TABLE 374-2 TESTS MOST FREQUENTLY USED TO DIAGNOSE A PRIMARY IMMUNE DEFICIENCY (PID)

Test	Information	PID Disease
• Blood cell counts and cell morphology	Neutrophil counts[a]	↓ Severe congenital neutropenia, ↑↑ LAD
	Lymphocyte counts[a]	T cell ID
	Eosinophilia	WAS, hyper-IgE syndrome
	Howell-Jolly bodies	Asplenia
• Chest x-ray	Thymic shadow	SCID, DiGeorge syndrome
	Costochondral junctions	Adenosine deaminase deficiency
• Bone x-ray	Metaphyseal ends	Cartilage hair hypoplasia
• Immunoglobulin serum levels	IgG, IgA, IgM	B cell ID
	IgE	Hyper-IgE syndrome, WAS, T cell ID
• Lymphocyte phenotype	T, B lymphocyte counts	T cell ID, agammaglobulinemia
• Dihydrorhodamine fluorescence (DHR) assay Nitroblue tetrazolium (NBT) assay	Reactive oxygen species production by PMNs	Chronic granulomatous disease
• CH50, AP50	Classic and alternative complement pathways	Complement deficiencies
• Ultrasonography of the abdomen	Spleen size	Asplenia

[a]Normal counts vary with age. For example, the lymphocyte count is between 3000 and 9000/μL of blood below the age of 3 months and between 1500 and 2500/μL in adults.

Abbreviations: ID, immunodeficiency; LAD, leukocyte adhesion deficiency; PMNs, polymorphonuclear leukocytes; SCID, severe combined immunodeficiency; WAS, Wiskott-Aldrich syndrome.

absence of natural filtration of microbes in the blood, asplenia predisposes affected individuals to fulminant infections by encapsulated bacteria. Although most infections occur in the first years of life, cases may also arise in adulthood. The diagnosis is confirmed by abdominal ultrasonography and the detection of Howell-Jolly bodies in red blood cells. Effective prophylactic measures (twice-daily oral penicillin and appropriate vaccination programs) usually prevent fatal outcomes.

GATA2 DEFICIENCY

Recently an immunodeficiency combining monocytopenia and dendritic and lymphoid (B and natural killer [NK]) cell deficiency (DCML), also called monocytopenia with nontuberculous mycobacterial infections (mono-MAC), has been described as a consequence of a dominant mutation in the gene *GATA2*, a transcription factor involved in hematopoiesis. This condition also predisposes to lymphedema,

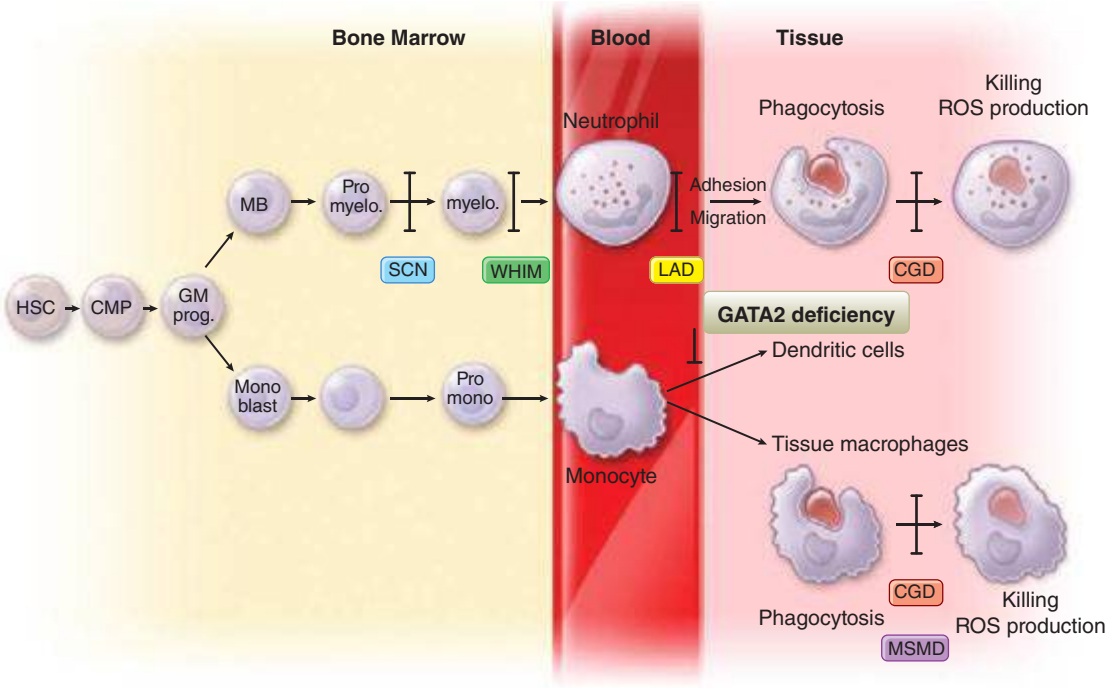

FIGURE 374-1 Differentiation of phagocytic cells and related primary immunodeficiencies (PIDs). Hematopoietic stem cells (HSCs) differentiate into common myeloid progenitors (CMPs) and then granulocyte-monocyte progenitors (GM-prog.), which, in turn, differentiate into neutrophils (MB: myeloblasts; Promyelo: promyelocytes; myelo: myelocytes) or monocytes (monoblasts and promonocytes). Upon activation, neutrophils adhere to the vascular endothelium, transmigrate, and phagocytose the targets. Reactive oxygen species (ROS) are delivered to the microorganism-containing phagosomes. Macrophages in tissues kill using the same mechanism. Following activation by interferon γ (not shown here), macrophages can be armed to kill intracellular pathogens such as mycobacteria. For sake of simplicity, not all cell differentiation stages are shown. The abbreviations for PIDs are contained in boxes placed at corresponding stages of the pathway. CGD, chronic granulomatous diseases; GATA2, zinc finger transcription factor; LAD, leukocyte adhesion deficiencies; MSMD, Mendelian susceptibility to mycobacterial disease; SCN, severe congenital neutropenia; WHIM, warts, hypogammaglobulinemia, infections, and myelokathexis.