

EPIDERMOLYSIS BULLOSA TESTING REQUISITION

All Information Must Be Completed Before Sample Can Be Processed

PATIENT INFORMATION

Patient Name: _____, _____, _____
Last First MI

Mailing Address: _____

Home Phone: _____

MR# _____

Date of Birth: ____/____/____ Gender: Male Female

ETHNIC/RACIAL BACKGROUND (Choose All)

- European American (White) African-American (Black)
 Native American or Alaskan Asian-American
 Pacific Islander Ashkenazi Jewish ancestry
 Latino-Hispanic _____
 (specify country/region of origin)
 Other _____
 (specify country/region of origin)

BILLING INFORMATION (Choose ONE method of payment)

REFERRING INSTITUTION

Institution: _____

Address: _____

City/State/Zip: _____

Accounts Payable Contact Name: _____

Phone: _____

Fax: _____

Email: _____

COMMERCIAL INSURANCE*

Insurance can only be billed if requested at the time of service.

Policy Holder Name: _____

Gender: _____ Date of Birth ____/____/____

Authorization Number: _____

Insurance ID Number: _____

Insurance Name: _____

Insurance Address: _____

City/State/Zip: _____

Insurance Phone Number: _____

*** PLEASE NOTE:**

- We will not bill Medicaid, Medicaid HMO, or Medicare except for the following: CCHMC Patients, CCHMC Providers, or Designated Regional Counties.
- If you have questions, please call 1-866-450-4198 for complete details.

SAMPLE/SPECIMEN INFORMATION

SPECIMEN TYPE: Amniotic fluid Blood Cytobrushes Saliva

Cord blood CVS Bone marrow Other _____

Tissue (specify): _____

Specimen Date: ____/____/____ Time: _____

Specimen Amount: _____

Each test requires 3 mL of whole blood in EDTA tube. Please call before sending alternate tissue samples, and for free cytobrush or saliva collection kits.

DRAWN BY: _____

*Phlebotomist must initial tube of specimen to confirm sample identity

REFERRING PHYSICIAN

Physician Name (print): _____

Address: _____

Phone: (____) _____ Fax: (____) _____

Email: _____

Genetic Counselor/Lab Contact Name: _____

Phone: (____) _____ Fax: (____) _____

Email: _____

____ Date: ____/____/____

Referring Physician Signature (REQUIRED)

Patient signed completed ABN

Medical Necessity Regulations: At the government's request, the Molecular Genetics Laboratories would like to remind all physicians that when ordering tests that will be paid under federal health care programs, including Medicare and Medicaid programs, that these programs will pay only for those tests the relevant program deems to be (1) included as covered services, (2) reasonable, (3) medically necessary for the treatment and diagnosis of the patient, and (4) not for screening purposes.

INDICATIONS/DIAGNOSIS/ICD-9 CODE

Reason for Testing:

- Mutation detection in suspected affected patient
- Carrier testing
- Prenatal diagnosis (by previous arrangement only)

PEDIGREE OR FAMILY HISTORY

Relative(s) with EB symptoms: Y N

If yes, please specify relationship to patient: _____

Parental Consanguinity Y N Unknown

Pedigree: _____

TEST(S) REQUESTED

Next Generation Sequencing Panel

EBSeq Epidermolysis Bullosa Panel

(*CD151, CDSN, CHST8, COL17A1, COL7A1, DSP, DST, EXPH5, FERMT1 (KIND1), ITGA3, ITGA6, ITGB4, JUP, KLHL24, KRT5, KRT14, LAMA3, LAMB3, LAMC2, PKP1, PLEC1, TGM5*)

- Reflex to deletion/duplication of entire panel'
- Reflex to deletion/duplication of single gene(s) (specify): _____

Each gene listed is also available for order as an individual test through custom gene sequencing (see below).

Deletion/Duplication analysis of *KRT14* and *KLHL24* is not available at this time.

Targeted (family specific) mutation analysis for _____ gene

If testing was **not** performed at CCHMC, please include proband's report and at least 100ng of proband's DNA to use as a positive control.

Proband's name _____

Proband's DOB _____

Proband's mutation _____

Please call 513-636-4474 to discuss any family-specific mutation analysis with genetic counselor prior to shipment.

DNA extraction and storage

CUSTOM GENE SEQUENCING

Gene(s) to be sequenced (specify): _____

Only genes with clear published functional relationship to rare diseases are accepted.

Suspected syndrome/ condition: _____

Please choose one of the following:

- Full gene(s) sequencing
- Full gene(s) sequencing with reflex to deletion and duplication analysis, if indicated (**please see list of genes available for del/dup at www.cincinnatichildrens.org/deldup**)
- Familial mutation analysis

Proband's name: _____

Proband's DOB: _____

Proband's mutation: _____

Patient's relation to proband: _____

If testing was **not** performed at CCHMC, please include proband's report and at least 100ng of proband's DNA to use as a positive control.

DELETION AND DUPLICATION ASSAY

Gene(s) to be analyzed (specify): _____

Please see list of available genes at: www.cincinnatichildrens.org/deldup

Suspected syndrome/ condition: _____

Please choose one of the following:

- Deletion and duplication analysis of gene(s) specified above
- Deletion and duplication analysis of gene(s) specified above with reflex to sequencing, if indicated
- Analysis of gene(s) specified above from previously analyzed deletion and duplication
- Familial deletion analysis

Proband's name: _____

Proband's DOB: _____

Proband's mutation: _____

Patient's relation to proband: _____

If testing was **not** performed at CCHMC, please include proband's report and at least 100ng of proband's DNA to use as a positive control.

CLINICAL HISTORY

Suspected Diagnosis _____

Severity _____

Age of Onset _____

Check All That Apply

- Anemia
- Blistering
 - Localized
 - Generalized
 - Mucosal involvement
- Congenital absence of skin
- Corneal erosions
- Dilated cardiomyopathy
- Dystrophic nails
- EB nevi
- Esophageal Dilatation
- Erythema
- Gastrostomy tube
- Hyperpigmentation
- Hypopigmentation
- Keratoderma
- Malnutrition
- Mottled pigmentation
- Nephritis
- Osteopenia/Osteoperosis
- Peeling skin
- Pruritis
- Finger contractures/webbing
- Scarring and milia of the hands and feet
- Squamous cell carcinoma
- Tooth enamel involvement
- Sensorineural hearing loss

Other Symptoms/Congenital abnormalities/malformations /dysmorphic features *(Please specify)*

Previous Testing

Immunofluorescent antibody/antigen mapping: Y N

If Yes; specify result and attach report: _____

Transmission electron microscopy (TEM): Y N

If Yes; specify result and attach report: _____

Genetics Testing: Y N

If Yes; specify gene and results including variants: _____
