



WHOLE EXOME SEQUENCING (WES) REFLEX FORM

Please complete every field and tick box clearly.

PATIENT INFORMATION

<input type="text"/>	<input type="text"/>	<input type="text" value="MM/DD/YYYY"/>
Patient's First Name	Middle Initial	Patient's Date of Birth

<input type="text"/>	<input type="text"/>
Patient's Last Name	Patient ID/MR Number

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

Patient's Street Address

<input type="text"/>	<input type="text"/>	<input type="text"/>
City / Town	State	Zip Code

<input type="text"/>	<input type="text"/>
Country	Patient's Preferred Phone

Patient's Email

Ethnicity (check all that apply):

<input type="radio"/> African-American	<input type="radio"/> Asian (China, Japan, Korea)
<input type="radio"/> Caucasian/N. European/S. European	<input type="radio"/> Finnish
<input type="radio"/> Hispanic	<input type="radio"/> French Canadian
<input type="radio"/> Jewish - Ashkenazi	<input type="radio"/> Jewish - Sephardic
<input type="radio"/> Mediterranean	<input type="radio"/> Middle Eastern (Saudi Arabia, Qatar, Iraq, Turkey)
<input type="radio"/> Native American	<input type="radio"/> OE. Indian
<input type="radio"/> Southeast Asian (Vietnam, Cambodia, Thailand)	<input type="radio"/> South Asian (India, Pakistan)
<input type="radio"/> Other (specify) <input type="text"/>	

ORDERING PROVIDER

Provider's First and Last Name

<input type="text"/>	<input type="text"/>
PKIG Ordering Provider Account Number	NPI

Clinic/Hospital/Institution Name

<input type="text"/>	<input type="text"/>
Provider's Email	Provider's Phone

Provider's Street Address

<input type="text"/>	<input type="text"/>	<input type="text"/>
City / Town	State	Zip Code

<input type="text"/>	<input type="text"/>
Country	Provider's Fax

SEND ADDITIONAL COPY OF RESULTS TO (If applicable)

Name

<input type="text"/>	<input type="text"/>
PKIG Ordering Provider Account Number	Phone Number

<input type="text"/>	<input type="text"/>
Email Address	Fax Number

PHYSICIAN CONFIRMATION OF INFORMED CONSENT AND MEDICAL NECESSITY

The undersigned person (or representative thereof) ensures he/she is a licensed medical professional authorized to order genetic testing and confirms that the patient has given appropriate informed consent for the testing ordered, including a discussion of the benefits and limitations. I confirm that testing is medically necessary and that test results may impact medical management for the patient. Furthermore, all information on this TRF is true to the best of my knowledge. My signature applies to the informed consent and/or attached letter of medical necessity.

Signature _____ Date _____

PATIENT SAMPLE INFORMATION

SAMPLE TYPE: Saliva Swab Whole Blood Dried Blood Spots Other _____

Collection Date:

Was this sample collected in NY State: yes no

INDICATION FOR TESTING (Required)

ICD10 Code(s): _____
 Clinical Diagnosis: _____
 Age at Initial Presentation: _____

TEST MENU

D9999 Reflex to proband WES (from previous comprehensive curated panel of 99+ genes or PanelPlus or AnyPanel™ test)

Information from Previous Testing (REQUIRED)

I certify that this patient previously had comprehensive curated panel (99+ genes or PanelPlus) testing or AnyPanel™ test at PerkinElmer Genomics

<input type="text"/>	<input type="text" value="MM/DD/YYYY"/>
Original Accession ID Number†	Report Date†

! † Can be found at the top of the first page of original PerkinElmer Genomics test report.

WES

To order whole exome sequencing after a smaller multigene panel, please submit a new sample and select one of the following:

- D1000 Whole Exome Sequencing Proband Only
- D1300 Whole Exome Sequencing TRIO
- D1301 Whole Exome Sequencing TRIO with Parental Reports
- D1010 STAT Whole Exome Sequencing Proband Only
- D1310 STAT Whole Exome Sequencing TRIO
- D1311 STAT Whole Exome Sequencing TRIO with Parental Reports

BILLING INFORMATION

Institutional Billing

Institution/Organization Name

PerkinElmer Genomics Billing Account ID

<input type="text"/>	<input type="text"/>
Contact Name	Contact Phone

Patient Billing

- Check:** \$ _____ Amount Enclosed (Please make checks payable to: PerkinElmer Genetics, Inc.)
- Credit Card** (Please fill out all information)

<input type="text"/>	<input type="text"/>	<input type="text" value="MM/YY"/>
Credit Card Number	CVV	Card Exp. Date

Cardholder Printed Name as Appears on Card

Credit Card Billing Street Address

City/Town

<input type="text"/>	<input type="text"/>	<input type="text"/>
State	Zip Code	Cardholder Phone

Cardholder Signature

FOR INTERNAL USE ONLY					
Date Rec'd	Rec'd				
TEMP	SPEC	COL	#TUBES	VOL	
R/C/F					
R/C/F					
R/C/F					

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

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

Clinical diagnosis: _____

Age of manifestation: _____ ICD-10 Codes: _____

<p>A. NEUROLOGY</p> <p>1. Behavioral abnormality</p> <p><input type="checkbox"/> 1.1 Autism</p> <p><input type="checkbox"/> 1.2 Attention deficit disorder</p> <p><input type="checkbox"/> 1.3 Psychiatric diseases</p> <p>2. Brain imaging</p> <p><input type="checkbox"/> 2.1 Abnormal myelination</p> <p><input type="checkbox"/> 2.2 Abnormal cortical gyration</p> <p><input type="checkbox"/> 2.3 Agenesis of corpus callosum</p> <p><input type="checkbox"/> 2.4 Brain atrophy</p> <p><input type="checkbox"/> 2.5 Cerebellar hypoplasia</p> <p><input type="checkbox"/> 2.6 Heterotopia</p> <p><input type="checkbox"/> 2.7 Holoprosencephaly</p> <p><input type="checkbox"/> 2.8 Hydrocephalus</p> <p><input type="checkbox"/> 2.9 Leukodystrophy</p> <p><input type="checkbox"/> 2.10 Lissencephaly</p> <p>3. Developmental delay</p> <p><input type="checkbox"/> 3.1 Delayed motor development</p> <p><input type="checkbox"/> 3.2 Delayed language development</p> <p><input type="checkbox"/> 3.3 Developmental regression</p> <p><input type="checkbox"/> 3.4 Intellectual disability</p> <p>4. Movement abnormality</p> <p><input type="checkbox"/> 4.1 Ataxia</p> <p><input type="checkbox"/> 4.2 Chorea</p> <p><input type="checkbox"/> 4.3 Dystonia</p> <p><input type="checkbox"/> 4.4 Parkinsonism</p> <p>5. Neuromuscular abnormality</p> <p><input type="checkbox"/> 5.1 Muscular hypotonia</p> <p><input type="checkbox"/> 5.2 Muscular hypertonia</p> <p><input type="checkbox"/> 5.3 Hyperreflexia</p> <p><input type="checkbox"/> 5.4 Spasticity</p> <p>6. Seizures</p> <p><input type="checkbox"/> 6.1 Febrile seizures</p> <p><input type="checkbox"/> 6.2 Focal seizures</p> <p><input type="checkbox"/> 6.3 Generalized seizures</p> <p>7. Others</p> <p><input type="checkbox"/> 7.1 Craniosynostosis</p> <p><input type="checkbox"/> 7.2 Dementia</p> <p><input type="checkbox"/> 7.3 Encephalopathy</p> <p><input type="checkbox"/> 7.4 Headache / Migraine</p> <p><input type="checkbox"/> 7.5 Macrocephaly</p> <p><input type="checkbox"/> 7.6 Microcephaly</p> <p><input type="checkbox"/> 7.7 Neuropathy</p> <p><input type="checkbox"/> 7.8 Stroke</p>	<p>B. METABOLISM</p> <p><input type="checkbox"/> 1. Abnormal creatine kinase</p> <p><input type="checkbox"/> 2. Decreased plasma carnitine</p> <p><input type="checkbox"/> 3. Hyperalaninemia</p> <p><input type="checkbox"/> 4. Hypoglycemia</p> <p><input type="checkbox"/> 5. Increased CSF lactate</p> <p><input type="checkbox"/> 6. Increased serum pyruvate</p> <p><input type="checkbox"/> 7. Ketosis</p> <p><input type="checkbox"/> 8. Lactic acidosis</p> <p><input type="checkbox"/> 9. Organic aciduria</p> <p>C. EYE</p> <p><input type="checkbox"/> 1. Blepharospasm</p> <p><input type="checkbox"/> 2. Cataract</p> <p><input type="checkbox"/> 3. Coloboma</p> <p><input type="checkbox"/> 4. Glaucoma</p> <p><input type="checkbox"/> 5. Microphthalmos</p> <p><input type="checkbox"/> 6. Nystagmus</p> <p><input type="checkbox"/> 7. Ophthalmoplegia</p> <p><input type="checkbox"/> 8. Optic atrophy</p> <p><input type="checkbox"/> 9. Ptosis</p> <p><input type="checkbox"/> 10. Retinitis pigmentosa</p> <p><input type="checkbox"/> 11. Retinoblastoma</p> <p><input type="checkbox"/> 12. Strabismus</p> <p><input type="checkbox"/> 13. Visual impairment</p> <p>D. MOUTH, THROAT AND EAR</p> <p><input type="checkbox"/> 1. Abnormality of dental color</p> <p><input type="checkbox"/> 2. Cleft lip / palate</p> <p><input type="checkbox"/> 3. Conductive hearing impair.</p> <p><input type="checkbox"/> 4. External ear malformation</p> <p><input type="checkbox"/> 5. Hypodontia</p> <p><input type="checkbox"/> 6. Sensorineural hearing impair.</p> <p>E. SKIN, INTEGUMENT AND SKELETAL</p> <p>1. Skeletal</p> <p><input type="checkbox"/> 1.1 Abnormal limb morphology</p> <p><input type="checkbox"/> 1.2 Abnormal skeletal system</p> <p><input type="checkbox"/> 1.3 Abnormal vertebral column</p> <p><input type="checkbox"/> 1.4 Joint hypermobility</p> <p><input type="checkbox"/> 1.5 Multiple joint contractures</p> <p><input type="checkbox"/> 1.6 Polydactyly</p> <p><input type="checkbox"/> 1.7 Scoliosis</p> <p><input type="checkbox"/> 1.8 Syndactyly</p> <p><input type="checkbox"/> 1.9 Talipes equinovarus</p>	<p>2. Skin and integument</p> <p><input type="checkbox"/> 2.1 Abnormal skin pigmentation</p> <p><input type="checkbox"/> 2.2 Abnormal hair</p> <p><input type="checkbox"/> 2.3 Abnormal nail</p> <p><input type="checkbox"/> 2.4 Hyperextensible skin</p> <p><input type="checkbox"/> 2.5 Ichthyosis</p> <p>F. CARDIOVASCULAR</p> <p><input type="checkbox"/> 1. Angioedema</p> <p><input type="checkbox"/> 2. Aortic dilatation</p> <p><input type="checkbox"/> 3. Arrhythmia</p> <p><input type="checkbox"/> 4. Coarctation of aorta</p> <p><input type="checkbox"/> 5. Defect of atrial septum</p> <p><input type="checkbox"/> 6. Defect of ventricular septum</p> <p><input type="checkbox"/> 7. Dilated cardiomyopathy</p> <p><input type="checkbox"/> 8. Hypertension</p> <p><input type="checkbox"/> 9. Hypertrophic cardiomyopathy</p> <p><input type="checkbox"/> 10. Hypotension</p> <p><input type="checkbox"/> 11. Lymphedema</p> <p><input type="checkbox"/> 12. Malf. of heart and great vessels</p> <p><input type="checkbox"/> 13. Myocardial infarction</p> <p><input type="checkbox"/> 14. Stroke</p> <p><input type="checkbox"/> 15. Tetralogy of Fallot</p> <p><input type="checkbox"/> 16. Vasculitis</p> <p>G. GASTROINTESTINAL, GENITOURINARY, ENDOCRINE</p> <p>1. Gastrointestinal</p> <p><input type="checkbox"/> 1.1 Aganglionic megacolon</p> <p><input type="checkbox"/> 1.2 Constipation</p> <p><input type="checkbox"/> 1.3 Diarrhea</p> <p><input type="checkbox"/> 1.4 High hepatic transaminases</p> <p><input type="checkbox"/> 1.5 Gastroschisis</p> <p><input type="checkbox"/> 1.6 Hepatic failure</p> <p><input type="checkbox"/> 1.7 Hepatomegaly</p> <p><input type="checkbox"/> 1.8 Obesity</p> <p><input type="checkbox"/> 1.9 Pyloric stenosis</p> <p><input type="checkbox"/> 1.10 Vomiting</p> <p>2. Genitourinary</p> <p><input type="checkbox"/> 2.1 Abnormal renal morphology</p> <p><input type="checkbox"/> 2.2 Abnormal urinary system</p> <p><input type="checkbox"/> 2.3 Hydronephrosis</p> <p><input type="checkbox"/> 2.4 Renal agenesis</p> <p><input type="checkbox"/> 2.5 Renal cyst</p> <p><input type="checkbox"/> 2.6 Renal tubular dysfunction</p>	<p>3. Endocrine</p> <p><input type="checkbox"/> 3.1 Diabetes mellitus</p> <p><input type="checkbox"/> 3.2 Hypo / hyperparathyroidism</p> <p><input type="checkbox"/> 3.3 Hypo / hyperthyroidism</p> <p>H. REPRODUCTION</p> <p><input type="checkbox"/> 1. Abnormal external genitalia</p> <p><input type="checkbox"/> 2. Abnormal internal genitalia</p> <p><input type="checkbox"/> 3. Hypogonadism</p> <p><input type="checkbox"/> 4. Hypospadias</p> <p><input type="checkbox"/> 5. Infertility</p> <p>I. ONCOLOGY</p> <p><input type="checkbox"/> 1. Adenomatous polyposis</p> <p><input type="checkbox"/> 2. Breast carcinoma</p> <p><input type="checkbox"/> 3. Colorectal carcinoma</p> <p><input type="checkbox"/> 4. Leukemia</p> <p><input type="checkbox"/> 5. Myelofibrosis</p> <p><input type="checkbox"/> 6. Neoplasm of the lung</p> <p><input type="checkbox"/> 7. Neoplasm of the skin</p> <p><input type="checkbox"/> 8. Paraganglioma</p> <p><input type="checkbox"/> 9. Pheochromocytoma</p> <p>J. HEMATOLOGY AND IMMUNOLOGY</p> <p><input type="checkbox"/> 1. Abnormality of coagulation</p> <p><input type="checkbox"/> 2. Anemia</p> <p><input type="checkbox"/> 3. Immunodeficiency</p> <p><input type="checkbox"/> 4. Neutropenia</p> <p><input type="checkbox"/> 5. Pancytopenia</p> <p><input type="checkbox"/> 6. Abnormal hemoglobin</p> <p><input type="checkbox"/> 7. Splenomegaly</p> <p><input type="checkbox"/> 8. Thrombocytopenia</p> <p>K. PRENATAL AND DEVELOPMENT</p> <p><input type="checkbox"/> 1. Dysmorphic facial features</p> <p><input type="checkbox"/> 2. Failure to thrive</p> <p><input type="checkbox"/> 3. Hemihypertrophy</p> <p><input type="checkbox"/> 4. Hydrops fetalis</p> <p><input type="checkbox"/> 5. IUGR</p> <p><input type="checkbox"/> 6. Oligohydramnios</p> <p><input type="checkbox"/> 7. Overgrowth</p> <p><input type="checkbox"/> 8. Polyhydramnios</p> <p><input type="checkbox"/> 9. Premature birth</p> <p><input type="checkbox"/> 10. Short stature</p> <p><input type="checkbox"/> 11. Tall stature²</p>
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OTHER: _____