

## FSHD Requisition Form

Please complete every field and tick box clearly.

### PATIENT INFORMATION

Patient's First Name Middle Initial Patient's Date of Birth

Patient's Last Name Patient ID/MR 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)

### ORDERING PROVIDER

Provider's First and Last Name

PKIG Ordering Provider Account Number NPI

Clinic/Hospital/Institution Name

Provider's Email Provider's Phone

Provider's Street Address

City / Town State Zip Code

Country Provider's Fax

### SEND ADDITIONAL COPY OF RESULTS TO (If applicable)

Name

PKIG Ordering Provider Account Number Phone Number

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:**  Whole Blood  Dried Blood Spots  Saliva  Other: \_\_\_\_\_  
 Collection Date: \_\_\_\_\_  
 Was this sample collected in NY State:  Yes  No

### INDICATION FOR TESTING (Required)

ICD 10 Code(s): \_\_\_\_\_  
 Clinical Diagnosis: \_\_\_\_\_  
 Age at Initial Presentation: \_\_\_\_\_

### TEST MENU

Test Code	Test Name	Sample Type
<input type="radio"/> D8000	FSHD Type 1 Testing (4q D4Z4 repeat size)	WB
<input type="radio"/> D5132	FSHD Type 2 Testing (SMCHD1 gene)	DBS, Saliva, WB, DNA
<input type="radio"/> D4035	Comprehensive Neuromuscular Panel (does not include FSHD Type 1)	DBS, Saliva, WB, DNA

### IMPORTANT SHIPPING AND HANDLING INSTRUCTIONS

For any order that includes FSHD1 testing, please follow the shipping and handling instructions below to ensure specimens are viable for FSHD1 analysis.

- All samples should be shipped to the lab the same day of draw.
  - Due to the time-sensitive nature of this test, **the sample must arrive in the lab within four days of collection.**
- Please include a completed requisition form marked with appropriate FSHD order to avoid delays in processing. **Date of Collection is a REQUIRED field.**
- The sample should be shipped at refrigeration temperature and include an ice pack within the box. **Do not freeze the specimen.**
  - Please note that shipping conditions can dramatically affect the temperature of sample while in transit.
- If using a PerkinElmer Genomics collection pack, please package the specimen in the provided box and activate the included coolant pack to help regulate temperature during transit.

#### FOR INTERNAL USE ONLY

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

## ■ INSTITUTIONAL BILLING

<input type="text"/>	<input type="text"/>
Institution/Organization Name	PerkinElmer Genomics Billing Account ID
<input type="text"/>	<input type="text"/>
Contact Name	Contact Phone

## ■ PATIENT (SELF) PAYMENT

By providing payment information, you are authorizing PerkinElmer to process payment at the associated charge for tests ordered. Test cost 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 PerkinElmer within 30 days, the test order may be canceled. Please note that failure by the patient to respond in a timely fashion to PerkinElmer's attempts to obtain payment may cause a delay in the receipt of the results report.

**CHECK:** \$\_\_\_\_\_ Amount Enclosed (Please make checks payable to: PerkinElmer Genetics, Inc.)

**CREDIT CARD** (Please fill out all information):

<input type="text"/>	<input type="text" value="MM/YY"/>	<input type="text"/>	<input type="text"/>	<input type="text"/>
Credit Card Number	Card Exp. Date	CVV	Cardholder Printed Name as Appears on Card	Amount
<input type="text"/>	<input type="text"/>	<input type="text"/>	<input type="text"/>	<input type="text"/>
Credit Card Billing Street Address	City / Town	State	Zip Code	
<input type="text"/>	<input type="text"/>	<input type="text"/>	<input type="text"/>	
Cardholder Signature	Cardholder Phone			

**CONTACT PATIENT FOR PAYMENT INFORMATION**

<input type="text"/>	<input type="text"/>
Mobile Phone	Home Phone
<input type="text"/>	
Email Address	

**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)**

Face2Gene ID (if applicable): \_\_\_\_\_

Clinical diagnosis: \_\_\_\_\_

Age of manifestation: \_\_\_\_\_ ICD-10 Codes: \_\_\_\_\_

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

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:

- 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.
- 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.
- 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.
- 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 variant(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.

