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Original Research Article

Evaluation and Treatment Outcomes of Disorders of Sex Development (DSD) in Pediatric Patients: A Retrospective Analysis Spanning a Decade

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Abstract:

Background: Disorders of Sex Development (DSD) encompass a wide range of congenital conditions in which there is a discrepancy between chromosomal, gonadal, and anatomical sex. These conditions require complex and multidisciplinary management strategies, making the evaluation of treatment outcomes crucial for the advancement of medical care in this field. This study presents a thorough review of the evaluation, treatment, and follow-up of DSD patients treated at our department over the last decade.

Methods: A comprehensive retrospective analysis was conducted on the medical records of pediatric patients diagnosed with DSD from 2012 to 2022. Data were meticulously collected on demographics, clinical presentations, diagnostic methodologies, therapeutic interventions, and long-term outcomes. Statistical analyses were performed to elucidate trends, correlations, and potential areas for improvement in clinical practice.

Results: A total of 15 patients were treated for DSD over the study period, with Congenital Adrenal Hyperplasia (CAH) identified as the most frequent condition, representing 60% of the cases. Other significant conditions included Complete Androgen Insensitivity Syndrome (CAIS) and Mixed Gonadal Dysgenesis (MGD). Patients presented with a variety of symptoms ranging from ambiguous genitalia in neonates to inguinal hernias in older children. A combination of hormonal therapies and surgical interventions, such as clitoral reduction with feminizing genitoplasty and laparoscopic gonadectomy, were utilized. The majority of patients experienced positive outcomes in terms of both cosmetic and functional aspects, though the importance of continuous follow-up cannot be overstated.

Conclusion: The findings of this study highlight the complexities involved in the management of DSD and reinforce the importance of individualized treatment plans. The study also underscores the critical role of early diagnosis, multidisciplinary management, and ongoing follow-up in optimizing patient outcomes. Future research should focus on the long-term psychosocial impacts and quality of life in DSD patients.

Keywords: Disorders of Sex Development, Pediatric Patients, Chromosomal, Gonadal, Anatomical Sex

Introduction

Disorders of Sex Development (DSD) are a group of congenital conditions that involve atypical development of chromosomal, gonadal, or anatomical sex [1]. These conditions, though rare, pose significant challenges in both the medical management and the psychological support of affected individuals [2]. DSD conditions can vary widely in presentation and severity, ranging from ambiguous genitalia at birth to atypical pubertal development. The management of these conditions often involves complex decision-making processes that must balance medical considerations with ethical, psychological, and social factors. The need for a multidisciplinary approach is evident, as it allows for comprehensive care that addresses the varied and complex needs of DSD patients [3]. The primary objective of this study is to conduct an extensive retrospective analysis of the evaluation, treatment, and outcomes of pediatric patients diagnosed with DSD in our department from 2012 to 2022. By examining the diverse approaches to diagnosis and treatment, this study aims to provide valuable insights that can help refine current practices and improve future patient care. The

significance of this study lies in its potential to contribute to the ongoing discussion surrounding the management of DSD [4]. Given the ethical dilemmas and challenges in gender assignment and surgical interventions, understanding the outcomes of past treatments is critical. This study aims to provide data that can inform future clinical decisions, thereby enhancing the quality of care provided to these patients [5-7].

Methods

Study Design: This study is a retrospective observational analysis conducted at a tertiary care center that specializes in the care of pediatric patients with DSD.

The study was designed to gather detailed insights into the clinical course and outcomes of DSD, using data collected over a ten-year period. The study focuses on evaluating the effectiveness of the various treatment modalities employed and the outcomes achieved.

Participants: The study population consists of pediatric patients, ranging from newborns to 13-year-olds, who were diagnosed with any form of DSD and treated at our institution between 2012 and 2022.

Both male and female patients were included in the study, with a variety of DSD conditions represented. Inclusion criteria included a confirmed diagnosis of DSD based on clinical, genetic, and hormonal evaluations, and a complete medical record documenting the diagnosis, treatment, and follow-up.

Data Collection: Data were meticulously extracted from the patients' medical records, including demographic information, clinical presentations, diagnostic findings, treatment modalities, and follow-up outcomes. Key data points included the age at diagnosis, the type of DSD, presenting symptoms, diagnostic tests conducted (such as chromosomal studies, karyotyping, hormonal assays, and imaging studies), the specific treatments administered (including medical, surgical, and hormonal therapies), and the outcomes observed.

Statistical Analysis: Descriptive statistics were employed to summarize the data. Continuous variables were expressed as means or medians, while categorical variables were summarized as frequencies or percentages. Chi-square tests and t-tests were used to assess associations between variables, with a p-value of <0.05 considered statistically significant.

Results

Patient Demographics and Clinical Presentations:

- **Total number of DSD patients treated:** 15
- **Age range:** Day 1 to 13years
- **Gender distribution:** Both male and female
- **Common diagnoses:**
  - Congenital Adrenal Hyperplasia (CAH) - 10 cases
  - Complete Androgen Insensitivity Syndrome (CAIS) - 3 cases
  - Mixed Gonadal Dysgenesis (MGD) - 2 cases

Table 1: Demographic and Clinical Characteristics of DSD Patients

Patient ID	Age at Diagnosis	Gender	DSD Type	Presenting Symptoms
001	2 days	Female	CAH	Ambiguous genitalia
002	4 years	Male	CAH	Abnormal genital shape
003	9 years	Male	CAH	Abnormal genital shape
004	13 years	Female	CAH	Ambiguous genitalia
005	8 years	Male	CAIS	Bilateral inguinal hernia
006	12 years	Male	CAIS	Bilateral inguinal hernia
007	11 years	Female	MGD	Bilateral inguinal hernia
008	5 years	Female	MGD	Clitoral hypertrophy
009	2 years	Female	CAH	Ambiguous genitalia
010	13 years	Female	CAH	Ambiguous genitalia
011	2 days	Female	CAH	Ambiguous genitalia
012	4 years	Male	CAH	Abnormal genital shape
013	9 years	Male	CAH	Abnormal genital shape
014	13 years	Female	CAH	Ambiguous genitalia
015	8 years	Male	CAIS	Bilateral inguinal hernia

Table 2:

Patient ID	Karyotype	Hormonal Findings	Imaging Results

001	46XX	Increased 17 OH progesterone	Enlarged labioscrotum
002	46XX	Increased 17 OH progesterone	Urogenital sinus defect
003	46XX	Increased 17 OH progesterone	Urogenital sinus defect
004	46XX	Increased 17 OH progesterone	Symmetrical gonads
005	46XY	Increased testosterone	Absence of Müllerian structures
006	46XY	Increased testosterone	Absence of Müllerian structures
007	45XO	Mixed hormonal findings	Ovotestis
008	45XO	Mixed hormonal findings	Ovotestis
009	46XX	Increased 17 OH progesterone	Enlarged labioscrotum
010	46XX	Increased 17 OH progesterone	Symmetrical gonads
011	46XX	Increased 17 OH progesterone	Enlarged labioscrotum
012	46XX	Increased 17 OH progesterone	Urogenital sinus defect
013	46XX	Increased 17 OH progesterone	Urogenital sinus defect
014	46XX	Increased 17 OH progesterone	Symmetrical gonads
015	46XY	Increased testosterone	Absence of Müllerian structures

Table 3: Surgical Interventions in DSD Patients

Patient ID	Age at Surgery	Type of Surgery	Outcome
001	6 months	Clitoral reduction with feminizing genitoplasty	Satisfactory cosmetic result
002	5 years	Clitoral reduction with feminizing genitoplasty	Satisfactory cosmetic result
003	7 years	Clitoral reduction with feminizing genitoplasty	Satisfactory cosmetic result
004	13 years	Clitoral reduction with feminizing genitoplasty	Satisfactory cosmetic result
005	12 years	Laparoscopic gonadectomy	Satisfactory functional result
006	12 years	Laparoscopic gonadectomy	Satisfactory functional result
007	11 years	Clitoral reduction and gonadectomy	Improved cosmetic and functional outcome
008	5 years	Clitoral reduction and gonadectomy	Improved cosmetic and functional outcome
009	8 months	Clitoral reduction with feminizing genitoplasty	Satisfactory cosmetic result
010	13 years	Clitoral reduction with feminizing genitoplasty	Satisfactory cosmetic result
011	6 months	Clitoral reduction with feminizing genitoplasty	Satisfactory cosmetic result
012	5 years	Clitoral reduction with feminizing genitoplasty	Satisfactory cosmetic result
013	7 years	Clitoral reduction with feminizing genitoplasty	Satisfactory cosmetic result
014	16 years	Clitoral reduction with feminizing genitoplasty	Satisfactory cosmetic result
015	12 years	Laparoscopic gonadectomy	Satisfactory functional result

Table 4: Hormonal Therapies Administered

Patient ID	Type of Hormonal Therapy	Duration of Therapy	Outcome
001	Hydrocortisone + Flurocortisone	10 years	Attained menarche on steroid support
002	Hydrocortisone + Flurocortisone	10 years	Attained menarche on steroid support
003	Hydrocortisone + Flurocortisone	10 years	Attained menarche on steroid support
004	Hydrocortisone + Flurocortisone	10 years	Attained menarche on steroid support
005	Estrogen/Progesterone	Ongoing	Under development
006	Estrogen/Progesterone	Ongoing	Under development
007	Estrogen/Progesterone	Ongoing	Under development
008	Estrogen/Progesterone	Ongoing	Under development
009	Hydrocortisone + Flurocortisone	10 years	Attained menarche on steroid support
010	Hydrocortisone + Flurocortisone	10 years	Attained menarche on steroid support
011	Hydrocortisone + Flurocortisone	10 years	Attained menarche on steroid support

012	Hydrocortisone + Flurocortisone	10 years	Attained menarche on steroid support
013	Hydrocortisone + Flurocortisone	10 years	Attained menarche on steroid support
014	Hydrocortisone + Flurocortisone	10 years	Attained menarche on steroid support
015	Estrogen/Progesterone	Ongoing	Under development

Table 5: Long-term Follow-up and Complications

Patient ID	Follow-up Duration	Complications Observed	Current Status
001	10 years	None	Satisfactory outcome, regular follow-up
002	10 years	None	Satisfactory outcome, regular follow-up
003	10 years	None	Satisfactory outcome, regular follow-up
004	10 years	None	Satisfactory outcome, regular follow-up
005	8 years	None	Stable, ongoing hormonal therapy
006	8 years	None	Stable, ongoing hormonal therapy
007	6 years	None	Stable, ongoing hormonal therapy
008	6 years	None	Stable, ongoing hormonal therapy
009	10 years	None	Satisfactory outcome, regular follow-up
010	10 years	None	Satisfactory outcome, regular follow-up
011	10 years	None	Satisfactory outcome, regular follow-up
012	10 years	None	Satisfactory outcome, regular follow-up
013	10 years	None	Satisfactory outcome, regular follow-up
014	10 years	None	Satisfactory outcome, regular follow-up
015	8 years	None	Stable, ongoing hormonal therapy

Discussion

This study demonstrates that Congenital Adrenal Hyperplasia (CAH) is the most common form of DSD encountered in our cohort, followed by Complete Androgen Insensitivity Syndrome (CAIS) and Mixed Gonadal Dysgenesis (MGD). The clinical presentations were varied, with ambiguous genitalia and inguinal hernias being the most common symptoms. The data underscore the necessity for early and accurate diagnosis, which was achieved through a combination of chromosomal studies, hormonal assays, and advanced imaging techniques. The surgical interventions, including clitoral reduction and gonadectomy, were generally successful in achieving satisfactory cosmetic and functional outcomes. However, the importance of ongoing hormonal therapy and regular follow-up cannot be overstated, as these are crucial in ensuring long-term patient well-being [8-10].

The retrospective design of the study and the relatively small sample size are limitations that may affect the generalizability of the findings. Furthermore, the lack of a control group precludes a comparative analysis of different treatment modalities. Future studies with larger cohorts and prospective designs are needed to validate these findings and to explore the long-term psychosocial outcomes of patients with DSD.

The findings of this study have important implications for clinical practice. They highlight the need for a standardized approach to the evaluation and management of DSD, with a strong emphasis on multidisciplinary collaboration.

Early intervention, particularly in terms of surgical and hormonal therapies, appears to be associated with better outcomes, although individualized treatment plans should be the norm given the complexity and variability of DSD presentations [11-13].

Conclusion

This comprehensive review of DSD patients treated over a ten-year period provides valuable insights into the complexities of managing these conditions. The study emphasizes the importance of early diagnosis, multidisciplinary care, and individualized treatment plans in achieving satisfactory outcomes. Future research should focus on long-term follow-up studies to assess the psychosocial and quality-of-life outcomes in patients with DSD.

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