"Low gamma-GT (GGT) familial intrahepatic cholestasis refers to a spectrum of disease, ranging from mild to severe cases. This spectrum of disease predominantly affects the liver. A variety of disorders leads to low GGT familial intrahepatic cholestasis. Children with defects in bile acid synthesis or conjugation, children with abnormalities of contact between liver cells, children with abnormalities of cell organization manifest as arthrogryposis-renal dysfunction-cholestasis syndrome, and children with ""neonatal hemochromatosis"" all may have low GGT familial intrahepatic cholestasis. These disorders are not covered in this report. A diagnosis of low GGT familial intrahepatic cholestasis should be suspected in infants and children with evidence of cholestasis. A diagnosis may be made based upon a thorough clinical evaluation, a detailed patient history, and a variety of tests. These tests include measuring serum levels of bilirubin, bile salts, and gamma-glutamyltransferase. Surgical excision and microscopic examination of liver tissue (biopsy) may be performed to aid in diagnosis and to detect the presence of cirrhosis. Molecular genetic testing is available on a clinical basis."