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BEYOND CHROMOSOMES: MULTIDISCIPLINARY LESSONS FROM A PAEDIATRIC DSD CASE IN INDIA

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ABSTRACT Disorders of sexual development (DSD) represent complex congenital conditions leading to discordance between chromosomal, gonadal, or phenotypic sex. Early recognition and a multidisciplinary approach are essential in diagnosis and management, especially in resource-limited settings. We report a newborn female with ambiguous genitalia, whose clinical evaluation, hormonal and genetic analyses revealed 46,XX DSD. This case highlights contemporary diagnostic challenges, evolving ethical considerations, and underscores the importance of parental counseling and multidisciplinary care in the Indian context.

KEYWORDS : Disorders of sexual development, ambiguous genitalia, multidisciplinary care, 46, XX DSD, pediatrics

INTRODUCTION

Disorders of sexual development (DSDs), encompassing variations in chromosomal, gonadal, or anatomical sex differentiation, challenge traditional male-female binaries and require nuanced clinical care. Globally, approximately 1 in 5,000 live births present with DSD, placing a significant burden on families and healthcare systems. In India, congenital adrenal hyperplasia remains a predominant etiology, but many cases remain undiagnosed or misclassified due to limited resources. Beyond genetics, psychosocial support and ethical considerations are now recognized as integral to holistic DSD management.

CASE REPORT

A neonate with phenotypic female characteristics and 46,XX karyotype, born to a G4P3L2D1 mother via cesarean at term, weighed 3.3 kg with APGAR scores of 7 and 9 at 1 and 5 minutes.. The infant was active, alert, stable, and feeding well.

Clinical Examination Revealed:

- Hyperpigmentation
- Low-set ears
- Depressed nasal bridge
- Enlarged clitoris and labia majora
- Patent anal orifice

Laboratory And Radiological Findings:

Parameter	Result	Normal Range
17-OH Progesterone	0.47 ng/ml	0.26–5.68 ng/ml
Cortisol	12.8 mcg/dl	6.2–19.4 mcg/dl
Testosterone	5.68 ng/dl	–
Karyotype	46,XX	–
Serum Electrolytes	Normal	–
Ultrasound Abdomen	Bilateral ovaries, uterus 2.6 × 0.4 cm	–

Imaging:

- Bilateral ovaries visualized
- Uterus present, measuring 2.6 × 0.4 cm

Figure Legends

Figure 1: Neonate presenting with ambiguous genitalia, including an enlarged phallic structure and partially fused genital folds. Findings are consistent with a disorder of sexual development (DSD).

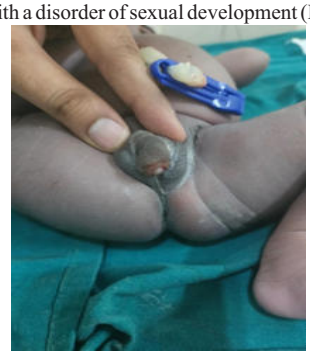

Figure 2: External genitalia of a newborn with ambiguous genitalia showing enlarged phallus with a single perineal opening and fused labioscrotal folds-features consistent with a disorder of sex development (DSD).

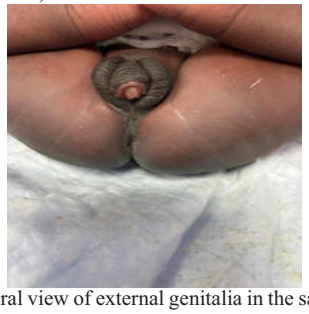

Figure 3: Lateral view of external genitalia in the same newborn with DSD, showing a prominent phallus and labioscrotal fusion. The appearance is suggestive of virilized external genitalia, likely due to a 46,XX DSD (e.g., congenital adrenal hyperplasia).

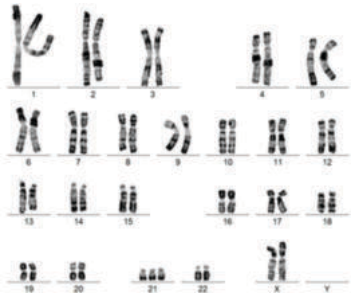

Figure 4: Karyotype analysis demonstrating a 46,XX chromosomal

pattern, confirming genetic sex consistent with female development.

DISCUSSION

Molecular And Clinical Insights:

The SRY gene on the Y chromosome triggers testicular differentiation; its absence generally directs ovarian development. Advances in genetic sequencing reveal that nearly 50% of DSD cases elude definitive genetic diagnosis, highlighting ongoing gaps.

Multidisciplinary Management:

Effective DSD care encompasses endocrinology, genetics, psychology, and ethics. International programs like Empower-DSD demonstrate benefits in psychosocial support and shared decision-making for families. In India, systematic evaluation—clinical, hormonal, radiological-prior to gender assignment is critical to optimize outcomes.

Ethical And Social Perspectives:

Global discourse advocates for patient autonomy and delayed irreversible interventions, reflecting current human rights standards. Cultural sensitivity and robust counseling are vital in the Indian setting, given social stigma.

CONCLUSION

This case underscores the need for early identification of DSDs and a multidisciplinary, culturally aware approach. Combining traditional evaluation with advanced diagnostics and counseling aligns with international standards, improving long-term outcomes.

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