

46,XX/SRY-negative true hermaphrodite

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Objective: To describe genetic evaluation and response to surgery and letrozole therapy of a 46,XX/SRY-negative true hermaphrodite. **Design:** Case report. **Setting:** University Medical Center. **Patient(s):** Nineteen-year-old male with penile hypospadias, micropenis, and cryptorchidism at the time of birth. **Intervention(s):** Unilateral gonadectomy, and contralateral conservative gonadal surgery, followed by therapy with letrozole. **Main Outcome Measure(s):** Histopathologic, genetic and hormonal studies. **Result(s):** Genetic analysis showed that the subject was 46,XX/SRY-negative. Gonadectomy of the left gonad was performed at 16 years. The gonad resected was an ovotestes. The patient's estradiol was high (492 ± 25 pmol/L), whereas the testosterone was low (4.2 ± 0.5 nmol/L). Nineteen months later, conservative gonadal surgery of the contralateral gonad was performed to resect ovarian tissue, and treatment with letrozole was started. During letrozole treatment, testosterone was significantly increas