

A diagnosis of chronic granulomatous disease is made based upon a thorough clinical evaluation, a detailed patient history, and specialized procedures to measure oxidant production by white blood cells. In healthy individuals, the white blood cells produce a chemical oxidant that destroys bacteria. One blood test for diagnosis of CGD uses a molecule called dihydrorhodamine 123 (DHR) to determine whether or not white blood cells are making these oxidants normally. Oxidants cause the DHR to fluoresce, which is markedly reduced or absent in CGD white blood cells. This test can be performed on blood samples shipped to clinical laboratories certified to perform this test. Another blood test for CGD is called the nitroblue tetrazolium (NBT) slide test. In this test, NBT is mixed with the white blood cells, which are then activated to produce oxidants that react with NBT, turning it a deep blue color. If this reaction does not occur, then these important oxidants are not being produced by an individual's white blood cells. If CGD is diagnosed based on defects in blood cell oxidant production, genetic testing is typically recommended to determine the specific type of CGD.