

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? _____ cDNA, genomic DNA
Gene(s) studied? _____

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? _____

