Imperforate anus is a rare inborn abnormality characterized by the absence or abnormal localization of the anus. The rectum or the colon may be connected to the vagina or the bladder by a tunnel (fistula). With surgical correction, normal elimination can become possible. Imperforate anus is an abnormality present at birth, and characterized by the absence of the normal opening of the anus. Elimination of feces may not be possible until surgery is performed. In some cases the rectum opens into the lower part of the vagina in females, or close to the scrotum in males. Imperforate anus and other related abnormalities of the anus and rectum (anorectal malformations) occur in approximately one in 4,000 to 5,000 newborns in the United States. Reported instances of imperforate anus include affected individuals in whom the condition appeared to occur sporadically, members of certain multigenerational families (kindreds), and individuals in whom the condition occurred in association with other birth defects or malformation syndromes (e.g., VACTERL association). Imperforate anus is surgically corrected by dilating, enlarging or repositioning the external opening, or other ways of providing an adequate rectal opening. Genetic counseling may be of benefit to patients and their families.