Decode Duchenne Requisition Form

Free genetic testing and counseling support for Duchenne and Becker muscular dystrophy

This test requisition form can be used to submit a specimen to the Decode Duchenne diagnostic program for patients in the US and Canada. This program is provided through a partnership with Parent Project Muscular Dystrophy (PPMD) with support from Sarepta Therapeutics, PTC Therapeutics, Vertex Pharmaceuticals and NS Pharma. Please confirm the patient meets the required INDICATIONS FOR TESTING (below) for the program. The Decode Duchenne program is for diagnostic testing only. For carrier testing, please visit: http://www.parentprojectmd.org/decode to submit an application. Please complete every field and tick box clearly.

INDICATIONS FOR TESTING (more than one selection may apply)

- ○ Suspected or confirmed diagnosis of Duchenne or Becker muscular dystrophy
- ○ Elevated creatine kinase (CK) levels

*Please note that this test requisition is not intended for carrier screening of unaffected individuals

PATIENT INFORMATION

Patient's First Name | Patient's Date of Birth (MM/ DD /YYYY)

Patient's Last Name

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) _____			

PROVIDER

Provider's First and Last Name

B0007 (Account #) | Provider's Phone

Provider's Email

Clinic/Hospital/Institution Name

Provider's Street Address

City / Town | State | Zip Code

Country | Provider's Fax

PATIENT SAMPLE INFORMATION

SAMPLE TYPE:
- ○ Saliva Swab
- ○ Whole Blood
- ○ Dried Blood Spots
- Other_____

Collection Date: MM/DD/YY

ICD10 CODES: _____

_____

_____

TEST MENU

- ○ DD006 DMD Creatine Kinase Activity
- ○ DD4045 Comprehensive DMD Test (Seq & Del/Dup)
- ○ DD999 Targeted CNV Analysis*
- ○ DD600 Sanger confirmation and interpretation (per variant)*

*These tests are typically ordered in families with a positive family history. If you have questions, please contact PPMD's Decode Duchenne program at 888-520-8675 option 1

X INSTITUTIONAL BILLING

PPMD (Institution/Organization Name) | B0007 (Institution/Organization Account #)

ADDITIONAL PROVIDER/GENETIC COUNSELOR (IF APPLICABLE)

Ann Martin, MS, CGC (Provider/Genetic Counselor's Name) | B0007 (Provider /Genetic Counselor's Account #)

decode@parentprojectmd.org (Provider/Genetic Counselor's Email) | / (Provider/Genetic Counselor's Phone / Fax)

FAMILIAL SINGLE-SITE TESTING (FOR DD600 SANGER CONFIRMATION)

Proband Last Name, First Name | Proband DOB (MM/ DD /YYYY)

Proband's Accession ID | Relationship to Proband

Positive Control Sample: ○ Already at PKIG ○ To be sent later ○ Not available

Gene	Coding Name (c.)	Protein Name (p.)

FAMILIAL COPY NUMBER VARIANT TESTING (FOR DD999 TARGETED CNV ANALYSIS)

Proband Last Name, First Name | Proband DOB (MM/ DD /YYYY)

Proband's Accession ID | Relationship to Proband

Positive Control Sample: ○ Already at PKIG ○ To be sent later ○ Not available

Gene	CN Event / Size / Exon	Additional CN Event / Size / Exon

Please include a copy of relative's report, if available!

PHYSICIAN STATEMENT

Confirmation of informed and medical necessity for genetic testing

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. De-identified data from the Decode Duchenne program may be shared with the industry sponsors listed above. This de-identified data may be used for diagnostic and therapeutic disease research, such as understanding the incidence of certain dystrophin gene mutations, evaluating and improving the diagnosis of Duchenne/Becker, and developing novel mutation-specific therapeutic strategies.

My signature applies to the informed consent and/or attached letter of medical necessity, if applicable (unless this box is checked). ☐

My signature gives PerkinElmer permission to provide PPMD with this test requisition form and for PPMD to share my name, contact information, and the date on which I executed this TRF with the industry sponsors listed above (unless this box is checked). ☐

Signature_____ Date_____

For general questions on the collection and return of samples, please call: PerkinElmer Genetics at 1-866-354-2910 or email: Genomics@perkinelmer.com
For other information related to this testing or on carrier testing, please contact PPMD and the Decode Duchenne Program at 888-520-8675 option 1 or http://www.parentprojectmd.org/decode.

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

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PHENOTYPE(S) / PATIENT HISTORY (CHECK ALL THAT APPLIES)

Clinical diagnosis: _____

Age of manifestation: _____ ICD-10 Codes: _____

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

MUSCULOSKELETAL

- O Gait difficulties
- O Ambulatory
- O Non-ambulatory
  - Age at which ambulation was lost (if known):_____
- O Calf hypertrophy
- O Muscle weakness/wasting
- O Gowers' maneuver
- O Pain/Cramps
- O Toe walking/Achilles tendon contracture
- O Other:_____

RESPIRATORY

- O Breathing difficulties/reduced pulmonary function on spirometry/ PFT's
- O Ventilator usage, day
- O Ventilator usage, night only
- O Weak cough/need for manual or mechanical assisted cough
- O Other:_____

CARDIAC

- O Diagnosed cardiomyopathy
- O Arrhythmia
- O Decreased cardiac function (shortening fraction or ejection fraction) on echo or cardiac MRI
- O Fibrosis noted on cardiac MRI
- O Other:_____

CENTRAL NERVOUS SYSTEM

- O Autism spectrum disorder (ASD)
- O Attention Deficit Hyperactivity Disorder/Attention Deficit Disorder (ADHD/ADD)
- O Anxiety Disorder
- O Intellectual disability
- O Learning disability
- O Global developmental delay
- O Gross motor delay
- O Fine motor delay
- O Speech delay
- O Other:_____

ORTHOPEDIC FINDINGS

- O Long bone fracture
- O Scoliosis
- O Vertebral fracture
- O Other:_____

LABORATORY

- O Elevated liver enzymes (AST/ALT)
- O Elevated creatine kinase (CK/CPK)
- O Diagnostic muscle biopsy (if yes, specific findings below)
  - Specific findings: _____

MEDICAL

- O Corticosteroid Use

DECODE DUCHENNE INFORMED CONSENT FORM

Decode Duchenne is a genetic testing program that provides free genetic testing to patients who have been unable to access testing due to financial barriers. Decode Duchenne is administered by Parent Project Muscular Dystrophy and is sponsored by Sarepta Therapeutics, PTC Therapeutics, Vertex Pharmaceuticals and NS Pharma.

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 enzyme activity, biomarker tests, and select genetic testing assays will be conducted in Pennsylvania, USA, and all other genetic testing will be conducted in Connecticut, USA.

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.

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 and to Parent Project Muscular Dystrophy (sponsor of the program), 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, no additional tests or anonymization will be carried out on your Sample; no results will be reported and your sample, reports and data that have not been anonymized will be destroyed.
  - ❑ Check here if you would like to opt out of anonymized sample retention. 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 internal statistical, quality analysis, research, scientific and technical development, and market research. PerkinElmer may also share your anonymized data and anonymized report with third parties including Parent Project Muscular Dystrophy.
  - ❑ Check here if you would like to opt out of anonymized data retention. Note, if not checked, this is interpreted as "consent given".
  - ❑ Check here if you would like to opt out of giving permission for PerkinElmer to provide PPMD with your test requisition form, which includes your name and contact information. Note, if not checked, this is interpreted as "consent given" and you understand and agree that PPMD may contact you with information about their programs and resources.

For residents of NY State:

❑ By checking here I give PerkinElmer permission to store my sample for longer than 60 days. Note, if not checked, this is interpreted as "consent not given".

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. My signature acknowledges that I consent to PerkinElmer providing PPMD with my genetic testing report, and that I understand that PPMD may share anonymized data from the Decode Duchenne program with industry sponsors.

Patient Signature (or Parent/Guardian if patient is minor) _____ Date _____

Patient Name _____ Name and Relationship (Parent/Guardian if patient is minor) _____

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.

Family Member Signature _____ Date _____

Family Member 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.

Family Member Signature _____ Date _____

Family Member Name _____ Relationship to Patient _____