

USIDNET DNA AND CELL REPOSITORY CORIELL CELL REPOSITORIES

SAMPLE SUBMISSION FORM

Please check or complete all applicable items.

| | | |
|--|-----|------|
| This box to be completed by CCR: Repository Number | | |
| Date Received: | | |
| | | |
| Month | Day | Year |

Subject Information

Year of birth _____ State Where Born _____ State of Residence _____

Gender _____ Age at time sample was taken _____
Male/Female

ID number assigned to sample _____
To be provided by Coriell along with the sample shipping container

Diagnosis (please indicate pattern of inheritance and subgroup if known)

Severe Combined Immunodeficiency (SCID) _____
x-linked, ADA, JAK3, $\text{c}\gamma$, RAG1-2, MHC class-II, Artemis, PNP, ZAP70, IL-7 R α CD45, CD25, CD3, unknown, etc.

Chronic Granulomatous Disease (CGD) _____ XLP _____
X-linked, p47phox, p22, p67

Leukocyte Adhesion Deficiency (LAD) _____ X-linked Agammaglobulinemia (XLA) _____
type 1, type 2

DiGeorge Syndrome _____ Wiskott-Aldrich Syndrome (WAS) _____
Classic, mild, XLT

Common Variable Immunodeficiency (CVID) _____
Comments

Hyper-IgM _____ Ataxia-Telangiectasia _____
X-linked, CD40L, CD40, IKK- γ , UNG, AID

Other (please describe) _____
Selective IgA deficiency, complement deficiency, Ig subclass deficiency, specific antibody deficiency, etc.

If an unknown immune deficiency: add age, sex, onset, illnesses, complications, other clinical or laboratory features, and immune defects that have been excluded:

Main clinical, immunological features, complications of this patient:

Molecular Diagnostic Studies (Provide a de-identified copy of the molecular diagnostics report if available.)

Has a molecular diagnosis been established for this patient? _____
Yes, no, kindred member has known mutation, none found

Were the studies done on this submitted cell line? _____ If no, what tissue or culture? _____

What DNA was used? _____ Gene(s) studied? _____
cDNA, genomic DNA

What reference sequence was used for comparison? _____

Mutation identified? y/n _____ Coding region _____ Upstream _____ Downstream _____

Nucleotide base affected by the mutation (e.g., G 1172 A) _____
1st nucleotide of transcript is labeled nucleotide 1, only exons counted

Codon affected (codon change if point mutation, e.g., CGT to IGT) _____

Deletion _____ Insertion _____ Splice site _____

Effects of mutation: Nonsense (direct stop) _____ Missense (aa substitution) _____

Frameshift & stop at _____ Multiple splicing _____ In frame _____

Type of assay used for mutation detection: _____
Sequencing, RFLP, etc.

Is the protein expressed? _____ Method of detection? _____
Yes, no, unknown Western, FACS, other

Additional Patient/Kindred Information

Is the patient in the Registry? _____ Registry number? _____
Yes, no, being simultaneously submitted

Has this patient/family been reported in the literature? _____
Yes, no, please give citation, if known

Have other specimens from this patient/family been stored in the USIDNet Repository? _____

If yes, what are their Repository numbers and what are their relationships? _____

Sample Information

Type of sample submitted? _____
Blood, tissue biopsy, B cell line, T cell line, fibroblast line, other cell type

If the sample is a cell culture, please provide the following information:

Passage number? _____ Number of population doublings? _____ Date culture established? _____
Month/Day/Year

How immortalized? (EBV, HTLV-1, SV40, etc.) _____ IL-2 dependent? _____

Culture medium in which submitted? _____
RPMI -1640, Eagles-Earles, Ham's F10, McCoys 5A, etc. % of each in mixtures

Serum Supplement? Fetal Bovine Serum _____ Other serum? _____ % Used _____

Heat inactivated? _____ Un-inactivated? _____ Special serum requirements? _____

Antimicrobials? Antibacterial _____ Antifungal _____
pen, strep, gentamycin, other fungizone, mycostatin, other

Growth additives? _____
pyruvate, glutamine, non-essential amino acids, other, concentration used?

Cytokines? _____
Identity, source, concentration, special instructions?

Special instructions for growth, handling, freezing? _____
Incubation temperature, %CO₂, % O₂ frequency of feeding, thawing conditions, etc.

Submitter Information

Name _____

Address _____

Telephone _____ Fax _____ E-mail _____

Release, Permission, and Consent Form

Submitter, please sign with your initials in the space provided for each statement that applies.

_____ I have obtained permission/consent for this sample to be stored in the USIDNet DNA and Cell Repository and for progeny cells and derived DNA to be distributed to qualified investigators.
Please provide an unsigned copy of the Informed Consent used to obtain the sample.

_____ No consent is required because the individual who provided the sample is deceased.

_____ No consent is required because my IRB has determined that this submission is exempt.
Please provide a copy of the waiver from your IRB.

_____ The individual from whom the sample was obtained would like the name of their physician to remain linked to the coded sample in the Repository records.

_____ The individual from whom the sample was obtained would like to be re-contacted, through their physician, about research results or with requests to participate in additional research studies.

Submitter's Signature _____ **Date** _____

Transmit the completed form with the submission to:

Barbara Frederick.
Coriell Cell Repositories
403 Haddon Avenue
Camden, NJ 08103
Telephone: 856-757-9690
E-mail: bfrederi@coriell.org

For information about this process, telephone: 800-752-3805 in USA / 856-757-4848 from other countries