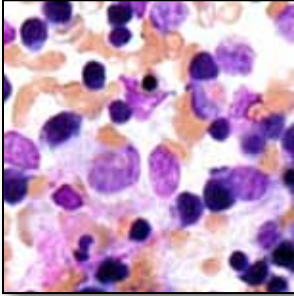


T CELL OR COMBINED DEFICIENCIES



Patients with T cell defects often have unusual infections or atypical infections with common organisms. In children, T cell defects are less common than antibody defects. In adults, T cell defects (other than those resulting from HIV infection) seldom will be new diagnoses. Recognition of patients with significant T cell deficiencies is imperative, because delay of therapy adversely affects outcome. In infants, the frequencies of HIV infection and severe combined immunodeficiency are nearly the same. Thus, in most situations where HIV testing is considered, primary T cell defects should also be considered.

Although severe combined immunodeficiency may have various presentations, one key characteristic is the persistence of infection. In the absence of T cell function, a simple upper respiratory tract infection progresses inexorably over one to two months. An infant with chronic diarrhea, failure to thrive, chronic wheezing, or recurrent Candida and a low lymphocyte count should be suspected.

Common T cell or Combined Deficiencies

Disease	Common Name	ICD 9 Code
Severe Combined Immunodeficiency	“Bubble Boy” Disease, SCID	279.2
DiGeorge Syndrome also known as 22q11 Deletion Syndrome	Thymic Aplasia	279.11
Ataxia Telangiectasia	AT	334.8
Wiskott-Aldrich Syndrome	WAS	279.12

Second tier testing from ADx

Screening laboratory tests

from Advanced Diagnostic Laboratories (ADx) can consist of initial complete and manual blood count, and quantitative serum immuno-globulin (IgG, IgA, IgM and IgE) levels.

	ADx Test Code
Antibody titers to vaccines	TET, DIPHT, PNU12
Lymphocyte phenotyping (CD3, 4, 8, 19, NK)	TBCDC
T cell proliferation to mitogens and recall antigens	LSCAN, LSCON, LSPHA, LSPWM, LSTET
NK cytotoxicity	NKASS
IFN γ /IL12 axis	IL12R, IFNGR, inquire
HLA-DR, CD45, CD16 phenotype	OLM1
Interleukin12, soluble	IL12
TRECs bearing lymphocytes (CD4/CD45RA/CD31)	Inquire
Regulatory T cell panel (CD4/CD25/FoxP3)	Inquire