- ☐ Hypospadias
- ☐ Nephrotic syndrome
- ☐ Renal agenesis
- ☐ Renal malformation
- ☐ Renal tubulopathy

Description:

Skin/Hair/Dental
- ☐ Abnormal fingernails
- ☐ Abnormal hair
- ☐ Abnormal skin
- ☐ Dental anomalies
- ☐ Hemangioma
- ☐ Hyperpigmentation
- ☐ Hypopigmentation

Description:

Neuromuscular
- ☐ Ataxia
- ☐ Autonomic dysfunction
- ☐ Cerebral palsy
- ☐ Dementia
- ☐ Dystonia
- ☐ Encephalopathy
- ☐ Hypotonia
- ☐ Muscle weakness
- ☐ Peripheral neuropathy
- ☐ Seizures
- ☐ Spasticity
- ☐ Stroke/TIAs
- ☐ Structural brain anomaly

Description:

Musculoskeletal
- ☐ Club foot
- ☐ Contractures
- ☐ Diaphragmatic hernia
- ☐ Foot deformity
- ☐ Joint laxity
- ☐ Limb anomaly
- ☐ Oligodactyly
- ☐ Polydactyly
- ☐ Scoliosis
- ☐ Skeletal dysplasia
- ☐ Syndactyly
- ☐ Vertebral anomaly

Description:

Ophthalmologic
- ☐ Aniridia
- ☐ Blindness
- ☐ Cataracts
- ☐ Coloboma
- ☐ Microphthalmia
- ☐ Myopia
- ☐ Ophthalmoplegia
- ☐ Optic atrophy
- ☐ Ptosis
- ☐ Retinitis pigmentosa

Description:

Cardiovascular
- ☐ Aortic dilatation/dissection
- ☐ Arrhythmia
- ☐ Arterial dilatation/dissection
- ☐ Atrial septal defect
- ☐ AV canal defect
- ☐ Bicuspid aortic valve
- ☐ Coarctation of the aorta
- ☐ Cardiomyopathy
- ☐ Hypoplastic left heart
- ☐ Pulmonic stenosis
- ☐ Tetralogy of Fallot
- ☐ Ventricular septal defect

Description:

Hearing
- ☐ Sensorineural hearing loss
- ☐ Conductive hearing loss
- ☐ Mixed hearing loss

Description:

Cancer/Neoplastic

Age of onset: _____

Tumor type: _____

Location: _____

Hematologic/Immunologic
- ☐ Anemia
- ☐ Immunodeficiency
- ☐ Iron deficiency
- ☐ Neutropenia
- ☐ Pancytopenia
- ☐ Thrombocytopenia

Description:

Endocrine
- ☐ Adrenal abnormality
- ☐ Diabetes, Type I
- ☐ Diabetes, Type II
- ☐ Gonadal abnormality
- ☐ Hypothalamic abnormality
- ☐ Parathyroid abnormality
- ☐ Pituitary abnormality
- ☐ Thyroid abnormality

Description:

Metabolic/Mitochondrial
- ☐ Abnormal CPK
- ☐ Abnormal plasma carnitine/acylcarnitine
- ☐ Elevated pyruvate
- ☐ Elevated alanine
- ☐ Hypoglycemia
- ☐ Ketosis
- ☐ Lactic acidosis
- ☐ Organic aciduria
- ☐ Ragged red fibers

Description:

Additional Details/Clinical History

Patient Name *(Last, First, Middle)*	Birth Date *(mm-dd-yyyy)*

MAYO CLINIC LABORATORIES

Whole Exome Sequencing Plus Pharmacogenomics: Informed Consent

This form is provided to ensure that you are informed about a genetic test called whole exome sequencing. Whole exome sequencing is a complex genetic test. Genetic counseling is recommended to help you more fully understand the risks and benefits associated with whole exome sequencing. It is your choice whether or not to have this test. Your signature is required on page 7 before the laboratory will proceed with whole exome sequencing.

What is Whole Exome Sequencing?

- Whole exome sequencing is a test that detects changes (variants) in a patient's genetic code (DNA) which may be causing a genetic disorder. Humans have approximately 20,000 genes. Variants in certain important portions of these genes, the exons (coding regions), account for the majority of the variants that cause genetic disorders. Taken together, all of our exons make up the "exome."
- The goal of whole exome sequencing is to identify genetic variants that may provide or confirm a specific diagnosis for a patient.

How is Whole Exome Sequencing performed?

- A blood draw or other procedure will be required to obtain samples from a patient and both biological parents. DNA is obtained from the 3 samples and sequenced to identify genetic variants in the exome.
- The laboratory evaluates certain characteristics of each variant (such as the type of genetic change, whether the parents have this change, and how common it is in the general population) in order to determine whether it could cause a genetic disorder in a patient.

What are the potential benefits of Whole Exome Sequencing?

- Genetic variants may be detected that will explain a patient's clinical features and provide a diagnosis.
- Establishing a specific diagnosis may allow for a better prediction of the outcome or course of a disorder. It may also help to determine the best medical management for a patient, such as surveillance, treatment, or preventive measures.
- Identification of a disease-causing variant may also allow for a more accurate risk estimate and/or diagnosis of at-risk or affected family members.

What are the potential risks of Whole Exome Sequencing?

- If a disease-causing variant is found and a specific diagnosis is made, it may not change the medical management that was previously recommended. There also may not be a treatment available for the disorder.
- In some cases, a health care provider may recommend additional tests to better understand the results from whole exome sequencing.
- Other possible risks, such as those associated with financial/insurance considerations, psychological effects, and implications for family members should be discussed with your health care provider.

What are the limitations of Whole Exome Sequencing?

- Whole exome sequencing will not establish a diagnosis for all patients who have the test.
- At this time, greater than 95% of the exome can be sequenced well enough to be interpreted. This means that about 5% of the exome will not be analyzed. If a disease-causing genetic variant exists in an unanalyzed region, it would not be detected.
- Because whole exome sequencing focuses on the most important regions of genes (the exons), variants in other areas of genes may be missed. Certain types of variants, such as large deletions/insertions or trinucleotide repeat expansions, may not be detected by this test.
- Scientific understanding of the role of genes and variants in human diseases is not complete. Therefore, the significance of some variants that are found may not be known. Patients are encouraged to contact their health care provider for updates regarding their test results, as understanding may change with time.
- The laboratory's interpretation is based upon the accuracy of the clinical information and family history provided by the ordering health care provider. If pertinent information is not provided, this may affect whether certain variants are reported.

What types of test results will the laboratory report?

- *Variants in genes associated with the patient's clinical features:* Variants in genes known to cause conditions that have features which overlap with the patient's clinical features will be reported (including carrier status for recessive conditions). Variants in these genes will be reported if they are known or expected to cause the genetic condition. Variants of uncertain significance in these genes will also be reported.
- *Variants in genes of uncertain significance:* Variants may be found in genes that are suspected, but not certain, to play a role in human disease. Variants in these genes of uncertain clinical significance may be reported if there is a high degree of suspicion that they are causing a patient's clinical features.
- *Pharmacogenomic variants:* Select genetic variants that impact how a person responds to certain medications will be reported.

Patient Name *(Last, First, Middle)*	Birth Date *(mm-dd-yyyy)*

Will secondary findings be reported?

- Secondary findings are variants that cause health problems for which some type of intervention or preventive measure is available, but that are unrelated to the reason that a patient is having whole exome sequencing. **You can choose whether or not to receive secondary findings.**
- Knowledge of a person's risk for these conditions can help to determine the medical actions available to maintain that person's health, such as screening for cancer or specific heart conditions.
- These results may lead to increased anxiety or worry. They may also result in additional medical interventions.
- Secondary findings will only be reported if they are in one of the 59 genes currently recommended by the American College of Medical Genetics and Genomics (ACMG), and only if they are known or expected to cause disease. Variants of uncertain significance will not be reported in these genes.

Why are parents tested and what types of test results will they receive?

- Parents' samples are required to analyze their child's results. Interpretation of genetic variants is more accurate when the laboratory is able to compare the results between the parents and their child.
- Parents will not receive full test reports for their samples.
- If the patient's reported genetic variants are inherited from a parent, this will be indicated in the patient's report (with the exception of pharmacogenomics results). Parents may learn about a diagnosis of a genetic condition, increased risk for health concerns, or carrier status for a recessive condition.
- Variants present in a parent that are absent from the patient will not be reported.

What else could whole exome sequencing results reveal about family members?

- Because samples from both biological parents are required for whole exome sequencing, it is possible to uncover that a parent is unrelated to a patient due to mis-attributed paternity, maternity, or adoption. In this situation, generating test results will not be possible and testing will be canceled. The patient may be charged for laboratory work that was already performed on the samples.
- In some cases, whole exome sequencing results may suggest that the parents of a patient are biologically related, such as first cousins or another familial relationship.

What types of test results will the laboratory not report?

- *Benign and likely benign variants:* Variants that are known or predicted to be benign (not disease causing) will not be reported.
- *Other secondary findings:* Variants in genes associated with conditions that are not related to a patient's reported clinical features will not be reported, with the possible exception of the secondary findings described above.

What does a negative whole exome sequencing report mean?

- A negative report means that whole exome sequencing did not detect a variant that could be related to a patient's clinical features. However, there may still be a genetic explanation for a patient's features. Because of the testing limitations noted above, a disease causing variant may have been missed.

How will the test results become available?

- The laboratory will release a patient's test report directly to the ordering health care provider and it will become part of a patient's medical record.
- Requests for the raw data obtained from whole exome sequencing should be directed to the laboratory. The laboratory is not responsible for providing software or other tools needed to visualize, filter, or interpret this data.

Will my test results be shared with databases or researchers?

- Mayo Clinic is an active participant in the National Institutes of Health-funded Clinical Genome Resource (ClinGen) and shares information about genetic variants identified through clinical genetic testing with publicly available databases (ClinVar).
- No patient-identifying information (ie, name, birth date) is shared.
- Genomic data sharing enables health care providers, clinical laboratories, and researchers to share experiences. This can lead to improved interpretations of genetic test results.

What will happen to my blood and DNA after testing is complete?

- The laboratory does not guarantee indefinite storage of patient samples and may discard them within 60 days of test completion, in accordance with state-specific regulations.
- Any sample remaining after testing is complete may be used for internal laboratory quality control or research purposes, after the removal of patient identifiers such as name and birth date. **You may request that your DNA sample not be used for these purposes by indicating this preference below.**

Patient Name *(Last, First, Middle)*	Birth Date *(mm-dd-yyyy)*

Informed Consent for Whole Exome Sequencing Plus Pharmacogenomics: Signature Page

Patient-specific preferences: If you wish to opt-out of receiving secondary findings or opt-out of participation in anonymized research studies, initial below. If either section is left blank, consent will be assumed for that preference.

Opt-Out of Secondary Findings:

- I choose to opt-out of receiving secondary findings from the 59 genes currently recommended by ACMG. Opting-out means that the laboratory will not look for variants in these genes and will not provide them in the test report unless it is a variant in a gene related to the patient's clinical features.
- Initial to Opt-Out: _____

Opt-Out of Anonymized Research Studies Using Remaining DNA:

- I choose to opt-out of participation in anonymized research studies using my DNA sample. All samples from New York clients will be disposed of 60 days after testing is complete. Opting-out means that my specimen will be destroyed upon completion of this test.
- Initial to Opt-Out: _____

Signatures: Sign below to indicate your understanding of the above information and your consent to proceed with whole exome sequencing.

Patient Signature

My signature below acknowledges my voluntary participation in this test for myself or my child.

Patient/Guardian Signature Signature Required	Date *(mm-dd-yyyy)*
Guardian Printed Name *(Last, First, Middle)*	Guardian Relationship to Patient

Parental Signatures

My signature below acknowledges my voluntary participation in this test. I understand that my sample is being provided to assist in the whole exome sequencing analysis for my child. I acknowledge that I will not receive a separate report for myself and that this testing does not include a complete assessment of my exome. I understand that this testing may reveal information about my own health.

Mother Signature Signature Required	Date *(mm-dd-yyyy)*
Mother Printed Name *(Last, First, Middle)*	Mother Birth Date *(mm-dd-yyyy)*

Father Signature Signature Required	Date *(mm-dd-yyyy)*
Father Printed Name *(Last, First, Middle)*	Father Birth Date *(mm-dd-yyyy)*

Provider/Genetic Counselor Signature

I have explained the above information regarding whole exome sequencing to this individual. I have addressed the limitations outlined above and have answered all questions to the best of my ability.

Provider/Genetic Counselor Signature Signature Required	Date *(mm-dd-yyyy)*
Provider/Genetic Counselor Printed Name *(Last, First)*	

Print