

DNA REPAIR ASSESSMENT (DDRFL) PATIENT INFORMATION FORM

Name: _____ MRN#: _____ Sex: <input type="checkbox"/> Male <input type="checkbox"/> Female <input type="checkbox"/> Other: _____ DOB: ____/____/____ MM/DD/YYYY Physician: _____ Institution/Hospital: _____ Specimen Information: Collection Date: ____/____/____ mm/dd/yyyy Collection Time: _____ <input type="checkbox"/> A.M. <input type="checkbox"/> P.M.	IF AVAILABLE, PLACE PATIENT LABEL HERE
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1. Does the patient have a known DNA repair defect? ☐ Yes ☐ No
 If yes, specify: _____

2. If genetic information is available, please provide the following:

- Gene: _____
- cDNA#: _____
- Protein variant: _____
- Zygosity: ☐ Heterozygous ☐ Cpd.Het ☐ Homozygous

3. Family Members affected? ☐ Yes* ☐ No *If yes, ☐ Mother ☐ Father ☐ Siblings, specify: _____

4. Clinical Phenotype:

- Lymphopenia? ☐ Yes* ☐ No *If yes: ☐ T-cell ☐ B-cell ☐ NK cell ☐ Not Available
- ALC from CBC: _____ Date: _____
- Facial dysmorphism? ☐ Yes ☐ No
- Short telomeres? ☐ Yes* ☐ No *If yes, _____%ile Lymphocyte _____%ile Granulocytes _____ ☐ Not Available
- Bone marrow failure? ☐ Yes ☐ No
- Malignancy? ☐ Yes* ☐ No *If yes, specify: _____
- Excessive toxicity to chemotherapy? ☐ Yes ☐ No
- Immunodeficiency (susceptibility to infection)? ☐ Yes ☐ No
- Immune dysregulation? ☐ Yes ☐ No
- Autoimmunity? ☐ Yes ☐ No
- Cytopenias? ☐ Yes ☐ No
- Dermatological findings? ☐ Yes ☐ No
- Maternal engraftment assessed? ☐ Yes ☐ No *If yes, are there maternal T cells present?: ☐ Yes, _____ (%ile) ☐ No
- Age of onset of symptoms: _____ yrs

Abnormal TREC – Newborn screen for SCID: ☐ Yes ☐ No *If yes, result: _____ (copies/uL, Cq, MoM)
 Relative to normal range: ☐ Low ☐ Absent

- Syndromic features? ☐ Yes ☐ No
- Other comments: _____

Treatment:

- Post-HCT: ☐ Yes* ☐ No *If yes, ☐ Myeloablative Conditioning ☐ Reduced Intensity Conditioning ☐ No Conditioning
- Chemotherapy? ☐ Yes ☐ No
- Other relevant treatment: _____

****Note:** In addition to this form, please complete and send the **Diagnostic Immunology Testing requisition form** by adding all the details for the patient, sample, institution, billing details and marking the appropriate tests to be ordered on the sample.