Bethlem and Ullrich Myopathies

(COL6A1, 6A2, 6A3) Sequencing Test

University of Utah Genome Depot
15 North 2030 East
Eccles Institute Human Genetics (EIHG) Room 2260
Salt Lake City, UT 84112-8934
Phone 801-585-3436 FAX 801-585-7177
Contact: Diane Dunn (Laboratory Supervisor)
Email: ddunn@genetics.utah.edu

DNA TEST REQUISITION FORM

CLIA #

46D1010280

Requesting Laboratory Information

Hospital / Lab Name ______________________ Contact Name ______________________

Address ______________________

Phone Number ______________ Fax Number ______________ Email ______________

Ordering Physician ______________ Phone Number ______________ Fax Number ______________

Address ______________________ UPIN ______________

Additional Person to Release Information to: Name ______________________

Address ______________________

Test Information

Date of specimen collection ______________

Specimen preparation and shipping. Specimen preparation and shipping: yellow top ACD or purple top EDTA tubes containing whole blood samples or genomic DNA (at least 2 μg) should be sent via courier at room temperature with this form enclosed. Shipping instructions are available at www.genome.utah.edu/col6a-clinical-testing.

Patient Information

Name Last ______________ First ______________ Middle ______________ Male ☐ Female ☐ Date of Birth ______________

Address ______________________

Phone ______________ Identification Number ______________

Obligations

1. The authorizing physician is obliged to explain this testing and provide genetic counseling. The DNA Testing Consent Form must be signed by the patient and the physician (or genetic counselor).
2. The test results will be returned to the requesting physician. Please contact the referring physician after six weeks.

Prepayment Agreement

Payment is the responsibility of the patient or patient's family. The University of Utah Dept. of Human Genetics does not bill insurance companies, but will issue a receipt for patients to seek insurance reimbursement. Self-payment must be received before any testing is performed; payment is accepted by check or credit card only, and payment must be in US dollars. The Department of Human Genetics will bill ordering labs or institutions.

Test Requested

1 ☐ Direct sequencing analysis of Collagen6A1, 6A2 and 6A3 genes **$2400**

2 ☐ Sequencing of a single exon—for family members of patients with previously detected mutations **$250**
***Please include a copy of the analysis identifying the family member's mutation.

Payment/Billing Information

Check Number __________ Credit Card (circle one) Visa/MC Card # ______________ Exp Date __________ Sec. Code __________

Authorized Signature ______________________ Date ______________

By signing the above you authorize the University of Utah Genome Center to deposit your check or process you credit card, as indicated above, for charges for this testing.

☐ Bill laboratory/institution (must be listed at top of form). Contact information if different than above:

____

Clinical Information **PATIENT'S NAME:** ________________________________

In order to facilitate accurate interpretation of the mutation analysis, accurate clinical information regarding the patient's phenotype is required.

Please fill out the following by checking the appropriate boxes.

1.) Clinical diagnosis: ☐ Ullrich's Congenital Muscular Dystrophy ☐ Bethlem Myopathy ☐ Intermediate

Is the patient: ☐ Symptomatic ☐ Carrier ☐ Pre-Symptomatic

2.) Presenting symptom(s) **(Check all that apply):**
☐ weakness ☐ contractures ☐ hypotonia ☐ delayed motor milestones

☐ Present at birth **or** Age at onset = ________________ years / months (circle one)

3.) CK: ☐ elevated (value = ______________) ☐ normal ☐ unknown

4.) Muscle biopsy collagen staining: ☐ Normal ☐ Abnormal ☐ No biopsy performed ☐ Biopsy results unknown

5.) Skin biopsy collagen expression: ☐ Normal ☐ Abnormal ☐ No biopsy performed ☐ Biopsy results unknown

6.) Current ambulation status: ☐ Walking unassisted ☐ Walking with assistive devices ☐ Not walking
Age at first ambulation ______________________ Age at loss of ambulation ________________________

7.) Feeding difficulties or failure to thrive: ☐ Present ☐ Not present ☐ Unknown
G-tube feeds: ☐ Yes 100% ☐ Yes partial ☐ No ☐ Unknown

8.) Current respiratory support: ☐ None ☐ Part-time (eg. nocturnal) ☐ Full time

9.) Family history of disease: ☐ Yes ☐ No ☐ Unknown (☐ Adopted)
Consanguinity: ☐ Yes ☐ No ☐ Unknown
Inheritance: ☐ Dominant ☐ Recessive
Affected family members____

10.) Physical features **(Check all that apply)**:

Distribution of weakness: ☐ proximal ☐ distal ☐ axial ☐ arms ☐ legs

Contractures: ☐ shoulders ☐ elbows ☐ wrists ☐ fingers ☐ hips ☐ knees ☐ ankles

Joint laxity: ☐ distal ☐ proximal

Skin findings: ☐ keloid ☐ hyperkeratosis ☐ other, specify________________________________

Scoliosis ☐ Yes ☐ No ☐ Unknown/NA

Rigid spine ☐ Yes ☐ No ☐ Unknown/NA

DNA TEST REQUISITION FORM

Consent for Diagnostic DNA Testing
University of Utah
Department of Human Genetics
Patient's Name: __________________________________Date of Birth:________________

The blood or tissue sample I (my child) have (has) provided is required to isolate DNA with which to undertake molecular genetic testing at the University of Utah Genome Center.

- The molecular genetic testing may provide a diagnosis of or indication of risk for myself or my offspring for he condition specified above.
- I understand that this test may not yield results for any combination of the following reasons: 1) The causative mutation may not be detected; 2) Unforeseen technical reasons; 3) Other. I understand that this test does not detect duplications and deletions greater than 1 exon, except for deletions for X-linked genes from male samples.
- I understand that a physician's order is necessary for testing, and that the results will be returned to the ordering physician or laboratory.
- In the case of carrier testing, I understand that in rare cases gene testing of DNA from blood may fail to detect mutations carried in the parental germ-line (ovaries and testes). Because of this I understand that a negative test report cannot absolutely exclude the possibility that I am a carrier.
- I understand that the procedure used to collect the blood or tissue samples has inherent minimal risks which have been explained to me (my child).
- An additional blood or tissue sample may have to be obtained or if the results are inconclusive, or due to unforeseen circumstances
- My (my child's) DNA will be stored in the DNA bank at the University of Utah, Dept. of Human Genetics, Salt Lake City or its responsible delegate
- **I DO** ☐ **DO NOT** ☐ **Initial** _______ agree to allow my (my child's) DNA samples to be used for the purpose of diagnosis, research and development, or quality control at the laboratory. I understand that any information identifying me (my child) will be kept confidential and that any exchange of samples or information will be coded.
- No compensation will be given to me (my child) nor will funds be forthcoming to me (my child) due to invention resulting from research and development using my (my child's) DNA.

Your signature on this form indicates that you have understood to your satisfaction the information regarding molecular genetic testing and agree to participate. If you have further questions concerning matters related to this consent, please discuss them with your medical geneticist, genetic counselor, or referring physician. For other questions, please contact Dr. Russell Butterfield at 801-587-9887.

____ __________
Signature of patient or legal guardian **Date**

____ __________
Signature of witness **Date**

Physician's/Counselor's Statement: I have explained DNA testing to this individual. I have addressed the limitations outlined above and I have answered this person's questions. I will provide appropriate genetic counseling regarding the results

____ __________
Signature of physician or genetic counselor **Date**