



# **Curated Multigene Panel TRF**

Please complete every field and tick box clearly.

PATIENT INFORMATION			PATIENT SAMPLE INFORMAT	ION	
		MM/ DD /YYYY	SAMPLE TYPE:	Collection Date: MM/DD/YY	
Patient's First Name	Middle Initial	Patient's Date of Birth	│ ○ Saliva Swab ○ Whole Blood	Was this sample collected in NY State: $\bigcirc$ yes $\bigcirc$ no	
- duonto i not ramo		T dione o bate of birtin	O Dried Blood Spots		
	] [		Other		
Patient's Last Name	Patient ID/MR	Number	INDICATION FOR TESTING (R	equired)	
Biological Sex: OMale OFemale OUnknown	own		ICD10 Code(s):	•	
Gender Identity (if different from above):			Clinical Diagnosis:		
			Age at Initial Presentation:		
Patient's Street Address			TEST MENU		
Patient's Street Address			Ocurated panel by clinical indication		
				disease-specific next-generation sequencing nical indications include neuromuscular, neurology,	
City / Town State Zip Code		cardiology, hereditary cancer,	and other categories such as hearing loss and		
				ring portal to select the correct panel at https://client.	
Country Patient's Pre	ferred Phone		perkinelmergenomics.com/log	<u>יווני</u>	
			TO ORDER ON PAPER:  Provide test code here:		
Patient's Email			Provide test name here:		
Ethnicity (check all that apply):   African-	American 04	Asian (China, Japan, Korea)			
			OD3000 AnyPanel™ Test	(with reflex options). Gene total may range from 2 to	
OCaucasian/N. European/S. European	innish	OFrench Canadian		exome sequencing is more efficient. Please submit	
	lewish - Sephardi	c OMediterranean	and include custom panel ID		
OMiddle Eastern (Saudi Arabia, Qatar, Iraq, Turkey)	lative American	OE. Indian	PROVIDE CUSTOM PANEL	ID HERE:	
OSoutheast Asian (Vietnam, Cambodia, Thailand	South	Asian (India, Pakistan)	○TRIO Panels*		
Other (specify)				I with TRIO capabilities, please select the correct box rated panels with TRIO options.	
			OTR001 Epilepsy TRIO Pane		
ORDERING PROVIDER			○TR002 Autism and Intellect	tual Disability TRIO Panel	
			<ul> <li>TR003 Neuromuscular TRI</li> <li>TR004 Nuclear Mitochondr</li> </ul>		
Provider's First and Last Name					
			* Please fill out family member received within 3 weeks.	section below. Family member samples MUST be	
PKIG Ordering Provider Account Number	NPI		FAMILIAL INFORMATION (Req	uired for TRIO panels)	
			FAMILY MEMBER 1:		
Clinic/Hospital/Institution Name			1 AMIEI WEWDER 1.		
			Last name, First name		
Provider's Email	Provider's P	hone	Last Hame, I list Hame		
			Relationship to Patient		
Provider's Street Address			Date of Birth: MM/DD/YYYY		
			Symptomatic (clinically affecte		
City / Town State		Code	Sample: OIncluded - Collect		
			l	3 10 30 00 Milator	
			FAMILY MEMBER 2:		
Country Provider's F					
SEND ADDITIONAL COPY OF RESULTS T	O (If applicable)		Last name, First name		
Name			Relationship to Patient		
			Date of Birth: MM/DD/YYYY	Y	
PKIG Ordering Provider Account Number	Phone Num	her	Symptomatic (clinically affecte	d)? Oyes Ono	
The ordering Frovider Account Number		DCI	Sample: OIncluded - Collect	ction Date MM/DD/YY O To be sent later	
Email Address	Fax Number				
PHYSICIAN CONFIRMATION OF INFORMED					
				ing and confirms that the patient has given appropriate sary and that test results may impact medical management	
for the patient. Furthermore, all information on th					
		•			
Signature		Date			
				FOR INTERNAL USE ONLY	





# **Curated Multigene Panel TRF**

■ INSTITUTIONAL BILLING					
Institution/Organization Name			PerkinElmer Genomics Billing Account ID		
Contact Name			Contact Phone		
■ PATIENT (SELF) PAYMENT					
By providing payment information, you are authorizing PerkinElmer is required prior to test initiation. The patient's sample will be placetest order may be canceled. Please note that failure by the patient	d on hold (for up to 30 days) until pay	yment is	secured. If the patient does not provide payment to Perkin	nElmer within 30	0 days, the
O CHECK: \$ Amount Enclosed (Please make	e checks payable to: PerkinElm	er Gen	etics, Inc.)		
O CREDIT CARD (Please fill out all information):					
	MM/YY				
Credit Card Number	Card Exp. Date CVV	Ca	dholder Printed Name as Appears on Card	Amount	
Credit Card Billing Street Address	City	/ Town		State	Zip Code
Cardholder Signature			Cardholder Phone		
O CONTACT PATIENT FOR PAYMENT INFORMATION					
Mobile Phone			Home Phone		
Email Address					





## **Curated Multigene Panel TRF**

Detailed medical records, clinical summary, pictures, and family history must be attached for all cases if you would like a phenotypically driven report. Such information is crucial for the accurate interpretation of results.

ADDITIONAL OPTIONAL PHENOTYPE / PATIENT HISTORY SECTION (Check all that apply) Clinical diagnosis: ICD-10 Codes: Age of manifestation: A. NEUROLOGY **B. METABOLISM** 2. Skin and integument 3. Endocrine 1. Behavioral abnormality O 1. Abnormal creatine kinase O 3.1 Diabetes mellitus O 2.1 Abnormal skin pigmentation O 1.1 Autism O 2. Decreased plasma carnitine O 2.2 Abnormal hair O 3.2 Hypo / hyperparathyroidism O 1.2 Attention deficit disorder O 3. Hyperalaninemia O 2.3 Abnormal nail O 3.3 Hypo / hyperthyroidism O 1.3 Psychiatric diseases O 4. Hypoglycemia O 2.4 Hyperextensible skin H. REPRODUCTION O 5. Increased CSF lactate O 2.5 Ichthyosis O 1. Abnormal external genitalia 2. Brain imaging O 2.1 Abnormal myelination O 6. Increased serum pyruvate F. CARDIOVASCULAR O 2. Abnormal internal genitalia O 2.2 Abnormal cortical gyration O 7. Ketosis O 1. Angioedema O 3. Hypogonadism O 2.3 Agenesis of corpus callosum O 8. Lactic acidosis O 2 Aortic dilatation O 4. Hypospadias O 3. Arrhythmia O 2.4 Brain atrophy O 9. Organic aciduria O 5. Infertility O 2.5 Cerebellar hypoplasia C. EYE O 4. Coarctation of aorta I. ONCOLOGY O 1. Blepharospasm O 5. Defect of atrial septum O 2.6 Heterotopia O 1. Adenomatous polyposis O 2.7 Holoprosencephaly O 2. Cataract O 6. Defect of ventricular septum O 2. Breast carcinoma O 2.8 Hydrocephalus O 3. Coloboma O 7. Dilated Cardiomyopathy O 3. Colorectal carcinoma O 2.9 Leukodystrophy O 4. Glaucoma O 8. Hypertension O 4. Leukemia O 2.10 Lissencephaly O 5. Microphthalmos O 9. Hypertrophic Cardiomyopathy O 5. Myelofibrosis 3. Developmental delay O 6. Nystagmus O 10. Hypotension O 6. Neoplasm of the lung O 3.1 Delayed motor development O 7. Ophthalmoplegia O 11. Lymphedema O 7. Neoplasm of the skin O 8. Optic atrophy O 12. Malf. of heart and great vessels O 3.2 Delayed language development O 8. Paraganglioma O 3.3 Developmental regression O 9 Ptosis O 13. Myocardial infarction O 9. Pheochromocytoma O 3.4 Intellectual disability O 14. Stroke J. HEMATOLOGY AND IMMUNOLOGY O 10. Retinitis pigmentosa O 15. Tetralogy of Fallot O 11. Retinoblastoma 4. Movement abnormality O 1. Abnormality of coagulation O 4 1 Ataxia O 12 Strabismus O 16. Vasculitis O 2. Anemia O 4.2 Chorea O 13. Visual impairment O 3. Immunodeficiency G. GASTROINTESTINAL. GENITOURINARY, ENDOCRINE O 4.3 Dystonia D. MOUTH, THROAT AND EAR O 4 Neutropenia O 4 4 Parkinsonism O 1. Abnormality of dental color 1. Gastrointestinal O 5. Pancytopenia O 2. Cleft lip / palate 5. Neuromuscular abnormality O 1.1 Aganglionic megacolon O 6. Abnormal hemoglobin O 1.2 Constipation O 5.1 Muscular hypotonia O 3. Conductive hearing impair. O 7. Splenomegaly O 5.2 Muscular hypertonia O 4. External ear malformation O 1.3 Diarrhea O 8. Thrombocytopenia O 1.4 High hepatic transaminases K. PRENATAL AND DEVELOPMENT O 5.3 Hyperreflexia O 5. Hypodontia O 5.4 Spasticity O 6. Sensoneural hearing impair. O 1.5 Gastroschisis O 1. Dysmorphic facial features O 1.6 Hepatic failure 6. Seizures O 2. Failure to thrive E. SKIN. INTEGUMENT AND SKELETAL O 6.1 Febrile seizures O 1.7 Hepatomegaly O 3. Hemihypertrophy 1. Skeletal O 6.2 Focal seizures O 1.8 Obesity O 4. Hydrops fetalis O 1.1 Abnormal limb morphology O 1.9 Pyloric stenosis O 6.3 Generalized seizures O 5. IUGR O 1.2 Abnormal skeletal system O 1.10 Vomiting 7. Others O 6. Oligohydramnios O 1.3 Abnormal vertebral column 2. Genitourinary O 7.1 Craniosynostosis O 7. Overgrowth O 1.4 Joint hypermobility O 2.1 Abnormal renal morphology O 72 Dementia O 8. Polyhydramnios O 1.5 Multiple joint contractures O 7.3 Encephalopathy O 2.2 Abnormal urinary system O 9 Premature birth O 1.6 Polydactyly O 7.4 Headache / Migraine O 2.3 Hydronephrosis O 10. Short stature O 1.7 Scoliosis O 7.5 Macrocephaly O 2.4 Renal agenesis O 11. Tall stature O 7.6 Microcephaly O 1.8 Syndactyly O 2.5 Renal cyst O 1.9 Talipes equinovarus O 7.7 Neuropathy O 2.6 Renal tubular dysfunction

O 7.8 Stroke





### U.S. CLINICAL INFORMED CONSENT FORM

PerkinElmer Genetics, Inc., ("PerkinElmer") requires a completed Patient's Informed Consent Form (ICF) for testing to be performed. The ICF must be completed by the patient, or a legally authorized representative of the patient (or by the healthcare provider where permitted under applicable law or regulation). For any patient below the age of majority, the ICF must be completed by the patient's legally authorized representative.

The purpose of this 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. Given the complexity of the type of the Test, it is recommended that you and/or your child receive genetic counseling by a trained genetics professional before and after the testing is performed.

#### **TEST INFORMATION**

Your healthcare provider ("HCP") has recommended that you, or your child, receive enzymatic, biochemical or molecular genetics 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 having you tested for, please consult with your HCP. You are free to decide if you want this Test performed or not. Providing a Sample and undergoing the Test is voluntary and you may withdraw your consent without penalty at any time.

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 child is carrying which is causing the specific disease or condition 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. Only the genes identified on the Requisition will be analyzed. In some cases, we may not be able to determine with certainty which gene is actually causing the disease.

#### **TEST METHOD**

If you consent to the Test, your HCP will take a sample of your and/or your child's blood, saliva, body fluid, tissue or other sample type. Your Sample will be sent to PerkinElmer'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; PerkinElmer will not provide a diagnosis. PerkinElmer will report Test results only to your HCP 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 child 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. A positive genetic test 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.
- 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: In rare instances, 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

Reported disease-causing variants are described as pathogenic variant(s), likely pathogenic variants(s), or variant(s) of uncertain significance in genes interpreted to be responsible for, or potentially contributing to, a disease or condition. In addition, variants in genes not known to be associated with disease but for which there is evidence to suggest an association with disease may also be reported. 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 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, and available reporting options. For prenatal samples, secondary findings for the proband are not available.

## 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 WES/WGS test results. These Tests could be part of a TRIO Test or as stand-alone targeted testing. PerkinElmer, in consultation with the HCP, will decide if other family members need to be tested. If the HCP recommends testing for additional family members, only the Test performed will be reported. If undergoing a TRIO WES or WGS test, family members will 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 selected (for an additional charge). If family members elect to receive information about secondary findings either as part of the proband report or as a standalone report, the family member must sign all applicable sections on page 3 and/or 4 of this form.

#### **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 that is uninterpretable or of unknown significance may require further testing 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.





### U.S. CLINICAL INFORMED CONSENT FORM

### CONFIDENTIALITY

You have the right to confidential treatment of the Sample and your PHI. Your HCP will provide PerkinElmer 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 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, PerkinElmer 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 PerkinElmer, or you can contact PerkinElmer directly by visiting www.perkinelmergenomics.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 PerkinElmer 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 PerkinElmer indefinitely, unless otherwise noted. In some instances, it may be beneficial to you for PerkinElmer to retain your sample for a longer period of time in order to conduct additional testing, and PerkinElmer will do so with appropriate documentation from you or your HCP.

PerkinElmer is requesting consent to keep you and/or your child's anonymized sample and data indefinitely for ongoing test development, scientific research, and/or other activities. This consent is optional, and the Test will be performed whether or not you provide consent to the following:

- · PerkinElmer will anonymize and retain your Sample indefinitely for internal quality control, test validation, assay development and improvement. By allowing PerkinElmer 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 PerkinElmer Genetics, Inc. If you withdraw your consent to use of your anonymized sample, no further anonymization will be performed.
  - 🖵 Check here if you would like to opt out of anonymized sample retention (NY State residents, please see section below). Note, if not checked, this is interpreted as "consent given"
- · PerkinElmer will anonymize your data and retain the anonymized data and related anonymized reports from your Tests indefinitely for statistical and quality analysis, research, scientific and technical development, and market research. PerkinElmer may also share your anonymized data and anonymized report with third parties.
  - 🖵 Check here if you would like to opt out of anonymized data retention. Note, if not checked, this is interpreted as "consent given"

REQUIRED FOR SAMPLES	S COLLECTED IN NEW YORK STATE OF	NLY
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No tests other than those authorized shall be performed on the biological sample submitted for testing, and any material derived from the sample (i.e., DNA); this includes testing for internal research and/or quality control purposes. The sample shall be destroyed no more than 60 days after the sample was taken or at the end of the testing process, whichever occurs later, unless indicated below.

By checking here and signing at right, I consent to PerkinElmer keeping my sample for longer than 60 days, and to using my de-identified	
sample for internal research and/or quality control purposes. Note, if not checked and signed, this is interpreted as "consent not given."	Patient/Guardian Signature

#### **RESEARCH OPTIONS**

PerkinElmer 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, PerkinElmer may contact you or your HCP about the development of new testing, drug development, or other treatments. PerkinElmer 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. By checking here I would like to opt out of PerkinElmer being able to provide my contact information to outside researchers to contact me directly about

applicable research studies.

#### 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 PerkinElmer will not perform the Test unless I provide consent to the Test. 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 PerkinElmer via email at: Genomics@perkinelmer.com or toll-free

by telephone +1-866-354-2910 to request withdrawal.				
PATIENT CONSENT TO TESTING				
☐ By checking this box I attest:				
I have read and understood the Informed Consent Form in its entirety, including the explanation of why my sample is being tested, how genetic testing is performed and the risks associated with genetic testing. I have had the opportunity to ask my HCP questions about the information contained herein, and understand that I am entitled to a copy of this ICF. My signature below acknowledges my free consent to the Test, and to any additional consents indicated above, and such testing in no way guarantees my health, the health of an unborn child, or the health of other family members.				
Patient Signature (or Parent/Guardian if patient is minor)	Dat	Date		
Patient Name	Nar	Name and Relationship (Parent/Guardian if patient is minor)		
FAMILY MEMBER CONSENT TO TESTING (if applicable)				
genetic testing is performed and the risks associated with genetic te	sting. I have had the opportubelow acknowledges my free	s entirety, including the explanation of why my sample is being tested, how brunity to ask my HCP questions about the information contained herein, free consent to the Test, and to any additional consents indicated above, of other family members.		
Family Member Signature	Date Family Member Na	Name Relationship to Patient		
FAMILY MEMBER CONSENT TO TESTING (if applicable)				

By checking this box I attest: I have read and understood the Informed Consent Form in its entirety, including the explanation of why my sample is being tested, how genetic testing is performed and the risks associated with genetic testing. I have had the opportunity to ask my HCP questions about the information contained herein, and understand that I am entitled to a copy of this ICF. My signature below acknowledges my free consent to the Test, and to any additional consents indicated above, and such testing in no way guarantees my health, the health of an unborn child, or the health of other family members.

Date Family Member Name Family Member Signature Relationship to Patient