

Management of Disorders of Sexual Development' in Pakistan: Issues and Way Forward

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Abstract

The complex nature of DSD (Disorders of sexual development) warrants the correctness of diagnosis, psychological and cultural aspects of the problem and timely intervention. Genital Reconstructive Surgery is the most essential part of the management. The sensitive nature of the disease and overlapping corrective options, makes this disorder a management dilemma. More physicians' training is required for diagnoses and timely referral to multidisciplinary team with focus on ethical decision-making. This was the main aim of this panel discussion reported in this article. The content analysis of the discussion was phrased, coding done and themes were generated. The themes were then linked together to reach a consensus. The idea of capacity building amongst care givers and public awareness in this regard was also pressed upon. Consensus was the team approach and utilizing the human resource in effective and timely manner, as a way forward.

Keywords: DSD; Genital Reconstructive Surgery; Ethical Decision-Making; Timing of Surgery; Special Physician Training

Introduction

Disorders of sexual development (DSD) is a heterogeneous group of congenital conditions, resulting in discordance between an individual's sex chromosomes, gonads, and/or anatomic sex [1]. Clinical presentations range from the ambiguity of the external genitalia at birth, deviation of body physiology from assigned gender, to variation in social and psychological behavior of individuals. Their management has always been challenging especially the timing of gender assignment. Diverse Medical specialties are involved in the life long management and support of these patients, with an aim to make them a valuable member of society. Obstetricians at birth and pediatric endocrinologists come across in neonatal and early years of life, dealing with identification and diagnosis. Then, Pediatric surgeons/

urologists and gynecologists are involved in their anatomical and physiological problems. Psychosocial issues, especially “gender dissatisfaction”, are dealt with by Psychologists and intersex educator [1].

But there is often a disconnect between the disciplines. Timing of gender assignment and reconstructive genital surgeries are the main area of conflict among different specialties. There is a dire need of a combined approach to provide constant support and a smooth transition of care from pediatric to adult age group. Such multidisciplinary collaborative approach is in practice internationally [2,3]. However, in low- and middle-income countries (LMIC) like Pakistan, providing such care to a DSD patient is difficult, if not impossible.

Understanding the need and benefit of a collective approach, the Association of Pediatric Surgeon of Pakistan in its 29th International Conference, had arranged an hour-long session of joint panel discussion with the Society of Obstetrics and Gynecology Pakistan and the Pediatric Endocrine and Diabetes Society. Invited expert panelists were the representatives of pediatric endocrinology, pediatric surgery, gynecology, pediatric and adolescent gynecology (PAG), psychology and social advocate for DSD individuals. They all actively involved in managing DSD cases. Current report is an excerpt of the proceedings of the session.

Materials and Methods

The study was conducted after the approval from Institutional Ethical Review Board Ref. No. NICH/EX-19/21 and conference organizing committee. This was a content analysis of the scientific session held in the Pediatric Surgery conference in Karachi, Pakistan. The vision and perspective of the experts narrated by themselves were transcribed. The session was conducted in English therefore translation was not required. It was a hybrid conference and the proceedings were not only recorded but was also broadcast on social media channels. Field notes were taken by the moderator which helped in triangulation of data and transcripts were shared and validated by the panelists for member checking. Initials of panelists are used here for the sake of convenience. All the panelists are the authors of this report and therefore consent was exempted. An online meeting was arranged to clarify the doubts that arose during the discussion and to elaborate the points that could not be discussed due to time limitation.

Analysis was conducted by identifying keywords and phrases, these unedited verbatim provided the basis for coding. Codes were clustered as trends and overarching themes were extracted from these trends. These themes were then linked together to formulate a conclusion.

Results

The panelists elaborated upon diagnostic approach, management and counselling related issues of this complex health condition. The discussion generated was based on provider related issues. There were few areas where consensus was achieved and some points generated discussion with some difference of opinions. Following themes emerged from the content analysis after using a deductive coding approach.

	Theme	Sub-themes	
I	Importance of early diagnosis	a)	Universal Screening of (CAH) at birth
		b)	Genetic work-up
II	Individualisation of cases	a)	Psychosocial and Cultural aspect of gender assignment
		b)	Timing of surgeries (early vs delayed surgery)
III	Challenges in management of DSD in Pakistan	a)	Need of capacity building
		b)	Developing awareness (general public and health care providers)
IV	Effect of covid pandemic on elective surgeries of DSD		

Table 1: Themes and sub-themes.

Theme I: Importance of early diagnosis

Majority of DSD cases are not life threatening except CAH and it was agreed upon that a proactive approach can save cases of CAH especially from complications related to salt loss and severe dehydration. Consensus was also reached that early diagnosis of DSD is of paramount importance in view of saving lives and achieving socio-cultural norms as well as religious rituals. Such real life dilemmas are often faced by pediatricians and pediatric endocrinologists in Pakistan, 'DSD is still considered to be more of a social rather than a medical emergency in most developing countries [4].

CAH screening at birth is not being performed at present. Hormonal assays, karyotyping genitogram and MRI are being done in few centers and are very expensive. For genetic/molecular testing samples are still sent outside Pakistan.

Theme II: Individualization of cases

Psychosocial and cultural aspect of gender assignment

The birth of a baby with DSD is considered a social stigma in our society. In a male dominant culture like Pakistan, if the gender of a newborn is uncertain, it poses a great stress to the whole family. The parents usually opt for a male gender without considering the emotional and psychological impact on the child. Psychosexual assessment of these individuals reveals that they feel uncomfortable and insecure, fearing rejection.

Intersex educator and counselor on the panel was of the opinion that parents need to be counseled regarding complexities involved in the management along with emotional and social wellbeing of DSD individuals.

Another prospect emerged was that Pakistan being an Islamic country, western principles cannot be applied here. Physicians need to consider families' religious beliefs and concerns while counselling and deciding for surgery.

Early vs late surgery

Timing for gender assignment

Few experts were of the view that gender assignment should be done as early as possible. But pediatric surgeons were of the opinion that it is highly dependent upon type of the defects, parental wishes and cultural background. Considering these facts institutional protocol of performing early surgery where indicated should be devised.

Gynecologist's opinion was that the genital surgeries can be delayed until the stage for menarche or till the sexual activity is reached as early surgeries may lead to scarring and vaginal stenosis.

Theme III: Challenges in management of DSD in Pakistan

Developing awareness (society and health care providers)

There is a lack of awareness among families about the medical nature and the various treatment options available for DSD. Even primary care provider don't know the appropriate management. Raising awareness by support groups for public and by professional societies and key opinion leaders for practitioners can create better understanding.

Need of capacity building in management of DSD

Another important aspect identified was the need of having national DSD core groups with multidisciplinary teams at least at district level hospitals with liaison of these groups to tertiary level facilities. Developing a good referral chain can be a strategy to mitigate this challenge.

Theme IV: Effect of covid pandemic on elective surgeries of DSD

During Covid pandemics SOPs, elective surgeries were limited in order to use the resources on the Covid patient as well as reducing the exposure of health care workers. Genital reconstructive surgeries are elective procedures and deferring them till pandemic is over, has led to a significant backlog. For parents and health care providers these are psycho- social and cultural emergencies and need to be done on priority basis to lessen misery of patients and families.

Discussion

DSD, a social as well as medical emergency, always needs urgent diagnosis and management. Majority of these children are diagnosed late or remained un-diagnosed in Pakistan like other developing countries. Hormonal assays, karyotyping and imaging facilities are available in a very few centers.

Rational investigations are mandatory in a newborn with DSD to make a proper diagnosis and provide a direction for future management. Use of advanced genetic testing like next generation sequencing, whole exome sequencing and targeted CGH array after karyotyping aids in a molecular diagnosis [5]. This will permit more rational sex assignment, recognizing the natural history of the identified cause of DSD [6]. Pakistan is still far from this molecular diagnostic facility, though genetic disorders are quite prevalent here. Besides improving basic health needs we need to develop our own molecular and genetic test facilities.

The management of DSD depends upon many factors and needs to be individualized based on the type of the disorder, gonadal differentiation, age at presentation, economic factors, psychosocial and cultural values [7].

The major issue at neonatal age is assigning sex-of rearing, which determines subsequent social dynamics, trajectories, surgical interventions, and often lifelong sex hormone administration. It also affects psychosocial and psychosexual developmental ramifications for the child. Instead of chromosomal sex, position the gonads, internal and external genitalia determine the sex of rearing. It is technically easier to construct female genitalia as compared to male. However, in a country with strong socio-cultural compulsions towards male, a considerate decision is required before assigning the sex [8,9].

The principle of autonomy dictates that the patient should have ultimate authority and should be kept on board, but it is often not possible for a child. Therefore, the medical team must identify the appropriate surrogate decision-makers including parents and legal guardians. In addition, a multidisciplinary DSD team and mental health worker should provide objective information to help the legal guardians in making final decision [10] to let the child live normal healthy life [11]. These patients do not feel comfortable in romantic/sexual relationships because of their insecurity regarding their physique and genitalia. They have poor inter- personal relations because of their internal complexes as well as because of bullying by peers. Moreover, the children with late presentation face different dilemmas as disclosing about change of sex at older age results in embarrassment and low self-esteem [12].

Religious concepts also affect the decision-making process in gender assignment. According to Islamic Jurisprudence, many Islamic rituals, rights, or obligations are gender oriented [13]. Therefore, Muslim patients with DSDs and their families need to be counseled on

both the societal as well as religious implications of gender assignment. However, in some Muslim countries like Malaysia, sex of rearing is based on the external appearance of these patients [13,14].

The role and timing of surgical intervention is the most debated part of management of DSD and still debatable aspect since 2006 consensus guidelines [15]. Proponents of early surgery believe it helps the DSD patients to confirm their assigned sex and leads to better quality of the genital tissues, better vascularization secondary to postnatal maternal estrogens. The Pediatric Surgeons in this panel discussion were also of the same opinion.

The gynecologists among panelists were the strong proponents of delayed surgical intervention especially in girls with CAH, suggest feminizing genitoplasty to be deferred till puberty as according to them, vaginoplasty needs regular dilatation. However, clitoroplasty can be done in the early age.

In a case study on two similar patients who had contrasting decisions regarding sex assignment at birth and had different outcomes at adulthood. This signifies that gender assignment is not a simple phenomenon based on parental wishes and social acceptance but it's a complex one [16].

Pakistan like other Low and Middle Income Countries (LMIC) is facing the challenge of disparity in provision of healthcare facilities among different social strata. In cases of DSD the socioeconomic class and poverty is an even bigger challenge. Denial to accept this social taboo of DSD associated with these challenges make them to seek either no traditional incompetent healer.

The panelists in this study unanimously agreed for DSD advocacy and the need for increasing awareness among public and healthcare professionals. Kalra, *et al.* and Kraft, *et al.* also emphasized to educate public and physicians for the medical and psychological needs of patients with DSD [17,18].

In Pakistan, developing a high-quality training and mentorship program at few centers to train dedicated physicians and surgeons with skills for compassionate care is the need of time.

The panelists felt a significant need for dedicated multidisciplinary clinics and DSD boards (in person or virtual) to achieve better outcomes. It was suggested to reallocate resources for stepwise approach at the primary and advanced level. The training support for interested consultants from institutional level as well as through professional bodies is considered the way forward for having competent human resources. International support can also be sought for capacity building to meet the unmet needs [19,20] like Global surgery partnership including international visiting experts, outreach partnership programs between LMIC and HIC and fellowship and residency training [21].

The surgical management of DSD patients demands a lot of discussion and planning before, any intervention which in itself is a time-consuming process but once it is decided undue delay must be avoided as it is cumbersome for patient and the family.

During COVID-19 pandemic, deferring genital reconstructive surgeries profoundly effected management of DSD patients [22,23]. This results in psychological stress for patients and their families and increased the backlog for surgeon. Although DSD is not potentially a life-threatening condition, it certainly needs management on a priority basis.

Conclusion

The mainstay of this panel discussion was to identify the issues in the management of DSD patients and to develop a collaborative approach for long term management. It was unanimously decided to take all the stakeholders under consideration including the effected

patient and family, pediatric endocrinologist, pediatric Surgeon, pediatric Psychiatrist and gynecologist. We as clinicians may also develop more human resource and train and develop expertise in ethical decision-making process in collaboration as a multidisciplinary team.

Author's Contributions

IA and HY contributed to generating themes and writing manuscript. NZ, NT, TB and MI contribution in designing and writing the manuscript. TB and IA also reviewed the manuscript and finalized the report. SY and FS contributed in writing their verbatim part in the panel discussion.

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