Cincinnati Children's
changing the outcome together

GENETICS AND GENOMICS DIAGNOSTIC LABORATORY
For local courier service and/or inquiries, please contact 513-636-4474 • Fax: 513-636-4373
www.cincinnatichildrens.org/moleculargenetics • Email: LabGeneticCounselors@cchmc.org

Mailing Address:
3333 Burnet Avenue, Room R1042
Cincinnati, OH 45229

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.

Cincinnati Children's®
changing the outcome together

Patient Name: _____ Date of Birth: _____

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, KRT1, KRT10, KRT14, KRT2, KRT5, LAMA3, LAMB3, LAMC2, PKP1, PLEC1, TGM5)

☐ Reflex to deletion/duplication of entire panel[1]

☐ Reflex to deletion/duplication of single gene(s)[1] (specify): _____

_____

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

[1]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.

Cincinnati Children's
changing the outcome together

Patient Name: _____ Date of Birth: _____

CLINICAL HISTORY

Suspected Diagnosis _____

Severity _____

Age of Onset _____

Check All That Apply

- ☐ Anemia
- ☐ Blistering
  - ☐ Localized
  - ☐ Generalized
  - ☐ Mucosal involvement
- ☐ Bullous ichthyosiform erythroderma
- ☐ Congenital absence of skin
- ☐ Corneal erosions
- ☐ Dilated cardiomyopathy
- ☐ Dystrophic nails
- ☐ EB nevi
- ☐ Epidermolytic hyperkeratosis
- ☐ Epidermolytic ichthyosis
- ☐ Esophageal Dilation
- ☐ 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: _____

_____