Townes-Brocks syndrome (TBS) is an autosomal dominant genetic disorder characterized by absence of the anal opening (imperforate anus), abnormal ears associated with hearing impairment and thumb malformations. Abnormalities in the feet, heart and kidneys also occur frequently. Townes-Brocks syndrome is associated with a mutation in the SALL1 gene. The absence of the anal opening (imperforate anus) is the most common feature of TBS. Small ears and a folded rim of skin and cartilage around the outer ear are usually present. Congenital sensorineural and/or conductive hearing loss can range from mild to severe and can be progressive. The most common thumb malformations are three bones instead of two (triphalangeal thumbs) and duplicated thumbs (preaxial polydactyly). Feet anomalies occur less frequently and include a short third toe, overlapping toes and flat feet. Kidney dysfunction can occur with or without kidney malformations. Typical kidney anomalies include displaced or rotated kidneys, horseshoe kidney, polycystic kidneys, and underdeveloped kidneys. Abnormalities of the heart, eye and spine are infrequently associated with Townes-Brocks syndrome. Mental retardation occurs in approximately 10% of affected individuals. Townes-Brocks syndrome is an autosomal dominant genetic disorder. Dominant genetic disorders occur when only a single copy of an abnormal gene is necessary for the appearance of the disease. The SALL1 gene is the only gene known to be associated with Townes-Brocks syndrome. The abnormal gene can be inherited from either parent, or can be the result of a new mutation (gene change) in the affected individual. Approximately 50% of affected individuals have the condition as a result of a new mutation. The risk of passing the abnormal gene from affected parent to offspring is 50% for each pregnancy regardless of the sex of the resulting child. The prevalence of Townes-Brock syndrome is not known but has been estimated to be at least 1 in 250,000 births. This condition affects males and females in equal numbers. The diagnosis of Townes-Brocks syndrome is based on clinical symptoms. Molecular genetic testing for mutations in the SALL1 gene is available to confirm the diagnosis.