JOHNS HOPKINS
SCHOOL of MEDICINE

Johns Hopkins Genomics
DNA Diagnostic Laboratory

Clinical Exome Requisition Form - Page 1

v190409

Referrer Information

Physician: UPIN/NPI:

Genetic counselor: Email:

Institution:

Address:

Phone: Fax:

Additional reports to:

Address:

Phone: Fax:

MANDATORY SIGNATURE

I have confirmed that the patient has consented for the testing ordered and that two matching identifiers are present on each page of this requisition.

Signature:

Date:

Patient Information (*two of these identifiers MUST also appear on the sample)

Legal Name* (Last): (First):

Preferred Name (Last): (First):

Date of birth* (mm/dd/yy): Sex assigned at birth: Gender:

Patient ID/MRN*:

Billing Information (Contact Billing Coordinator at 443-287-2486 prior to submitting)

- ☐ Referring center
- ☐ Patient insurance
- ☐ Maryland Medicaid
- ☐ Self-pay
- ☐ Medicare

Billing contact:

Phone:

Fax:

Email:

Shipping Address

1812 Ashland Ave
Sample Intake Rm 245
Baltimore, MD 21205

For sample specific requirements please reference our website.

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Clinical Exome Requisition Form - Page 2

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Patient Name*: DOB*:

Patient Sample Information

Test Type

- ☐ Proband
- ☐ Trio
- ☐ Duo
- ☐ Quad

Sample Type

- ☐ Blood
- ☐ Saliva

Date collected

Submission Checklist

- ☐ Contact Billing Coordinator at 443-287-2486
- ☐ Obtain informed consent from proband and/or family members
- ☐ Complete Phenotypic Information section (Page 3)
- ☐ Sign page 1 indicating that the provider has ensured that all necessary information is complete
- ☐ If you are a Hopkins provider, enter test order in EPIC (place test order BEFORE proband blood is drawn)
- ☐ Send this requisition form, signed consent forms, and all samples to JH Genomics

Family Information

Name (Last, First)	DOB	Relationship	Sample Type	Date collected
			☐ Blood ☐ Saliva	
			☐ Blood ☐ Saliva	
			☐ Blood ☐ Saliva	

Ancestry

__ Caucasian
__ Northern European
__ Western European
__ Eastern European
__ Middle Eastern
__ African American
__ African
__ Hispanic
__ Central/South American
__ Caribbean
__ Asian
__ Pacific Islander
__ Native American
__ Other

Relevant Family Information/Pedigree

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Patient Name*: DOB*:

Phenotypic Information

Perinatal
__ IUGR
__ Oligohydramnios
__ Polyhydramnios
__ Prematurity
__

Developmental
__ Fine motor delay
__ Gross motor delay
__ Speech delay
__ Intellectual disability
__ Learning disability
__ Regression
__

Behavioral
__ Anxiety
__ Depression
__ Autism spectrum disorder
__ Autistic features
__ Obsessive-compulsive disorder
__

Craniofacial
__ Macrocephaly
__ Microcephaly
__ Craniosynostosis
__ Cleft lip/palate
__ Dental abnormalities
__ Facial dysmorphism:
__

Eyes and Vision
__ Myopia
__ Hyperopia
__ Visual Impairment
__ Blindness
__ Coloboma
__ Strabismus
__

Ears and Hearing
__ Sensorineural hearing loss
__ Conductive hearing loss
__ Mixed hearing loss
__ Preauricular pit
__ External ear malformation
__ Low-set ears
__

Brain Abnormalities
__ Agenesis of the corpus callosum
__ Holoproscencephaly
__ Lissencephaly
__ Encephalocele
__ Hydrocephalus
__ Cortical dysplasia
__ Brain atrophy
__

Neuromuscular
__ Ataxia
__ Chorea
__ Dystonia
__ Hypertonia
__ Hypotonia
__ Seizures
__

Skeletal
__ Tall stature
__ Short stature
__ Overgrowth
__ Joint hypermobility
__ Contractures
__ Polydactyly
__ Syndactyly
__ Scoliosis
__

Skin and Hair
__ Café-au-lait spots
__ Blistering
__ Ichthyosis
__ Abnormal pigmentation
__ Alopecia
__ Coarse hair
__ Brittle hair
__ Nail abnormalities
__

Cardiac
__ ASD
__ VSD
__ Tetralogy of Fallot
__ Cardiomyopathy
__ Arrhythmia
__

Gastrointestinal
__ Gastroschisis/omphalocele
__ Pyloric stenosis
__ Tracheoesophageal fistula
__ Gastroesophageal reflux
__ Hirschsprung disease
__ Constipation
__

Additional Clinical Information

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SCHOOL of MEDICINE

Johns Hopkins Genomics DNA Diagnostic Laboratory

Clinical Exome Informed Consent - Page 1

v190409

My Choices - Patient

Secondary Findings Reported to Me

Secondary findings are genetic changes that are likely to cause specific conditions other than the primary reason for testing in me/my child. Only conditions with clear management guidelines are included, and not every possible disease is covered. The DNA Diagnostic Laboratory follows the ACMG guidelines for reporting secondary findings.

- ☐ Yes, I would like secondary findings to be analyzed for me/my child.
- ☐ No, I would not like secondary findings to be analyzed for me/my child.

Research

De-identified clinical and genetic information may be used in academic case research and/or publications. The ordering provider may reach out to me to obtain additional information and/or photos. Additionally, if the results of clinical exome sequencing are negative, the ordering provider may re-contact me about follow-up research sequencing opportunities.

- ☐ Yes, I agree to my de-identified sample being used for research.
- ☐ No, I do not agree to my de-identified sample being used for research.

Raw Sequence Data Given to My Provider

My provider can have a copy of the raw sequence data from my exome test (called a variant call file or VCF). My provider may request the raw data to further analyze genetic changes that may be associated with my/my child's primary medical concerns. This information will not be used for research purposes or shared with other providers or insurers unless otherwise discussed with me.

- ☐ Yes, I allow the lab to release my raw data to my referring provider.
- ☐ No, I do not allow the lab to release my raw data to my referring provider.

Statement of Consent - Patient

My ordering provider has reviewed OR I have read the Clinical Exome Sequencing Informed Consent document in its entirety. I have had the opportunity to ask questions of the provider about Exome Sequencing. I grant permission for the DNA Diagnostic Laboratory at Johns Hopkins University to perform clinical exome sequencing for me and/or my child. I have chosen to either opt-in or opt-out of receiving secondary findings, being re-contacted for research, and allowing my referring provider to request access to the VCF as detailed above. I understand the benefits, risks, and limitations of exome sequencing

Patient name (print): _____

Signature: _____ Date: _____

Relationship to patient (if not self): _____

My Choices - Family Member

Secondary Findings Reported to Me

Secondary findings are genetic changes that are likely to cause specific conditions other than the primary reason for testing in the patient. Only conditions with clear management guidelines are included, and not every possible disease is covered. These findings will only be looked at in me if they are found to be present in my family member. The DNA Diagnostic Laboratory follows the ACMG guidelines for reporting secondary findings. I have received a copy of those guidelines.

- ☐ Yes, I would like secondary findings to be analyzed for me.
- ☐ No, I would not like secondary findings to be analyzed for me.

Research

De-identified clinical and genetic information may be used in research and/or publications. The ordering provider may reach out to me to obtain additional information and/or photos. Additionally, if the results of clinical exome sequencing are negative, the ordering provider may re-contact me about follow-up research sequencing opportunities.

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- ☐ Yes, I allow the lab to release my raw data to my referring provider.
- ☐ No, I do not allow the lab to release my raw data to my referring provider.

Statement of Consent - Family Member

My ordering provider has reviewed OR I have read the Clinical Exome Sequencing Informed Consent document in its entirety. I have had the opportunity to ask questions of the provider about Exome Sequencing. I grant permission for the DNA Diagnostic Laboratory at Johns Hopkins University to perform family member clinical exome sequencing for me and/or my child. I have chosen to either opt-in or opt-out of receiving secondary findings, being re-contacted for research, and allowing my referring provider to request access to the VCF as detailed above. I understand the benefits, risks, and limitations of exome sequencing.

Family member name (print): _____

Signature: _____ Date: _____

Relationship to patient: _____ DOB: _____

My Choices - Family Member

Secondary Findings Reported to Me

Secondary findings are genetic changes that are likely to cause specific conditions other than the primary reason for testing in the patient. Only conditions with clear management guidelines are included, and not every possible disease is covered. These findings will only be looked at in me if they are found to be present in my family member. The DNA Diagnostic Laboratory follows the ACMG guidelines for reporting secondary findings. I have received a copy of those guidelines.

- ☐ Yes, I would like secondary findings to be analyzed for me.
- ☐ No, I would not like secondary findings to be analyzed for me.

Research

De-identified clinical and genetic information may be used in research and/or publications. The ordering provider may reach out to me to obtain additional information and/or photos. Additionally, if the results of clinical exome sequencing are negative, the ordering provider may re-contact me about follow-up research sequencing opportunities.

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Family member name (print): _____

Signature: _____ Date: _____

Relationship to patient: _____ DOB: _____

Informed Consent

Exome Sequencing

Exome sequencing is a genetic test that analyzes a patient's genetic material, or DNA. Genes are the instructions that tell cells and bodies how to grow and develop. They made up of DNA. Changes in genes, or variants, may contribute to a patient's health concerns. All people have many changes in their genetic information. Only some of these variants are known to result in genetic conditions. Exome sequencing is able to analyze many genes at once to look for variants that may provide a genetic diagnosis. Understanding the cause of a patient's health concerns may provide insight into what can be expected for the patient in the future, whether other family members that may be at risk for carrying the variant, and what the risk is for recurrence. Although exome sequencing is able to read through many genes, it is not able to read through the entirety of the patient's genetic information. The goal of exome sequencing is to identify a genetic cause for a patient's health concerns.

Types of Results

There are several types of results that may be reported by exome sequencing:

POSITIVE: A positive result indicates that a genetic change has been identified in a gene known to be responsible for a genetic condition. This may or may not provide a cause or diagnosis for the patient's health concerns. It is possible that this test may identify more than one genetic change. It is possible that other family members may carry the same genetic change.

NEGATIVE: A negative result indicates that no known genetic cause for the patient's medical concerns was found. A negative result does not mean that there is not a genetic cause for the patient's health concerns. Future genetic testing may be able to identify additional genetic changes.

UNCERTAIN: A variant of uncertain significance (VUS) indicates that a genetic change was identified in a gene but that there is not yet enough information known about the consequences of a particular change or gene to determine whether it has health care significance. Testing of additional family members may be recommended to better understand the effect of an uncertain variant.

Family Member Testing

Obtaining samples from the patient's biological family members may aid in the interpretation of exome sequencing results. If a genetic change is identified in a patient, the family member samples may be tested for the same change. This may indicate whether or not the change was inherited or de novo (new to the patient). Family member samples will only be analyzed in the event that a genetic change is identified in the patient.

Secondary Findings

Exome sequencing analyzes many genes all at once, accordingly, it is possible to find genetic changes in genes that are not related the patient's primary health concerns. These results are called secondary findings. The American College of Medical Genetics and Genomics (ACMG) recommends that laboratories report such findings in genes that are known to cause specific actionable inherited conditions. Examples include hereditary cancer and heart syndromes, among others (please see attached table). Some of these conditions may not present until adulthood and may have a significant impact on the patient's and family members' healthcare and/or reproductive risk. If the patient is found to have a genetic change associated with one of these conditions, the family member samples will be analyzed for the same change. A complete list of these genes will be provided to the patient/parent/guardian. Secondary findings will only be analyzed and reported if the patient, parent, or guardian consents to receive them.

Informed Consent (continued)

Results Reporting

Results of exome sequencing will be reported to your ordering provider. Additionally, the provider may wish to get a copy of the raw sequence data, also known as the variant call file (VCF), after results are returned. Results that will be reported include positive results in the genes analyzed, variants of uncertain significance in the genes analyzed, and secondary findings if the patient/parent/guardian consents to receive them. A negative result does not rule out a disease-causing genetic change in the genes analyzed. Changes that are not believed to affect the patient's health will not be reported. Changes that are known to be risk-factors but not causative of disease may not be reported.

Risks

It is possible that this test may result in an uncertain result or identify unexpected, secondary findings. It is possible that this test may reveal unexpected familial relationships (i.e. consanguinity, non-paternity, etc.). Results of this test may affect the healthcare and/or reproductive decisions of both the patient and their family members. Results may also affect the patient's and/or family member's ability to buy life, disability, and long-term care insurance in the future. Additionally, it is possible that exome sequencing may not be covered in full by the patient's health insurance plan. Although unlikely, there is a possibility for laboratory error to occur. Genetic counseling is recommended prior to consent for exome sequencing and after results are returned.

Limitations

Although exome sequencing analyzes many genes at once, it does not analyze all genes and all types of genetic changes. It is possible that this test may not identify the genetic change responsible for the patient's medical concerns. This test may identify a change in a gene, but does not have the ability to predict long-term prognosis. Interpretation of results is based on our current understanding of genetics. It is possible that results may change in the future upon reanalysis.

Research

If the patient/parent/guardian provides permission, de-identified clinical and genetic information may be used in academic case reports and publications. The ordering provider may reach out to the patient/parent/guardian for additional information and/or photos. Additionally, if the results of clinical exome sequencing are negative, the ordering provider may re-contact the patient/parent/guardian regarding follow-up research sequencing opportunities.

Privacy Protections

The results of clinical exome sequencing will be released only to providers authorized by the patient/parent/guardian. In addition, The Genetic Information Nondiscrimination Act (GINA) protects most individuals from discrimination by employers and/or health insurers on the basis of genetic test results. In an attempt to better understand the field of genetics and variant interpretation, the DNA Diagnostic Laboratory may share de-identified genetic information in healthcare databases.