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Clinical Case Reports

When Identity Feels Divided: A Case Report On Gender Dysphoria

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Abstract

Summary: Gender dysphoria is a psychological condition in which an individual's feeling of gender is not congruent with the assigned biological sex.

We present the case of a 17-year-old girl who presented with primary amenorrhoea to the endocrinology department and was referred to the psychiatry department. This case report highlights the importance of a multidisciplinary approach to treatment.

Introduction

Gender dysphoria is a psychological condition in which an individual's own feeling of gender is not congruent with their assigned biological sex. 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 sexual development disorders in males. The patient presented with 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 individuals.

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 amenorrhoea. The patient was legally and socially raised as female.

As the patient was distressed by being perceived as female, this 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 undeveloped. Pubic hair was absent. The external genitalia were also ambiguous. There was a clitoromegaly/phallic-like structure and a vaginal slit.

On mental state examination, a young girl of the 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 profiling are presented 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. 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 generalised anxiety and depression.

IQ was assessed using Raven's Standard Progressive Matrices average range (55th percentile). There was no evidence of cognitive impairment.

Based on a 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
ATN IJM KKM ZU - Critical Review

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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, including an endocrinologist, plastic surgeon, gynaecologist, psychiatrist, and radiologist, was involved in the study.

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 the DSM-5, Gender dysphoria is a psychological condition in which a person feels incongruent with their biological gender. This shows that the definition of gender dysphoria in children and adults is the same. This is similar to our case, in which a young girl had primary amenorrhoea and felt incongruent with her gender.

Over the last 10 years, several countries have witnessed 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. The prevalence of gender dysphoria has rapidly increased in birth-registered females. The most prevalent mood disorder was gender dysphoria and depression, which was more common in females.[3] The patient who presented to us was being raised as a female. She was expected to fulfil the social responsibility of a female, which resulted in her mood symptoms.

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

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

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

Psychiatric comorbidities include depression, anxiety, personality-related disorders, and suicidality. It is recommended to delay surgery until gender identity is confirmed. Family counselling is crucial before 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 multidisciplinary approach.

This report aims to contribute to the growing understanding of gender dysphoria by emphasising 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, evidence-based care for all individuals navigating gender identity.

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