Nationwide Children's
When your child needs a hospital, everything matters.℠

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DNA REPAIR ASSESSMENT (DDRFL) PATIENT INFORMATION FORM

Name: _____

MRN#: _____

Sex: [ ] Male [ ] Female [ ] Other:_____ DOB: ____ / ____ / _____ MM/DD/YYYY

Physican: _____

Institution/ Hospital: _____

Specimen Information:

Collection Date: _____ / _____ / _____ mm/dd/yyyy Collection Time: _____ [ ] A.M. [ ] P.M.

IF AVAILABLE, PLACE PATIENT LABEL HERE

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: _____
  _____
  _____