Complete gonadal dysgenesis (CGD, *Swyer's syndrome*) is associated with streak gonads, müllerian structures (due to insufficient AMH/MIS secretion), and a complete absence of androgenization. Phenotypic females with this condition often present because of absent pubertal development and are found to have a 46,XY karyotype. Serum sex steroids, AMH/MIS, and inhibin B are low, and LH and FSH are elevated. Individuals with CGD typically identify as female. The risk of GCC is high, and intraabdominal gonads should be removed. In contrast, patients with *partial gonadal dysgenesis* (PGD, *dysgenetic testes*) may produce enough MIS to regress the uterus and sufficient testosterone for partial androgenization and, therefore, usually present in the newborn period with atypical genitalia, highlighting the spectrum of features that are typically seen with many DSDs.

Testicular dysgenesis can result from disruption of testis-promoting genes (e.g., *WT1, SF1, SRY, SOX9, MAP3K1, DHH, DHX37*, and others) or, rarely, duplication of chromosomal loci containing "antitestis" genes (e.g., *DAX1*) **(Table 390-4)**. Among these, deletions or mutations of *SRY* and heterozygous mutations of *SF1* (*NR5A1*) or *DHX37* appear to be most common but still account collectively for <30% of cases. Associated clinical features may be present, reflecting additional functional roles for these genes. For example, renal dysfunction occurs in patients with specific *WT1* mutations (Denys-Drash and Frasier's syndromes), primary adrenal insufficiency occurs in a minority of patients with disruption of *SF1*, and severe cartilage abnormalities (campomelic dysplasia) are the predominant clinical feature of pathogenic variants in *SOX9*. A family history of DSD, hypospadias, infertility, or early menopause is important because variations in some genes (e.g., *SF1/NR5A1, SOX8*) can be associated with a range of reproductive phenotypes. *SF1* variants are sometimes inherited from a mother in a sex-limited dominant manner (which can mimic X-linked inheritance), and a woman may later develop primary ovarian insufficiency because of the effect of *SF1* on the ovary. Gender identity can be variable in PGD. Dysgenetic testes have an increased risk of GCC. For descended testes, monitoring via physical examination is