# When Identity Feels Divided: A Case Report On Gender Dysphoria

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## Abstract

**Summary:** A psychological condition in which an individual's feeling of gender is not in congruence with the assigned biological sex is known as gender dysphoria.
We shall present the case of a 17-year-old girl who presented with primary amenorrhea to the endocrinology department and was referred to psychiatry. This case report highlights the importance of a multidisciplinary approach.

## Introduction

A psychological condition in which an individual's own feeling of gender is not congruent with the assigned biological sex is known as gender dysphoria. About two-year-old children have gender dysphoria, and it persists into adolescent years, which accounts for between 12% and 27% of the cases.[1]
Gender dysphoria is related to 5 alpha reductase deficiency, a rare cause of disorder of sexual development in males. It is presented as a 46XY karyotype. This case of a 17-year-old girl highlights the importance of a multidisciplinary approach to mitigate the distress and suffering of the individual.

## Case Presentation

A 17-year-old female patient, educated till 6th grade, resident of Rawalpindi, was referred to the Institute of Psychiatry, Benazir Bhutto Hospital, Rawalpindi, in January 2025 by the Endocrinology department of Holy Family Hospital, where she was admitted for evaluation of primary amenorrhea.
The patient presented with 3 years of distress during early adolescence (13-14 years of age). The patient had a strong desire to be treated as male. The informant reported that the patient had no genital ambiguity at the time of birth, but the secondary sexual characteristics were absent, and the patient had primary amenorrhea. The patient was legally and socially raised as female.
As the patient was distressed by being perceived as female, it resulted in social withdrawal and reduced interest in activities. At 15 years of age, the patient noted phallic enlargement concurrent with the first testicular descent.
On examination, the breast was not developed. The pubic hair was absent. The external genitalia were ambiguous. There was a clitoromegaly/phallic-like structure and vaginal slit.
On mental state examination, a young girl of stated age, wearing an abaya and veil, had feminine features. Her thought content as she stated.

مجھے سمجھ نہیں آتا میں کیا ہوں ...میں کون ہوں ...نہ مجھے میرا جسم ٹھیک لگتا ہے، نہ مجھے اپنا آپ باقی لڑکیوں جیسا لگتا ہے۔ میں کبھی نارمل زندگی گزار سکوں گی؟

**Investigations:**
The results of imaging karyotyping and hormonal profile are mentioned in Tables 1 and 2, respectively.

**Table 1: Results of Abdominal Ultrasound, Pelvic Ultrasound, and Cytogenetic Study**

| Investigation | Findings |
|---|---|
| **Abdominal Ultrasound** | Unremarkable |
| **Pelvic Ultrasound** | A soft tissue was visualized posterior to the urinary bladder and anterior to the rectum, likely representing the prostate with a volume of 8ml.<br>Normal testis visualized bilaterally in a **malformed scrotal sac.** |
| **Cytogenetic study** | **46XY Karyotype** |

Using the Revised Child Anxiety and Depression Scale, scores indicated borderline to clinical levels of generalized anxiety and depression.
The IQ was assessed using Raven's Standard Progressive Matrices- average range (55th percentile). No evidence of cognitive impairment.
Based on the detailed evaluation, the patient was diagnosed with Disorders of Sex Development (DSD)- 5 Alpha Reductase Deficiency with Gender Incongruence (ICD-11) or Gender Dysphoria (DSM-5-TR)

**Contributions:**
ATN IJM KKM ZU - Conception, Design
ATN IJM KKM ZU - Acquisition, Analysis, Interpretation
ATN IJM KKM ZU - Drafting
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**Treatment:**
The patient was recommended gender affirming surgery to align the patient's physical characteristics with male gender identity, as advised by the Endocrinology department. A multidisciplinary team was involved, including an endocrinologist, plastic surgeon, gynecologist, psychiatrist, and radiologist.

**Table 2: Hormonal Profile**

| Name Of Test | Level |
|---|---|
| **Serum Estradiol** | 21.50 pg/ml |
| **Ratio of Testosterone/DHT** | 21 |
| **LH** | 5.1 mIU/mL |
| **Testosterone** | 315 ng/mL |
| **Serum 17 OH Progesterone** | 0.17 ng/mL |
| **Dihydrotestosterone** | 149.39pg/ml |

## Discussion

According to DSM-5, Gender dysphoria is a psychological condition in which a person feels incongruent with one's biological gender. This shows that the definition for gender dysphoria in children and adults is the same. It is like our case in which a young girl had primary amenorrhea and felt incongruent with her gender.

A rapid rise has been seen in the number of children and adolescents presenting with gender dysphoria in their dysphoria/incongruence or identifying as transgender in several countries in the last 10 years. The prevalence of gender dysphoria has shown a rapid rise in birth-registered females. The most prevalent mood disorder was gender dysphoria was depression, which was more common in females.[3] The patient who presented to us was being raised as a female. She was expected to fulfill the social responsibility as a female, which resulted in her mood symptoms.

The prevalence of gender dysphoria is increasing, particularly in children. The factors that cause this increase are the surge in role models like gender non-conforming celebrities and media exposure. As the children are not legally allowed to give informed consent, they rely on caregivers and health professionals to make decisions on their behalf. The dysphoria is accompanied by psychosocial stressors and psychiatric illnesses. Although it is important to diagnose gender dysphoria, the psychiatric comorbidities persist over a long time and affect the future of the child.[1]

The deficiency of 5-alpha reductase is a rare cause of sex development-related disorders.

This enzyme is responsible for converting testosterone to 5α-dihydrotestosterone (DHT). The absence leads to impaired virilization and phenotypic ambiguity. The deficiency of the second type of 5α-reductase enzyme affects urogenital development. About 60% of the patients with ambiguous genitalia were raised as females who transitioned to male during puberty. Studies have shown that early diagnosis can limit medical and psychosexual complications due to gender dysphoria.[2]

The psychiatric comorbidities include depression, anxiety, personality-related disorders, and suicidality. It is recommended to have delayed surgery until the gender identity is confirmed. Family counselling is very important for irreversible genital surgery. It is a difficult and delicate task for a multidisciplinary team.[4]

Our case highlights that the presence of 5-alpha-reductase deficiency and gender dysphoria necessitates a holistic and multidisciplinary approach. This report aims to contribute to the growing understanding of gender dysphoria, emphasizing the crucial role of empathy, patient autonomy, and collaborative management in clinical practice. Continued awareness, education, and research in this domain are essential to ensure compassionate and evidence-based care for all individuals navigating gender identity.

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