

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 (diaphragmatic hernia 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
