DOI: 10.4274/jcrpe.galenos.2026.2026-4-6

Letter to the Editor

Anorexia Nervosa Emerging After Swyer Syndrome Diagnosis

Aygün Arı D and Çetinkaya D. Anorexia Nervosa After Swyer Syndrome

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11.04.2026
16.05.2026

Epub: 22.05.2026

Keywords: Swyer syndrome, complete gonadal dysgenesis, anorexia nervosa, psychiatric comorbidity

Dear Editor,

Swyer syndrome (46,XY complete gonadal dysgenesis) is a rare disorder of sex development (DSD) characterized by female external genitalia, hypergonadotropic hypogonadism, primary amenorrhea, and increased risk of gonadal malignancy (1,2). Beyond endocrine and oncologic complications, psychosocial difficulties are increasingly recognized in individuals with DSD. We report an adolescent with Swyer syndrome who developed anorexia nervosa during follow-up after diagnosis and treatment.

A 15.5-year-old phenotypic female presented with primary amenorrhea despite spontaneous breast and pubic hair development since age 11. Physical examination showed Tanner stage IV breast and pubic hair development with normal female external genitalia. Laboratory evaluation demonstrated hypergonadotropic hypogonadism (FSH 138.07 mIU/mL, LH 48.35 mIU/mL, estradiol <12.1 pg/mL). Pelvic imaging revealed absent ovarian tissue and a left adnexal nodular lesion. Karyotype analysis showed 46,XY with positive SRY expression. During the diagnostic process, tumor markers increased (AFP 5.6 ng/mL, β-hCG 6.6 mIU/mL), raising suspicion for gonadal malignancy. Laparoscopic bilateral gonadectomy was performed, and histopathological examination demonstrated gonadoblastoma in both gonads with coexisting dysgerminoma in the left gonad. Postoperatively, estrogen replacement therapy was initiated and later transitioned to cyclic estrogen–progesterone therapy.

At initial psychosocial assessment, no apparent psychiatric risk factors were identified. However, six months after diagnosis, she had intentionally lost 16 kg over three months through severe caloric restriction. Further psychosocial assessment revealed increasing distress related to her diagnosis, concerns regarding gender identity, and dissatisfaction with physical appearance. Her body mass index (BMI) had decreased to 16.7 kg/m² (0.4th percentile), corresponding to 78.8% of median BMI. She demonstrated distorted body image and intense fear of gaining weight and was diagnosed with anorexia nervosa according to DSM-5 criteria. As she remained medically stable, outpatient management was initiated. Her caloric intake was gradually increased, and she was closely monitored during follow-up. No signs of refeeding syndrome or other medical complications were observed during nutritional rehabilitation. She was also evaluated regularly by psychiatry and dietetics teams as part of multidisciplinary care. During follow-up, she regained a healthy weight and maintained positive body image.

Patients with DSD are known to experience increased rates of psychological distress, including anxiety, depression, stigma, impaired self-esteem, and body dissatisfaction (3-5). Current consensus guidelines emphasize psychosocial support as part of multidisciplinary care (4). A large European study reported eating disorders in 11.1% of adults with DSD, particularly among individuals with an XY karyotype raised as females (3). In contrast, studies in adolescents with DSD have focused mainly on anxiety, depressive symptoms, and attention-deficit/hyperactivity disorder, while eating disorders have rarely been discussed (5). To our knowledge, reports of anorexia nervosa in adolescents with Swyer syndrome are scarce. Our case highlights that eating disorders may emerge shortly after diagnosis and treatment, even in adolescents without previous psychosocial risk factors.

This report emphasizes the importance of multidisciplinary management in Swyer syndrome, including routine psychosocial assessment during follow-up. Clinicians caring for adolescents with DSD should remain aware of the potential for eating disorders and body image concerns during the diagnostic and treatment process.

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