CRF: PARTICIPANT INFORMATION

Participant Name	
Participant is de-identified	◯ Yes ◯ No
First Name	_____
Middle Name	_____
Last Name	_____
Maiden / Other Name	_____
Patient Initials	_____

Participant Information	
Date of Birth	_____ / _____ / _____
State of Birth	_____
Sex	◯ Male ◯ Female

Race and Ethnicity	
Participant race **(check all that apply)**	□ Caucasian / White □ American Indian / Alaska Native □ Asian □ Native Hawaiian or Pacific Islander □ Black or African American □ More than One Race □ Other (specify) □ Unknown or Not Reported
Other race (specify)	_____
Participant is Hispanic or Latino	◯ Yes ◯ No

Contact Information	
Address	_____
City	_____
State	_____
Zip	_____
Phone	_____
Email	_____

CRF: CONSENT INFORMATION

Participant Consent	
Note: Unconsented data must be de-identified	
Consent Type	◯ Institution-Specific IRB ◯ Schulman IRB ◯ IRB Waiver
Reason for waiver	◯ Patient is deceased ◯ Other
Other reason (specify)	_____
Per consent, patient allows USIDNET to contact him/her.	◯ Yes ◯ No
Per consent, patient allows USIDNET to contact his/her physician.	◯ Yes ◯ No
Consent allows sharing of data with CIBMTR and related research data platforms.	◯ Yes ◯ No
CIBMTR Recipient ID Number	_____
PIDTC Recipient ID Number	_____
Additional data platforms per consent (if applicable)	_____ _____ _____ _____ _____

CRF: FAMILY HISTORY

Family History of PI	
Are there other patients with a primary immunodeficiency in the family?	◯ Yes ◯ No ◯ Unknown
How many relatives also have a PI?	_____
Did a positive family history prompt testing of this patient?	◯ Yes ◯ No ◯ Unknown
Are the parents of the patient related to each other?	◯ Yes ◯ No ◯ Unknown

Information on Affected Family Members		
Relation	**Diagnosis**	**Listed in Registry?**
_____	_____	◯ Yes ◯ No ◯ Unknown
_____	_____	◯ Yes ◯ No ◯ Unknown
_____	_____	◯ Yes ◯ No ◯ Unknown
_____	_____	◯ Yes ◯ No ◯ Unknown
_____	_____	◯ Yes ◯ No ◯ Unknown
_____	_____	◯ Yes ◯ No ◯ Unknown
_____	_____	◯ Yes ◯ No ◯ Unknown

Family History of Other Diseases		
	Has family history of disease	**Disease present in…**
Autoimmune Diseases	◯ Yes ◯ No ◯ Unknown	□ Mother □ Father □ Sister □ Brother □ Daughter □ Son □ Other Female Relative □ Other Male Relative □ Relative Not Specified
Inflammatory Diseases	◯ Yes ◯ No ◯ Unknown	□ Mother □ Father □ Sister □ Brother □ Daughter □ Son □ Other Female Relative □ Other Male Relative □ Relative Not Specified
Malignancies	◯ Yes ◯ No ◯ Unknown	□ Mother □ Father □ Sister □ Brother □ Daughter □ Son □ Other Female Relative □ Other Male Relative □ Relative Not Specified

Family History of Other Diseases			
Relation	**Genetic Testing?**	**Test Results**	**Gene Tested**
______________________	◯ Yes ◯ No ◯ Unknown	◯ No Pathologic Variants ◯ X-Recessive Carrier ◯ Autosomal Recessive Carrier ◯ Same variant as proband without current health issues ◯ Unknown	____________
______________________	◯ Yes ◯ No ◯ Unknown	◯ No Pathologic Variants ◯ X-Recessive Carrier ◯ Autosomal Recessive Carrier ◯ Same variant as proband without current health issues ◯ Unknown	____________
______________________	◯ Yes ◯ No ◯ Unknown	◯ No Pathologic Variants ◯ X-Recessive Carrier ◯ Autosomal Recessive Carrier ◯ Same variant as proband without current health issues ◯ Unknown	____________
______________________	◯ Yes ◯ No ◯ Unknown	◯ No Pathologic Variants ◯ X-Recessive Carrier ◯ Autosomal Recessive Carrier ◯ Same variant as proband without current health issues ◯ Unknown	____________
______________________	◯ Yes ◯ No ◯ Unknown	◯ No Pathologic Variants ◯ X-Recessive Carrier ◯ Autosomal Recessive Carrier ◯ Same variant as proband without current health issues ◯ Unknown	____________

CRF: VISIT INFORMATION

Welcome to the FILL Program!

To begin, please select the appropriate evaluation

Initial Evaluation	**Birth up to 6 months of age**
Mid-year Evaluation (Follow-up)	**From Initial Evaluation up to 1 year of age** **(After receiving primary series of killed vaccines, if given)**
Annual Evaluation (Follow-up)	**1 year up to 18 months of age, and annually thereafter**

☑ This is a FILL visit

Visit covers up to the following date*	_____ / _____ / _____
Which evaluation are you completing?	◯ Initial Evaluation ◯ Mid-Year Evaluation ◯ Annual Evaluation

CRF: INITIAL REGISTRATION

Parent or Legal Guardian Information	
First Name of Parent/Guardian	_____
Last Name of Parent/Guardian	_____
Email of Parent/Guardian	_____
Phone of Parent/Guardian	_____

General Patient Information	
Gestational age at birth	_____ Weeks
Birth Weight	_____ Grams
Twin or multiple birth?	◯ Yes ◯ No
Breastfed?	◯ Yes ◯ No

Evaluation Details	
Initial evaluation prompted by: (select all that apply)	☐ Abnormal newborn screen test ☐ Positive family history ☐ Clinical events or finding
Infection Type	◯ Bacteria ◯ Fungal ◯ Viral ◯ No Proven Organism
Physical Findings	☐ Heart Disease ☐ Hypoglycemia ☐ Syndromic Features
Other (specify)	_____

Syndromic Features	
List features in the space provided.	_____ _____ _____ _____ _____

CRF: CLINICAL HISTORY

NO CLINICAL CONDITIONS PRESENT AT THIS TIME ☐ Select if applicable

Clinical Conditions	
Select all that apply	☐ Hydrops, edema, third spacing ☐ Hygroma or A-V malformation ☐ Chylous effusion ☐ GI malformation, atresia, gastroschisis ☐ Lymphangiectasia
Heart Anomaly	◯ Yes ◯ No ◯ Unknown
Specific anomaly (if known)	_____
Heart surgery in the past 30 days	◯ Yes ◯ No ◯ Unknown
Thymectomy?	◯ Yes ◯ No ◯ Unknown
Infection	☐ Bacteria ☐ Fungal ☐ Viral ☐ No Proven Organism
Neurological Abnormality	☐ Seizures ☐ Hypotonia ☐ Other
Other neurological disorder (specify)	_____
Rash	☐ Erythroderma ☐ Desquamation ☐ Maculopapular ☐ Eczema ☐ Other
Other rash (specify)	_____

Syndromic Features not Listed Above	
Please list all syndromic features for this patient.	_____ _____ _____ _____ _____

Non-Syndromic Features not Listed Above

Please list all non-syndromic features for this patient.

_____

_____

_____

_____

_____

CRF: IMMUNIZATIONS, INFUSIONS, TRANSFUSIONS

Live Vaccinations Given

Note: enter first valid determination for any of the following studies that were performed on your patient. You may leave blank any tests which were not performed or for which the data is not available.

	# of Shots
Measles	◯ 1 ◯ 2 ◯ Total not known
Mumps	◯ 1 ◯ 2 ◯ Total not known
Rotavirus	◯ 1 ◯ 2 ◯ 3 ◯ Total not known
Rubella	◯ 1 ◯ 2 ◯ Total not known
Varicella	◯ 1 ◯ 2 ◯ 3 ◯ 4 ◯ Total not known

Vaccines Withheld

Withhold live rotavirus vaccine?	◯ Yes ◯ No ◯ Unknown
Withhold other vaccine(s)	◯ Yes ◯ No ◯ Unknown

Other Vaccines Withheld

Please include withheld vaccines in this space.

_____

_____

_____

_____

_____

Immunoglobulin Infusions		
Has patient ever received Ig Replacement Therapy?		◯ Yes ◯ No ◯ Unknown
Is patient currently on Ig replacement therapy?		◯ Yes ◯ No ◯ Unknown
Initiation	**Start Date** **Start Age**	_____ / _____ / _____ _____ months _____ years
Termination	**End Date** **End Age**	_____ / _____ / _____ _____ months _____ years
Dose		_____ mg/kg _____ Total grams
Route		◯ IV ◯ SC ◯ IM ◯ Unknown ◯ Other
	Other route (specify)	_____
Frequency		_____ days
Was a port placed specifically for IVIG?		◯ Yes ◯ No ◯ Unknown

Transfusion Treatments	
Transfusion Precautions Advised **(CMV negative, leuko-reduced, irradiated)**	◯ Yes ◯ No ◯ Unknown

CRF: MEDICATIONS AND SUPPLEMENTS

Historical Anti-Infective Treatment	
Palivizumab (synagis) used during RSV activity in community	◯ Yes ◯ No ◯ Unknown

Prophylactic Anti-Infectives			
Please include all antibiotics, anti-virals, and anti-fungals (topical and systemic)			
Drug Name	**Course**	**Adverse Reaction**	**Reaction**
______________________	☐ Continuous ☐ Intermittent ☐ Rotating	◯ Yes ◯ No ◯ Unknown	______________________
______________________	☐ Continuous ☐ Intermittent ☐ Rotating	◯ Yes ◯ No ◯ Unknown	______________________
______________________	☐ Continuous ☐ Intermittent ☐ Rotating	◯ Yes ◯ No ◯ Unknown	______________________
______________________	☐ Continuous ☐ Intermittent ☐ Rotating	◯ Yes ◯ No ◯ Unknown	______________________
______________________	☐ Continuous ☐ Intermittent ☐ Rotating	◯ Yes ◯ No ◯ Unknown	______________________
______________________	☐ Continuous ☐ Intermittent ☐ Rotating	◯ Yes ◯ No ◯ Unknown	______________________
______________________	☐ Continuous ☐ Intermittent ☐ Rotating	◯ Yes ◯ No ◯ Unknown	______________________
______________________	☐ Continuous ☐ Intermittent ☐ Rotating	◯ Yes ◯ No ◯ Unknown	______________________

Historical Immunomodulator Treatment	
Patient has used immunomodulatory medication <u>in the past year</u>	◯ Yes ◯ No ◯ Unknown

Non-Transplant Immunomodulator Medications					
Please include all immunomodulator medications prescribed during the time period of this visit					
Drug Name	**Indication**	**Specify**	**Improvement**	**Adverse Reaction**	**Reaction**
____________	◯ Treat Disease ◯ Prophylaxis ◯ Unknown	____________	◯ Yes ◯ No ◯ Unknown	◯ Yes ◯ No ◯ Unknown	____________
____________	◯ Treat Disease ◯ Prophylaxis ◯ Unknown	____________	◯ Yes ◯ No ◯ Unknown	◯ Yes ◯ No ◯ Unknown	____________
____________	◯ Treat Disease ◯ Prophylaxis ◯ Unknown	____________	◯ Yes ◯ No ◯ Unknown	◯ Yes ◯ No ◯ Unknown	____________
____________	◯ Treat Disease ◯ Prophylaxis ◯ Unknown	____________	◯ Yes ◯ No ◯ Unknown	◯ Yes ◯ No ◯ Unknown	____________
____________	◯ Treat Disease ◯ Prophylaxis ◯ Unknown	____________	◯ Yes ◯ No ◯ Unknown	◯ Yes ◯ No ◯ Unknown	____________
____________	◯ Treat Disease ◯ Prophylaxis ◯ Unknown	____________	◯ Yes ◯ No ◯ Unknown	◯ Yes ◯ No ◯ Unknown	____________
____________	◯ Treat Disease ◯ Prophylaxis ◯ Unknown	____________	◯ Yes ◯ No ◯ Unknown	◯ Yes ◯ No ◯ Unknown	____________

For-Transplant Immunomodulator Medication			
Please include all immunomodulator medications prescribed during the time period of this visit			
Drug Name	**Indication**	**Adverse Reaction**	**Reaction**
________________	◯ Stem Cell: PRE-transplant ◯ Stem Cell: POST-transplant ◯ Organ: PRE-transplant ◯ Organ: POST-transplant ◯ Unknown	◯ Yes ◯ No ◯ Unknown	________________
________________	◯ Stem Cell: PRE-transplant ◯ Stem Cell: POST-transplant ◯ Organ: PRE-transplant ◯ Organ: POST-transplant ◯ Unknown	◯ Yes ◯ No ◯ Unknown	________________
________________	◯ Stem Cell: PRE-transplant ◯ Stem Cell: POST-transplant ◯ Organ: PRE-transplant ◯ Organ: POST-transplant ◯ Unknown	◯ Yes ◯ No ◯ Unknown	________________
________________	◯ Stem Cell: PRE-transplant ◯ Stem Cell: POST-transplant ◯ Organ: PRE-transplant ◯ Organ: POST-transplant ◯ Unknown	◯ Yes ◯ No ◯ Unknown	________________
________________	◯ Stem Cell: PRE-transplant ◯ Stem Cell: POST-transplant ◯ Organ: PRE-transplant ◯ Organ: POST-transplant ◯ Unknown	◯ Yes ◯ No ◯ Unknown	________________
________________	◯ Stem Cell: PRE-transplant ◯ Stem Cell: POST-transplant ◯ Organ: PRE-transplant ◯ Organ: POST-transplant ◯ Unknown	◯ Yes ◯ No ◯ Unknown	________________
________________	◯ Stem Cell: PRE-transplant ◯ Stem Cell: POST-transplant ◯ Organ: PRE-transplant ◯ Organ: POST-transplant ◯ Unknown	◯ Yes ◯ No ◯ Unknown	________________
________________	◯ Stem Cell: PRE-transplant ◯ Stem Cell: POST-transplant ◯ Organ: PRE-transplant ◯ Organ: POST-transplant ◯ Unknown	◯ Yes ◯ No ◯ Unknown	________________

CRF: TRANSPLANTATION AND GENE THERAPY

Transplantation	
Patient has undergone transplant	◯ Yes ◯ No ◯ Unknown
Type of transplant	☐ Hematopoietic Stem Cells ☐ Solid Organ ☐ Unknown

Stem Cell Transplant		
Date of Transplant	**Date**	_____ / _____ / _____
	Age	_____ months _____ years

Solid Organ Transplant		
Date of Transplant	**Date**	_____ / _____ / _____
	Age	_____ months _____ years
Which Solid Organ?		◯ Kidney ◯ Liver ◯ Lung ◯ Heart ◯ Cornea ◯ Thymus ◯ Intestines ◯ Other
	Other (specify)	_____

Transplant Not Performed	
Transplant considered and not performed?	◯ Yes ◯ No
Solid Organ **Other (specify)**	☐ Lack of donor ☐ Age or condition of the patient ☐ Unfavorable probability of success ☐ Religious objections ☐ Insurance denial ☐ Hyper-immune status ☐ Other _____
Hematopoietic Stem Cell Transplant **Other (specify)**	☐ Lack of donor ☐ Age or condition of the patient ☐ Unfavorable probability of success ☐ Religious objections ☐ Insurance denial ☐ Other _____

Gene Therapy	
Patient has undergone gene therapy	◯ Yes ◯ No ◯ Unknown
Date of GT **Date**	_____ / _____ / _____
Age	_____ months _____ years

CRF: SURVIVAL & QUALITY OF LIFE

Patient Status	
Alive?	◯ Yes ◯ No ◯ Unknown
Date of Death	_____ / _____ / _____
Cause(s) of death	☐ Primary immunodeficiency ☐ Malignancy ☐ Infection ☐ Other Cause ☐ Unknown
Other (Specify)	_____

CRF: CORE LABORATORY TEST RESULTS

TREC SCREENING	
Dried blood spot TREC screen	◯ Done ◯ Not Done
Date of Test	_____ / _____ / _____
Testing Site	◯ State NBS Lab ◯ Other Program ◯ Other Lab
Other lab (specify)	_____
if TREC NON-NORMAL, Result	◯ Consistent with SCID ◯ Unspecified ◯ Inconclusive ◯ Incomplete (Poor PCR)

	Value	Type	Units	Result Term
Control DNA	_____	◯ B-actin ◯ RNAseP ◯ Other: _____ _____	◯ copies/uL ◯ Ct	◯ Normal-adequate ◯ Non-normal ◯ Not done / not applicable
TRECs	_____		◯ copies/uL ◯ Ct	◯ Normal ◯ Non-normal

OTHER STUDIES	
Immune studies completed	☐ PHA testing ☐ HIV Testing ☐ Maternal Lymphocyte Engraftment

OTHER STUDIES	
PHA studies performed	☐ PHA by Flow ☐ PHA by 3H-thymidine
PHA by Flow	
Date of Test	_____ / _____ / _____
CD45 cells proliferating	_____ %
Normal for CD45 Lab	_____ >%
CD3 cells proliferating	_____ %
Normal for CD3 Lab	_____ >%
PHA by 3H-thymidine	
Date of Test	_____ / _____ / _____
Patient CPM Medium	_____
Patient CPM PHA	_____
Lab control CPM range medium	_____
Lab control CPM range with PHA**	_____

OTHER STUDIES	
Infant HIV by PCR or Protein Test	
Infant HIV Results	◯ Positive (+) ◯ Negative (-)
HIV Maternal Antibody	
HIV Maternal Antibody Results	◯ Positive (+) ◯ Negative (-)
Maternal Lymphocyte Engraftment	
Results	◯ Searched for and Absent ◯ Searched for and Present
% of CD3	_____%

Complete Blood Count

Date of Test	_____ / _____ / _____			
WBC	_____	THOU/uL		
Platelets	_____	THOU/uL		
RBC	_____	MILL/uL		
Hgb	_____	g/dL		
Lymphocytes	_____	%	_____	/uL
PMN	_____	%	_____	/uL
Eosinophils	_____	%	_____	/uL
Monocytes	_____	%	_____	/uL

Lymphocyte Phenotype

Date of Test	_____ / _____ / _____			
Absolute Lymphocyte Count	_____	/uL		
CD3 T Cells	_____	%	_____	/uL
CD4 Helper T	_____	%	_____	/uL
CD8 Cytotoxic T	_____	%	_____	/uL
CD19 B Cells	_____	%	_____	/uL
CD56/CD16 NK cells	_____	%	_____	/uL

CD4 Subset Panel

Date of Test _____ / _____ / _____

Naive CD4+ T-Cells

- ◯ CD4+ CD45RA+
- ◯ CD4+ CCR7+ CD45RA+
- ◯ CD4+ CD27+ CD45RA+
- ◯ Other or unknown markers used

_____ % _____ /uL

Immunoglobulin Evaluation

Date of Test _____ / _____ / _____

IgG	_____	mg/dL	**IgG1**	_____	mg/dL
IgA	_____	mg/dL	**IgG2**	_____	mg/dL
IgM	_____	mg/dL	**IgG3**	_____	mg/dL
IgE	_____	IU/mL	**IgG4**	_____	mg/dL
IgD	_____	mg/dL			

OTHER STUDIES	
Vaccinations given	☐ Diphtheria ☐ Pertussis ☐ Tetanus ☐ Pneumococcus
Pneumococcus vaccine type	☐ Conjugated vaccine ☐ Unconjugated vaccine
Additional antibody tests	☐ Isohemagglutinin-Anti A ☐ Isohemagglutinin-Anti B

Vaccine Responses		
	# of shots	**Response**
Diphtheria	◯ 1 ◯ 2 ◯ 3 ◯ 4 ◯ Total not known	◯ Absent ◯ Low ◯ Normal
Tetanus	◯ 1 ◯ 2 ◯ 3 ◯ 4 ◯ Total not known	◯ Absent ◯ Low ◯ Normal
Pertussis	◯ 1 ◯ 2 ◯ 3 ◯ 4 ◯ Total not known	◯ Absent ◯ Low ◯ Normal
Isohemagglutinin – Anti A		◯ Absent ◯ Low ◯ Normal
Isohemagglutinin – Anti B		◯ Absent ◯ Low ◯ Normal

Unconjugated Pneumococcal Vaccine Response	
Date of Test	_____ / _____ / _____
# of serotypes producing a protective level	_____ serotypes
# of serotypes TESTED	_____ serotypes
# of times vaccine administered	_____ times

Conjugated Pneumococcal Vaccine Response	
Date of Test	_____ / _____ / _____
# of serotypes producing a protective level **# of serotypes TESTED**	_____ _____
Vaccine Name	□ Prevnar 7 (PCV7) □ Prevnar 13 (PCV13) □ Other □ Unknown
# of times **PCV7** administered # of times **PCV13** administered # of times **Other** administered # of times **Unknown** administered	_____ _____ _____ _____

CRF: MOLECULAR INFORMATION

Genetic Information		
Is this patient's PID linked to a gene mutation?	◯ ◯ ◯	Yes; PI is linked to gene No; Gene not identified Gene tested; no mutation found

Gene Mutation	
Gene	_____
Mutation Type	◯ Complex (hgvs/iscn) ◯ Deletion ◯ Deletion/Insertion (indel) ◯ Duplication ◯ Insertion ◯ Inversion ◯ Substitution
DNA Change	_____
Protein Change	_____
Zygosity	◯ Homozygous ◯ Heterozygous ◯ X-Linked Hemizygous ◯ Unknown
Comments on how mutation was detected	_____

Gene	_____
Mutation Type	◯ Complex (hgvs/iscn) ◯ Deletion ◯ Deletion/Insertion (indel) ◯ Duplication ◯ Insertion ◯ Inversion ◯ Substitution
DNA Change	_____
Protein Change	_____
Zygosity	◯ Homozygous ◯ Heterozygous ◯ X-Linked Hemizygous ◯ Unknown
Comments on how mutation was detected	_____

Gene	_____
Mutation Type	◯ Complex (hgvs/iscn) ◯ Deletion ◯ Deletion/Insertion (indel) ◯ Duplication ◯ Insertion ◯ Inversion ◯ Substitution
DNA Change	_____
Protein Change	_____
Zygosity	◯ Homozygous ◯ Heterozygous ◯ X-Linked Hemizygous ◯ Unknown
Comments on how mutation was detected	_____

Testing Done; Mutation not Found	
Mutation Not Found: Gene	_____

Genetic / Genomic Testing

Note: list positive result only; for pending results and negative results, fill in at later assessment times.

Testing Performed

- ☐ Insertion/Deletion Analysis
- ☐ FISH for 22q deletion / DiGeorge Syndrome
- ☐ Whole exome / genome sequencing
- ☐ Karyotype
- ☐ Other
- ☐ NO TESTING DONE AT THIS TIME

Insertion / Deletion Analysis

Testing Performed

- ☐ Copy number array (CGH array)
- ☐ Multiplex ligation-dependent probe amplification (MPLA)
- ☐ SNP array
- ☐ Other

FISH Testing

Result

- ◯ Deletion Detected
- ◯ No Deletion Detected

CGH Array

Copy Number Array: Result

- ◯ Normal or non-contributory
- ◯ 22q deletion
- ◯ Other abnormality _____

Other: Specify _____

Karyotype

Result

- ◯ Normal or Non-Contributory
- ◯ Other abnormality _____

Other Testing

Name of Test _____

Result

- ◯ Normal or Non-Contributory
- ◯ Other abnormality _____

Other: Specify _____

CRF: DISEASE-SPECIFIC QUESTIONS

Diagnostic Criteria	
HIV has been excluded	◯ Yes ◯ No
Select best diagnosis from list	◯ Typical SCID / Leaky SCID / Omenn Syndrome ◯ Preterm birth alone ◯ Syndrome with variable T cell impairment ◯ Secondary T lymphopenia ◯ Idiopathic T lymphopenia/not known at this time ◯ No significant T lymphopenia or normal

Typical / Leaky / Omenn	
Please Specify	◯ Typical SCID ◯ Leaky SCID ◯ Omenn Syndrome
Typical SCID	◯ Absent or very low (< 300 /uL) T cells & absent or very low (< 10% of lower limit of normal) T cell function ◯ T cells of maternal origin present, but with < 10% of lower limit of normal T cell function
Leaky SCID (1)	☐ < 1000/uL T cell # at < age 2 years ☐ < 800/uL T cell # at age 2 through < 4 years ☐ < 600/uL T cell # at > 4 years ☐ Maternal lymphocytes not detected
Leaky SCID (2)	☐ Absent proliferative responses to candida and tetanus toxoid antigens (post vaccination or exposure), with expression of HLA by flow/serology ☐ Rule-out of MHC Class I and II non-expression by flow cytometry (or histology) ☐ T cell function > 10% and < 30% of normal lower limit (as measured by response to PHA)
Omenn Syndrome	☐ Generalized skin rash ☐ Maternal lymphocytes not detected ☐ Absent or low (< 30% lower limit of normal) T cell proliferation to antigens ☐ > 80% of CD4 T cells are CD45R0+ (< 2 years of age)

Syndrome with variable T-cell impairment	
Select best diagnosis from list	◯ Ataxia telangiectasia ◯ Cartilage Hair Hypoplasia ◯ CHARGE syndrome (coloboma, heart defect, atresia choanae, retarded growth and development, genital and ear abnormality) ◯ CLOVES syndrome (congenital lipomatous overgrowth, vascular malformations, epidermal nevi, and spinal/skeletal anomalies) ◯ DiGeorge / 22q deletion (or TBX1 mutation) ☐ DOCK8 Deficiency ☐ ECC syndrome (ectodermal dysplasia, ectrodactyly and clefting) ☐ EXTL3 deficiency ☐ Fryns syndrome (diaphragmatichernia and other congenital anomalies) ☐ Jacobsen Syndrome (growth and psychomotor retardation, congenital abnormalities, chromosome 11qter deletion) ☐ Nijmegen breakage syndrome ☐ Noonan syndrome (multiple congenital anomalies) ☐ RAC2 defect ☐ Renpenning syndrome ☐ Schimke disease ☐ TAR syndrome (thrombocytopenia, absent radius) ☐ Trisomy 18 ☐ Trisomy 21 ☐ Other DIAGNOSED multi-system syndrome _____ ☐ Unknown or undefined syndrome: list symptoms _____ _____ _____

PEG-ADA Treatment	
Was this patient treated with PEG-ADA?	◯ Yes ◯ No ◯ Unknown
Is the patient currently on PEG-ADA?	◯ Yes ◯ No ◯ Unknown
Immune Reconstitution	◯ Full ◯ Partial ◯ RX Failure
Was ADA used as a "bridge" to another therapy?	◯ Yes ◯ No ◯ Unknown
Dose Schedule	_____ units per week