

Table 1: Summary of phenotype and genotype findings in the reported cases

Cases	Patient 1	Patient 2	Patient 3	Patient 4	Patient 5
Family Medical History	– Unrelated parents – Testicular ectopia in two 46XY brothers	– Unrelated parents	– Unrelated parents	– Unrelated parents	– Unrelated parents
Prenatal History	– Born at full term – Vaginal delivery	– Born at full term – Vaginal delivery – History of urinary infection – Prenatal diagnosis of an abdominal cystic formation	– Born at full term – Vaginal delivery	– Born at full term – Vaginal delivery	– Born at full term – Vaginal delivery
Age at presentation	Shortly after birth	Prenatal period	Shortly after birth	Shortly after birth	13 years
Birth sex assignment	Male	Male	Male	Female	Female
External genitalia phenotype	– Penile curvature – Posterior hypospadias – Penoscrotal transposition	– Normal phallus – Unilateral ectopic gonad	– Normal phallus size – Posterior hypospadias – Scrotum-located urogenital meatus – Hypoplastic left scrotum – Bilateral inguinal hernia	– 2.5-centimeter curved genital tubercle – Unique urogenital orifice – Fused labioscrotal swellings	– Normal prepubertal female
Internal genitalia phenotype	– Normal left vas deferens and related spermatic vessels	– vagina that ends in the posterior urethra	– No Müllerian-derived internal genitalia	– Normal internal female genitalia,	– Normal internal female genitalia,

	– No right vas deferens was found – Hypoplastic uterus with round ligament	– left fallopian tube cystic vagina – Uterus structure – Hypoplastic right-sided vas deferens		including and uterus and vagina	including an uterus and vagina
Genitography	– Normal masculinized urethra	– 5 cm urogenital sinus with oblong cavity in the median pelvic axis	– Normal masculinized urethra	– 2.5 cm urogenital sinus with vertical female urethra	Not done
Karyotype	45,X/46,XY mosaicism	45, X/46, XY mosaicism	45,X/46X,r(Y) mosaicism	45,X/46,X, der(Y) mosaicism	45X/46X,idic(Y) mosaicism
Gonads	Left: fully descended left testis	Left: streak gonad	– Left: inguinal left testis	Left: gonad not found by pelvic MRI	Left:Streak Gonad
	Right: not found	Right: Fully descended testis	– Right: Fully descended right testis	Right: gonad not found by pelvic MRI	Right:streak gonad
Management	– Removal of the hypoplastic uterine structure and its adnexa. – Testosterone hormone therapy to increase penile length. – Masculinizing surgery	– Resection of Müllerian-derived structures (vagina, uterus, fallopian tube, and left ovary) – The right vas deferens was also resected due to its adherence to the uterine corn	– Inguinal hernia surgery – Masculinizing surgery – scheduled for laparoscopic surgery with resection of the mullerian derivatives	– Feminizing surgery, including clitoral reduction, vaginoplasty, and labioplasty	– Removal of dysgenetic gonads – Hormonal replacement

Outcomes	Favorable	Favorable	Favorable	Favorable	Favorable
Follow-up	17 Years	4 years	3 years	8 years	2 years
Gender identity	Boy	Not yet clearly expressed	Not yet clearly expressed	Girl	Girl