

CFTR A diagnosis of cystic fibrosis may be suspected based upon newborn screening, identification of characteristic symptoms (e.g., pulmonary disease, pancreatic insufficiency) or a positive family history. The standard diagnostic test for cystic fibrosis is the sweat test, a painless and simple procedure that measures the amount of salt in the sweat. Genetic testing can identify carriers of the defective gene. In May 2005, the U.S. Food and Drug Administration (FDA) approved the first DNA-based blood test to help detect cystic fibrosis. The Tag-It Cystic Fibrosis Kit directly analyzes human DNA to find genetic variations indicative of the disease. All 50 States have newborn screening for CF. In most states, immunoreactive trypsinogen (IRT) assays are performed on dried blood spots from newborns. Trypsinogen is synthesized in the pancreas and IRT levels are elevated in CF. Abnormal IRT results are followed up with sweat testing and/or molecular genetic (DNA-based) testing to confirm the diagnosis.