Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is a rare disorder that affects women. It is characterized by the failure of the uterus and the vagina to develop properly in women who have normal ovarian function and normal external genitalia. Women with this disorder develop normal secondary sexual characteristics during puberty (e.g., breast development and pubic hair), but do not have a menstrual cycle (primary amenorrhea). Often, the failure to begin the menstrual cycle is the initial clinical sign of MRKH syndrome. The range and severity of MRKH syndrome can vary greatly and the disorder is generally broken down into type I, which occurs as an isolated finding, and type II, which occurs with abnormalities of additional organ systems including mainly the kidneys and the skeleton. Because of the nature of the disorder, MRKH syndrome can cause significant psychological challenges and counseling is recommended. The exact cause of MRKH syndrome remains largely unknown, but there is now no doubt of a genetic origin. In this respect, an update on the most recent research publications shows the involvement of several chromosomal segments, some of them including genes likely to account for the disorder.