



UK Duchenne Muscular Dystrophy Registry

powered by **TrialServe** technology

supported by **Morgan Cole** 

[Clinician's Name and Address]

[Your Name and Address]

[Date] _____

Dear [Clinicians Name] _____,

I am writing to you to ask for medical details so that I / my child (as appropriate) can participate in the UK Duchenne Muscular Dystrophy Registry ["the Registry"].

This Registration Document has been given to me by Parent Project UK Ltd ["PPUK"], who are responsible for the Registry. If you have any further questions about this document or the Registry please contact them at the following address:

PPUK
Epicentre
41 West Street
Leytonstone
London E11 4LJ

020 8556 9955
dmdregistry@ppuk.org

This document contains the following sections; please read them all carefully and **complete Section 6 Participant Medical Data Form** (see below):

1. (This cover letter)
2. **Registration Fact Sheet.** This explains what the Registry is
3. **Participant Consent Form.** This gives my legal consent to hold my data on the Registry
4. **Consent Contact Data Form.** This gives my details for PPUK to contact me
5. **Participant Medical Release Form.** Please read carefully. This gives my legal consent for my (or my child's) medical data to be released.
6. **Participant Medical Data Form.** This is a form that *you (the clinician) must read, fill in and complete.* This form will state whether the medical Information to be released is for me or my child
7. **Participant Genetic Data Form.** The DMD Registry is collecting data relating to the variations in my or my child's dystrophin gene. This information is vital for informing genetic trials and other research; where possible it will be completed online by a geneticist at the testing laboratory to avoid transcription errors, and PPUK will send me a full report documenting this and all data stored in the Registry
8. **Registry Administration Form.** For administrative use only

Once you have completed this information please return the Registration Document to me at the address above.

Thank you for your help and support.

Yours sincerely,

REGISTRATION FACT SHEET

INTRODUCTION

You or your child are being asked to register with the UK Duchenne Muscular Dystrophy Registry [“the Registry”] because it is understood that you or your child [“the Participant”] have a diagnosis of Duchenne or Becker Muscular Dystrophy. The Registry has been established by Parent Project UK Ltd [“PPUK”] who have appointed a Registry Manager. Please read this Registration Fact Sheet carefully and ask any questions you may have before making a decision whether or not to participate. It will be very important for you to understand the kind of information which PPUK intends to keep in the Registry and the purposes for which it will be used.

Please keep the copy we have sent of this Registration Document safe, as it contains important information to which you may wish to refer at a later date.

PURPOSE

The purpose of the Registry is to:

- Develop and encourage the development of more research into Muscular Dystrophy
- Facilitate research by collecting relevant data and making it available for specified research projects
- Use the information provided to understand the disease better
- Establish contact between clinicians, other health professionals, researchers and Registry participants
- Enhance clinicians’ and other health professionals’ ability to deliver treatments for this disease

IMPORTANT FACTS ABOUT THE REGISTRY

- Participation is totally voluntary
- You may ask that your (or your child’s) data be removed or irreversibly anonymised at any time
- No one can find out if you (or your child) are in the Registry, except for the purposes listed
- No information will be given to insurance or related companies
- Any decision whether to join the Registry or to withdraw from it will not change your (or your child’s) medical care or legal rights
- Authorised researchers will look at your (or your child’s) anonymised data for the purposes of research projects which have been approved by PPUK and the Registry Steering Committee
- Authorised health professionals will look at data on your (or your child’s) condition for a specific purpose which will benefit your (or your child’s) treatment and/or the treatment of other participants
- Your (or your child’s) personal details will not be shared with any authorised researcher, authorised health professional or any other third party without your consent. In most circumstances information will be anonymised
- The Registry is regulated by the Data Protection Act (1988) and notification has been made of the purposes of the Registry to the Information Commissioner
- Any research that uses the Registry must have been passed by a medical ethics authority (e.g. COREC in the UK) that is regulated by the EU Clinical Trials Directive 2001, or a non-medical ethical authority (e.g. a university ethical committee) that has been approved by our Steering Committee
- If the medical or genetic data for you (or your child) changes after registration you will have to complete and return a new Medical Release form so that we can obtain this data from your clinician. This form will be available on request or can be downloaded from the website
- The web address for the Registry is <http://www.dmdregistry.org>

SENSITIVE PERSONAL DATA

The Registry will hold the following categories of sensitive personal data about you (or your child):

- Contact information (name, address, telephone and email)
- Basic medical information (date of birth, gender, NHS number)
- Biopsy result and tissue location
- Genetic variation information (DNA mutation)
- Medical interventions (e.g. during research projects)
- Adverse symptoms (e.g. during research projects)
- Clinical assessments (e.g. during research projects)
- Clinical surveys (e.g. post-treatment)

BENEFITS OF PARTICIPATION

Joining the Registry may give you or your child the opportunity to participate in research studies. If you are enrolled, you will receive a newsletter at least once a year about Registry activities and research advances in Duchenne Muscular Dystrophy.

CONFIDENTIALITY

The Registry is controlled by Parent Project UK Ltd in compliance with the Data Protection Act (1988).

JOINING THE REGISTRY

The following section describes the forms that you will be asked to read carefully, fill in and sign, and the procedures that will be followed for you (or your child) to join the Registry.

The forms will be contained in a single document, to make the registration process more reliable. The forms you must fill in and sign are as follows:

- Section 1: Cover letter to your clinician
- Section 3: Participant Consent Form, which will give your legal consent for us to hold your (or your child's) data on the Registry
- Section 4: Consent Contact Data Form containing name, address, phone, and email details, which must be completed so that we can get in contact with you at a later point in time
- Section 5: Participant Medical Release Form, to be completed by you (or your child). This form is a request to your clinician and geneticist to release your (or your child's) basic medical data, disease-related interventions and genetic variation details. If you (or your child) have not yet had a genetic test you can contact your clinician to ask about genetic testing. You will be asked to mail the completed form to us in the enclosed pre-addressed postage-paid envelope

The person giving consent in these forms must be:

- A person with parental responsibility if the child is under the age of 12
- A person with parental responsibility if the child is aged between 12 to 18 and does not have the capacity to understand and to provide consent
- The child, if they are over the age of 12 and have the capacity to understand and to provide consent

The procedure for registration is as follows:

- You will receive two Registration Documents, one for you to keep, and the other for you to complete and send to your clinician
- Your clinician will return this document to you and you must then forward it on to PPUK
- We will review your consent forms and may call you to ask questions. The purpose of this call is to make sure that this information is correct

- We will then enter the data you have given us onto the Registry. Once the data has been entered, we will contact the genetics laboratory where the genetics tests were done with a copy of your medical release form, and ask them to enter your genetic data directly onto the Registry or they will complete Section 7. Geneticists are entering your data directly onto the Registry as a means of transferring the data to you, while reducing the transcription errors that are common when transferring complex genetic data
- Once all your data has been entered onto the Registry you will be sent a printed copy for you and your clinician to validate to ensure it has been entered correctly. You must then return a signed copy of the form to us
- Once you are entered onto the Registry we may contact you and your clinician about opportunities for you or your child to participate in research studies. We advise that you always consult your clinician before agreeing to take part in any proposed research study. If you agree only then will PPUK release your personal details to the researcher
- PPUK registry staff may contact you once a year to update information recorded in the Registry. If this happens it should take about 15 minutes to review the information and make changes. You are also encouraged to contact PPUK to update your details, for example if you move house or take a further biopsy or genetic test
- Once a child is 12, and they have the capacity to understand and to provide consent, they should give consent to join the Registry themselves. Once a child is 18 they will have the right to consent to join the Registry, and the right to withdraw from the Registry. When a child reaches the age of 18 the PPUK Registry Manager will contact the participant for their consent to stay in the Registry

DATA CONTROLLER

Parent Project UK Ltd will act as Data Controller (under the Data Protection Act). PPUK will appoint a Registry Manager who is responsible for collecting, storing, handling and processing your data. In order to administer the Registry, to maintain the data and to update the data they will have access to all the data in the Registry. They are also responsible for informing the Information Commissioner's Office of the purposes for which your data is held. All employees of PPUK have signed an express condition of confidentiality to keep your information confidential. PPUK will only access the data for the purposes for which the Registry is kept.

ELIGIBILITY

Registration is only authorised to people who are registered with a National Health Service consultant clinician in the United Kingdom because they have been diagnosed as having either Duchenne or Becker Muscular Dystrophy.

STEERING COMMITTEE

A Steering Committee made up of at least five senior health professionals and researchers will be responsible for authorising and approving all access to the Registry by any researcher and/or Health Professional for the purposes of a specific research project or for the purposes of a specific course of treatment.

DATA OWNER

Parent Project UK will be regarded as the Data Owner (under the Data Protection Act 1988). This means that PPUK are responsible for access to the data in the Registry.

YOUR RIGHT TO ACCESS DATA

Under the Data Protection Act you have the right to access any data held about you on the Registry. To exercise this right you can contact the Registry Manager. The Registry also holds data specific to research projects. This research data may sometimes not be made accessible until after the specific research project has been completed if the nature of the research project requires this to be released at a future date.

We would also like to offer you the opportunity to go online and securely view your own data using a secure internet connection and password. This opportunity will be available by contacting PPUK. The purpose of this is to encourage your involvement, and reduce the burden on PPUK to respond to data requests under the Data Protection Act.

ACCESS BY HEALTH PROFESSIONALS

Health professionals such as clinicians and geneticists will be asked to release your (or your child's) medical data using the Participant Medical Release Form (Section 5 of this document).

These health professionals will be able to see all participants medical data excluding personal contact details.

Other authorised health professionals may be granted permission by the Steering Committee to look at anonymised data on your (or your child's) condition for a specific purpose which will benefit your (or your child's) treatment and/or the treatment of other participants. If a health professional needs to contact you personally regarding a research project, the Registry Manager will pass on this request to you and your clinician. Your personal contact details will not be released to any health professional without your consent.

ACCESS BY RESEARCHERS

Because of the nature of the research it is not possible to declare all the researchers or organisations that, in the future, might have access to the data in the Registry. However, any access to the data in the Registry must firstly be authorised by the Steering Committee. Access to your data by researchers will be anonymous, as you will be identified using an anonymous reference code.

If a researcher needs to contact you personally regarding a research project, the PPUK Registry Manager will pass on this request to you and your clinician. Your personal contact details will not be released to any researcher or any other third party without your consent.

CONFIDENTIALITY OF RECORDS

We will keep your data private and secure. Your data is protected under the Data Protection Act. Unauthorised third party access will not be allowed.

Results of research carried out using anonymised data from the Registry may be presented at meetings or in publications.

Your health information will be accessed to help research and possibly to develop or plan new test procedures, treatments or products. Health information may be used to report results of research to sponsors and regulators. It may be audited by regulators to make sure that we are following regulations, policies and plans. In these circumstances the information will be accessed anonymously.

ETHICAL APPROVAL

Any research that uses the Registry must have been passed by a medical ethics authority (e.g. COREC in the UK) that is regulated by the EU Clinical Trials Directive 2001, or a non-medical ethical authority (e.g. a university ethical committee) that has been approved by our Steering Committee.

SECURITY

The computer on which the Registry runs will be sited in a secure data centre. This data centre features resilient infrastructure, including multiple levels of 24/7 security, stable high-capacity power supplies, multiple telecoms providers, fire protection, in-house support and environmental controls. The computer is accessed via a secure internet connections, it is password protected and has a firewall. The computer is owned and operated by Javelin Software Ltd on behalf of PPUK Ltd, solely for the purpose of the Registry.

Access to the Registry uses security that adheres to the NHS standards. The Registry is regularly backed up to prevent loss of data.

DATA TRANSFERS

In the event that a research project outside the Registry needs your data it will only be transferred when the following conditions are met:

- The research cannot be done within the Registry database. For example computing the frequency of genetic variants can be done on the Registry database
- Any data transferred is anonymous
- The Steering Committee (consisting of senior medical professionals) has validated that the research will help achieve the goals of the Registry

In addition, under the Data Protection Act a transfer of data can only be made outside the European Economic Area (EU and Iceland, Liechtenstein and Norway), where there is “adequate protection” for the rights and freedoms of the individuals. Before any decision to transfer data outside the European Union is made PPUK will take legal advice for the test of adequacy.

WITHDRAWAL OR COMPLETION

At some time in the future the Registry may cease to operate. It is anticipated that this will occur when it no longer is required to achieve the purposes for which it has been registered. For example if treatments have become widely available and long term studies have been completed. At that point in time you will be contacted to inform you that the Registry is closing and we will either seek consent for the data to be transferred to a clinical database or to be made irrevocably anonymous, or to inform you that the data will be deleted.

If you decided to participate in the Registry your data will be held in the Registry until the Registry ceases to operate or you indicate that you wish to withdraw your data. You can always cancel your consent to participate by writing to the Registry Manager. If you revoke your participation you may be contacted and given the option of your personal data being kept anonymously, rather than deleting it. Cancelling your participation will not change your medical care or legal rights.

By signing this consent form, you give us permission to use and/or share your data for the purposes stated above.

COMPLAINTS

Should you wish to make a complaint regarding the Registry please contact PPUK at info@ppuk.org or write to the address below.

CONTACT

For more information about the DMD Registry please contact the Registry Manager:

Registry Manager
Parent Project UK Ltd
Epicentre
41 West Street
Leytonstone
London
E11 4LJ

Tel: 020 8556 9955

Visit the Registry at <http://www.dmdregistry.org>

Visit Parent Project UK at <http://www.ppuk.org>

PARTICIPANT CONSENT FORM

This section explains consent to your participation in the Registry, if you are over the age of 12.

WHO CAN GIVE CONSENT

The law recognises that a child can consent to their data being held or accessed under the Data Protection Act if the child has reached an age when he/she can decide for himself/herself whether or not to agree. It is generally accepted that by the age of 12 a child can be expected to have sufficient understanding to decide whether or not to agree to their data being supplied, or accessed.

If the person with parental responsibility feels that the child does not have sufficient understanding to consent, then the parent can consent on the child's behalf until the child either reaches such understanding or the child reached the age of 18 whichever is the earlier.

Where a child is under the age of 12 then parental consent will always be required.

PARENTAL RESPONSIBILITY

The person(s) with parental responsibility will usually, but not invariably, be the child's birth parents. People with parental responsibility for a child include: the child's mother; the child's father if married to the mother at the child's conception, birth or later; a legally appointed guardian; adoptive parent; the local authority if the child is on a care order; or a person named in a residence order in respect of the child. Fathers who have never been married to the child's mother will only have parental responsibility if they are registered on the child's birth certificate, if they have a court order granting parental responsibility, or a parental responsibility agreement.

CONSENT, SIGNATURES AND DATES

(Tick to confirm)

I have received two identical copies of this consent form (one to keep and one to return) and have read the contents.

I confirm that I have read and understood the information provided and have had the opportunity to ask questions.

I confirm that I have had sufficient time to consider whether I want myself to participate in the Registry.

I understand that my participation is voluntary and that I am free to withdraw at any time, without giving any reason, without my medical care or legal rights being affected.

This section continues next page.

CONSENT, SIGNATURES AND DATES (CONTINUED)

I understand that my data may be looked at by a responsible individual authorised by Parent Project UK and the Steering Committee, for specific research or treatment. I give permission for these approved individuals to have access to my information.

I agree to take part in the Registry.

My child is under the age of 12 and I am signing on behalf of my child as a person with parental responsibility.

My child is aged between 12 and 18 and does not have the capacity to understand why his consent is being sought. I am giving consent as the person with parental responsibility.

I am over the age of 12 years and I have understood the purposes for which my consent is being requested which have been explained to me.

My Name: _____

Print Name

Signature

Date Signed

For participants between the age of 12 and 18:

I confirm that _____ is over the age of 12 and has had explained to them the purpose for which the data is to be supplied and accessed and has understood the explanation.

Witness: _____

Name

Address

Signed

Date Signed

CONSENT CONTACT DATA FORM

This form is to be filled in and signed by the person giving consent in Section 3.

* Required fields

ROLE*

- Participant
 Parent
 Guardian
 Clinician
 Researcher
 Administrator

CONTACT NAME*

Title*

Forename*

Family name*

CONTACT ADDRESS*

Line 1*

Line 2

Line 3

Line 4

County/City*

Postcode*

Phone 1*

Phone 2

Email

I am over 18 years old

I have read and understood the Registration Fact Sheet (Section 2) and Participant Consent Form (Section 3)

Signed: _____ **Dated:** _____

Thank you for your help.

PARTICIPANT MEDICAL RELEASE FORM

Please read this Participant Medical Release Form carefully and ask any questions you may have before making a decision whether or not to release your (or your child's) medical information. Please keep this form safe, as it contains important information to which you may wish to refer at a later date.

DESCRIPTION OF MEDICAL INFORMATION TO BE RELEASED

The following information is in Section 6 (Participant Medical Data Form):

- NHS Number, name, date of birth, gender
- Biopsy results and tissue location
- Disease, genomic variation related details
- Disease related interventions, (e.g. steroids, surgery, physiotherapy)

DESCRIPTION OF PURPOSE

The medical records will be used by Parent Project UK for purpose of the UK Duchenne Muscular Dystrophy Registry.

CONSENT, SIGNATURES AND DATES

(Tick to confirm)

I hereby give permission for my (or my child's) basic medical data, disease-related interventions and genetic variation details to be disclosed to Parent Project UK Ltd for the UK Duchenne Muscular Dystrophy Registry ["the Registry"].

I confirm that either I am over the age of 12 years, or I have parental responsibility over the child, and I have had explained to me and I have understood the purposes for which my or my child's data is to be held by PPUK.

I have read and understood Section 3 (the Registration Fact Sheet) signed Section 4 (the Participant Consent Form).

I confirm that I have read and understood the information provided and have had the opportunity to ask questions.

I confirm that I have had sufficient time to consider whether I want my or my child's' medical details released to the Registry.

Participant Name _____ Birth Date _____

Clinician's Name _____ Hospital Name _____

My Name _____ Print Name _____

Signature _____ Date Signed _____



PARTICIPANT MEDICAL DATA FORM

This form is to be used if you are releasing data to the **UK Duchenne Muscular Dystrophy Registry** ["The Registry"] for Parent Project UK Ltd ["PPUK"]. You must have a Medical Release Consent Form (Section 5) from the participant, parent or guardian before completing this form and sending it back to the participant. The participant will then forward this Registration Document to PPUK. Please give details of any biopsy that has been taken in 6:12 - if no biopsy has been taken please leave blank. To avoid transcription errors PPUK will send Section 7 of this document to the relevant Genetic Testing Laboratory for online completion. Participants will then be sent a full report of all data held by the Registry.

PARTICIPANT DETAILS (person diagnosed with DMD/BMD)

* Required fields

Title*

Forename*

Family name*

Gender* Male Female

Birth Date* / / DD/MM/YYYY

Disease* Duchenne MD Becker MD undiagnosed

Date of Diagnosis* / / DD/MM/YYYY

NHS Number* NHS 10 digit number

Hospital* Hospital ref*

Genetics Lab*

Important Information

BIOPSY (MUSCLE, BLOOD ETC...)

Date of Biopsy* / / DD/MM/YYYY

Biopsy Lab* Biopsy ref

Muscle Pathology Light Microscopic

Few Necrotic Fibres Mild Degenerative Myopathy Severe Degenerative Myopathy

Inflammation Normal Histopathology

DYSTROPHIN

Immunohistochemistry

Dystrophin 1	<input type="checkbox"/> Absent	<input type="checkbox"/> Focal Reduced	<input type="checkbox"/> Reduced	<input type="checkbox"/> Present
Dystrophin 2	<input type="checkbox"/> Absent	<input type="checkbox"/> Focal Reduced	<input type="checkbox"/> Reduced	<input type="checkbox"/> Present
Dystrophin 3	<input type="checkbox"/> Absent	<input type="checkbox"/> Focal Reduced	<input type="checkbox"/> Reduced	<input type="checkbox"/> Present
Revertant fibres	<input type="checkbox"/> Absent	<input type="checkbox"/> Focal Reduced	<input type="checkbox"/> Reduced	<input type="checkbox"/> Present

Immunoblot dystrophin

Protein level	<input type="checkbox"/> Absent	<input type="checkbox"/> Severely Reduced	<input type="checkbox"/> Reduced	<input type="checkbox"/> Normal
Protein weight	<input type="checkbox"/> Decrease	<input type="checkbox"/> Severely Reduced	<input type="checkbox"/> Reduced	<input type="checkbox"/> Normal

Dystrophin Comments

Utrophin Unregulated	<input type="checkbox"/> Yes	<input type="checkbox"/> No	<input type="checkbox"/> Unknown	
N-NOS	<input type="checkbox"/> Absent	<input type="checkbox"/> Focal Reduced	<input type="checkbox"/> Reduced	<input type="checkbox"/> Present
Laminin Alpha 2	<input type="checkbox"/> Absent	<input type="checkbox"/> Focal Reduced	<input type="checkbox"/> Reduced	<input type="checkbox"/> Present
Dystroglycan Alpha	<input type="checkbox"/> Absent	<input type="checkbox"/> Focal Reduced	<input type="checkbox"/> Reduced	<input type="checkbox"/> Present
Dystroglycan Beta	<input type="checkbox"/> Absent	<input type="checkbox"/> Focal Reduced	<input type="checkbox"/> Reduced	<input type="checkbox"/> Present
Collagen V & VI	<input type="checkbox"/> Absent	<input type="checkbox"/> Focal Reduced	<input type="checkbox"/> Reduced	<input type="checkbox"/> Present

CK Level - 1	<input type="text"/>	Date	<input type="text"/> / <input type="text"/> / <input type="text"/>	DD/MM/YYYY
CK Level - 2	<input type="text"/>	Date	<input type="text"/> / <input type="text"/> / <input type="text"/>	DD/MM/YYYY
CK Level - 3	<input type="text"/>	Date	<input type="text"/> / <input type="text"/> / <input type="text"/>	DD/MM/YYYY

SARCOGLYCAN

Immunohistochemistry

Sarcoglycan Alpha	<input type="checkbox"/> Absent	<input type="checkbox"/> Focal Reduced	<input type="checkbox"/> Reduced	<input type="checkbox"/> Present
Sarcoglycan Beta	<input type="checkbox"/> Absent	<input type="checkbox"/> Focal Reduced	<input type="checkbox"/> Reduced	<input type="checkbox"/> Present
Sarcoglycan Gamma	<input type="checkbox"/> Absent	<input type="checkbox"/> Focal Reduced	<input type="checkbox"/> Reduced	<input type="checkbox"/> Present
Sarcoglycan Delta	<input type="checkbox"/> Absent	<input type="checkbox"/> Focal Reduced	<input type="checkbox"/> Reduced	<input type="checkbox"/> Present

PARTICIPANT GENETIC DATA FORM

This form will be completed by geneticists at your genetic testing laboratory. To avoid transcription errors participants have agreed that wherever possible this should be done online. This form will NOT be completed by your clinician. PPUK will send this form with your Medical Release form to the Genetic Testing laboratory for completion. Participants will receive a full report of this genetic information and all information stored in the Registry after completion of the registration process.

Gene Ref Seq. NM_004006.1 Other

Familial Type* Mother Father Sporadic Unknown

Inheritance Type* Mosaic Unknown

Variant Map

. = Untested (dot)

+ = Unchanged (plus)

- = Deletion (minus)

D = Deletion

M = Point Variant

H = Heterozygous

? = Unknown

	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16	17	18	19	20
.																				
+	21	22	23	24	25	26	26	28	29	30	31	32	33	34	35	36	37	38	39	40
-																				
D	41	42	43	44	45	46	47	48	49	50	51	52	53	54	55	56	57	58	59	60
M																				
H	61	62	63	64	65	66	67	68	69	70	71	72	73	74	75	76	76	78	79	
?																				

DNA TEST

DNA Test Lab **Lab ref**

DNA Test Date / / DD/MM/YYYY

DNA Change (coding change)

DNA Technique DGGE DHPLC HD-CSCE
 MAPH MLPA QF-PCR
 RT-PCR SEQ Southern

Other

REGISTRY ADMINISTRATION FORM

Participant Name _____

Participant number

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AUDIT TRAIL

	D	D	/	M	M	/	Y	Y	Checked by
Date sent to participant			/			/			
Date returned to PPUK			/			/			
Date sent to geneticist			/			/			
Date returned to PPUK			/			/			
Date of final report to participant			/			/			
Date signed final report returned to PPUK			/			/			

AMENDMENTS MADE TO THE DOCUMENT

Date	Page no.	Comments	Amended by

REGISTRY MANAGER VALIDATION

- All data has been correctly transferred to the DMD Registry database
- The registration process is complete
- This document will be kept in a locked and secure place

Registry Manager _____ Print Name

_____ Signature

_____ Date Signed