The WT1 gene is located on chromosome 11p13 and encodes several protein isoforms involved in the urogenital system development. WT1 mutations are identified in patients with WAGR syndrome (Wilms tumor, aniridia, genitourinary tract abnormalities and mental retardation), Denys-Drash syndrome and Frasier syndrome (FS). Heterozygous mutations that compromise intron 9 are responsible for FS. The phenotypic expression of this syndrome is variable; in patients 46XY is characterized by female genitalia, kidney failure during second decade of life, dysgenetic gonads and over 60% risk for gonadoblastoma development. We report a female patient who consulted for primary amenorrhea with 46XY karyotype, female genitalia, kidney failure at age 19, elevated gonadotropins, bilateral ovotestis without gonadoblastoma, with a IVS9 +4C>T mutation in WT1 gene sequence. The presence of a sex development disorder with ovotestis without gonadoblastoma and germ cell tumor, are unusual presentations of this syndrome.