SERVIZIO SANITARIO REGIONALE
EMILIA-ROMAGNA
Azienda Ospedaliero - Universitaria di Ferrara

Department of Reproduction and Growth&Medical Science
MEDICAL GENETICS UNIT
"STOP-SEQ diagnostic project"

Please fill in the form with all requested information

PATIENT DETAILS

NAME AND SURNAME ____

DATE OF BIRTH_____________________________

GENDER

MALE FEMALE

RESIDENCE (street)______________________ Number______City________postalcode___________Country

Email and telephone____

Sent by (Clinician, please add an e-mail address):____

Positive family history: NO YES (if YES, attach pedigree)

CLINICAL DIAGNOSIS (check as appropriate)

DUCHENNE MUSCULAR DYSTROPHY

BECKER MUSCULAR DYSTROPHY

INTERMEDIATE MUSCULAR DYSTROPHY

HIGH CK

CRAMPS ONLY

CARRIER

CLINICAL DESCRIPTION

Age of onset: ______________

Clinical evaluation: (indicate date of last evaluation__________________________)

Muscle weakness: please indicate MRC score for each of the subsequent muscle group

Muscular district	MRC score right	MRC score left
Neck flexors		
Shoulder abductors		
Elbow flexors		
Elbow extensors		
Hip flexors		
Knee flexors		
Knee extensors		

Calf hypertrophy: NO YES

Contractures: NO YES (if YES, where?____ _)

- Functional abilities:

Indipendent walking

Walking with assistance

Loss of ambulation (from age _____________ years)

Rise from sitting position

Climb stairs with/ without assistance

Unable to climb stairs

- North Star Ambulatory Assessment (NSAA) at last evaluation____
- Six-Minute Walk Distance Test (6MWD) at last evaluation ____
- CPK at onset:_______________________ CPK on last evaluation (if available): ______________________________

- Cardiological evaluation: Dilated cardiomyopathy YES NO
  ECG (attach report) ECHO (attach report)
- Pulmunary evaluation: ____

Steroid therapy:

NO YES Please indicate:

start date____

dosage____

posology____

- Other drug therapy (describe) ____

MUSCLE BIOPSY

- Age: __________________ Muscle: ____
- Immunohistochemical/WB: NO YES (if **YES attach report**)

AVAILABLE SAMPLE/s FOR FURTHER ANALYSES

Muscular biopsy Skin biopsy Fibroblasts Myoblasts

Place, date______________________

Signature_____________________________________

(Clinician Name and Surname, email, fax)

____

Official Stamp

FAMILY PEDIGREE (please indicate the proband or index case by an arrow)