Quality of Life, Perceived Stigma, and Challenges Faced by the Parents of Girls with Congenital Adrenal Hyperplasia

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ABSTRACT

Objective: To assess the quality of life (QoL), perceived stigma, and extent of anxiety and/or depression among parents of female children with congenital adrenal hyperplasia (CAH).

Study Design: Observational cross-sectional study.

Place and Duration of the Study: Department of Paediatric Surgery and Endocrinology Clinics of National Institute of Child Health (NICH), Karachi, Pakistan, from June to December 2022.

Methodology: Parents of the female children diagnosed with CAH were enrolled. A validated parent self-report questionnaire, parent-focused items of the experiences and reactions questionnaire used for assessing disorders of sex development (DSD)-related experienced or anticipated stigma and the patient health questionnaire-4 (PHQ-4) were used to gather the information. The scores were calculated for each item and compared among mothers and fathers. Independent samples student t-test was used to find any significant differences between the mean scores. A value of $p < 0.05$ was considered statistically significant.

Results: Out of the 98 parents, there were 74 (75.5%) mothers and 24 (24.5%) fathers. The score recorded in all three tools showed multiple challenges that had a negative impact on parents. In QoL assessment for experiences and reactions, scores between mothers and fathers were similar. In the PHQ-4 tool, the scores were significantly related to the female gender of the parents as anxiety and depression were more reported in mothers ($p = 0.046$). In parents of children >7 years of age, the differences in scores were statistically significant for the domains such as talking to others, and stigma.

Conclusion: The parental QoL was affected resulting in limited social interactions with the feeling of being stigmatised. Many reported significant anxiety and/or depression.

Key Words: *Parental anxiety, Congenital adrenal hyperplasia, Disorders of sex development, Gender assignment, Gender.*

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INTRODUCTION

Disorders of sex development (DSD) include several conditions where gender assignment at birth remains challenging.[1] Lack of awareness on the part of healthcare providers and parents adds to the delay in diagnosing these children.[2] Gender assignment issues create a social dilemma.[3] In congenital adrenal hyperplasia (CAH), the genetically female newborns are exposed to the excess of androgen hormones *in-utero*. This results in variable degrees of masculinisation of the external genitalia in females.[4]

The first question usually asked at the birth of a newborn is about the sex of the baby. However, for many parents, this is the beginning of a life-long psychological torment.[5] In the genetically female babies who are severely masculinised at birth, a male gender is usually assigned by the parents. The CAH children who later identify themselves as females report poor quality of life (QoL).[6,7] The rearing of female children with CAH in whom gender is re-assigned later in life produces significant psychological trauma and stigma to the parents.[8,9] Multiple psycho-social issues may compromise the well-being of the parents as well.

In liberal societies, gender-related social issues are discussed openly. However, in many conservative communities, such as Pakistan, parents are inhibited or hide such issues because of various social and cultural taboos. This may culminate in anxiety and lead to mental health issues among the parents of these children.[10] Therefore, it is important to understand the perspectives of the parents and evaluate their well-being. This study was conducted to assess the QoL, presence of anxiety and/or depression among the parents of female children born with CAH.

METHODOLOGY

This observational cross-sectional study was conducted from June

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to December 2022 at the National Institute of Child Health (NICH), Karachi, Pakistan. The Institutional Review Board approved the research proposal (No: IERB-04/2022). Parents of the female children diagnosed as 46, XX-DSD-CAH, were enrolled from the Paediatric Surgery and Endocrine Clinics of the hospital. The other DSDs and males with CAH were excluded. The purpose of the research was explained to the parents and informed consent was taken.

The sample size was calculated using OpenEpi version 3. Taking the estimated reporting of psychosocial health disorder in parents of 46, XX-DSD children as 14.6%,[11] a margin of error of 7%, and a confidence level of 95%, the sample was calculated as 98. A validated tool, the parent self-report (PSR) tool, was used for this study.[12] This tool is comprised of eight components (healthcare communication and information, talking to others, future concerns, medication, clinic visits, surgery, stigma, and anxiety and depression). These items were taken from a previously validated PSR questionnaire, parent-focused items of the experiences and reactions questionnaire assessing DSD-related experience or anticipated stigma and the patient health questionnaire-4 (PHQ-4) which is used as a screening scale for measuring depression and/or anxiety.[13-15]

The PSR scoring was done by using a 5-point Likert scale starting from "totally agree" with score of 1 to "totally disagree" with a score of 5. The higher scores indicated better QoL and positive adaptation. The experiences and reactions (stigma) domain was scored on a 5-point Likert scale. The higher scores indicated a greater level of experience (or anticipated) stigma. The scores calculated for the PHQ-4 screening scale for anxiety and/or depression were labelled as normal (0-2), mild (3-5), moderate (6-8), and severe (9-12).

A face-to-face meeting with the parents, at their ease, during a scheduled visit to the endocrine outpatient clinic was arranged. A single family member was approached in one session. Parents were informed about the tool that was used for the assessment of their psychosocial health. A separate room was booked in the institution to maintain the privacy of the individuals. Parents were also informed about the confidentiality of the data obtained from the study. The questionnaire was filled out in the presence of the investigator. Any difficulty in understanding the statements of the tool used was clarified. The tool is in easy language meant for laypersons. Those individuals who were assessed as having anxiety and/or depression were facilitated for psychological consultation.

Data were entered and analysed using the Statistical Package for the Social Sciences (SPSS) for Windows version 23.0 (IBM Corp., Armonk, New York). Frequency and percentages were calculated for categorical variables such as gender and mean and standard deviation (SD) were reported for continuous variables such as age. Parents' health-related QoL, parents' report of stigma, and patient health questionnaire scores were compared for each parent (either father or mother) using independent samples Student's t-test. The ages of the female children with CAH were categorised as from birth to 90 months and from 91 months to 180 months. The scores calculated for the parents were compared between the two age groups of the children, considering that by the age of seven years, the child understands the biological sex and has a self-perceived gender identity. At this age, parents' stress was expected to increase further. A p-value of ≤0.05 was taken as significant.

RESULTS

In this study, 98 parents (either mother or father) of the children with CAH were included. The total number of mothers and fathers was 74 (75.5%) and 24 (24.5%), respectively. The mean age of the children with CAH was 61.77 ± 47.01 months (minimum of 3 months to maximum of 180 months). In the QoL score, future concerns showed a combined mean score of 11.04 ± 4.39 out of 35. Similarly, in the domain of medications, the mean score was 6.79 ± 2.66 out of 20. This highlighted the low adaptation of parents in the above-mentioned components. The mean score on the PSR tool for all the mothers and fathers separately is given in Table I. In QoL assessment for experiences and reactions, scores did not show any significant difference between mothers and fathers in any of the domains. The PHQ-4 scores were significantly related to the gender of the parents. Of the total, 52 (53%) parents suffered from some degree of anxiety and depression, of which, 12 were fathers and 40 were mothers (p = 0.046).

Table I: Parent Self-Report (PSR) and Patient Health Questionnaire (PHQ) scores and their association with the gender of the parents.

Parent Self-Report	**Items (Maximum score)**	**Domains derived from**	**Mean Scores**			**p-value**
			Fathers	**Mothers**	**Total sample**	
Communication and information	2 (10)	QOL-DSD	4.04 ± 2.59	3.95 ± 2.32	3.97 ± 2.38	0.865
Talking to others	5 (25)	QOL-DSD	15.46 ± 5.82	14.70 ± 5.80	14.89 ± 5.78	0.581
Future concerns	7 (35)	QOL-DSD	10.75 ± 5.16	11.14 ± 4.01	11.04 ± 4.39	0.705
Medication	4 (20)	QOL-DSD	6.67 ± 3.04	6.82 ± 2.55	6.79 ± 2.66	0.803
Clinic visit	4 (24)	QOL-DSD	11.50 ± 1.58	12.15 ± 1.87	11.99 ± 1.82	0.131
Surgery	4 (25)	QOL-DSD	22.37 ± 4.89	22.76 ± 9.28	22.66 ± 8.4	0.848
Experiences and reactions						
Stigma	10 (50)	Experiences and reactions: Parent focus	22.79 ± 9.65	21.24 ± 6.830	21.62 ± 7.59	0.388
Patient Health Questionnaire-4						
PHQ-4, mean ± SD	3 (12)	Patient health questionnaire-4	3.46 ± 2.97	2.85 ± 1.71	3.00 ± 2.09	0.219
Level of anxiety and depression among parents			**Fathers n (%)**	**Mothers n (%)**		
Normal (Score - 0-2), (n = 46 - 46.9%)			12 (50.0%)	34 (45.9%)		0.046*
Mild (Score - 3-5), (n = 45 - 45.9%)			10 (41.7%)	35 (47.3%)		
Moderate (Score - 6-8), (n = 5 - 5.2%)			0	5 (6.8%)		
Severe (Score - 9-12), (n = 2 - 2.0%)			2 (8.3%)	0		

**Significant by Independent Samples Student's t-test.*

Table II: Association of age of the congenital adrenal hyperplasia (CAH) children with the Parent Self-Report (PSR) tool and Patient Health

Questionnaire (PHQ) scores of their parents.

Questionnaire domains	Age groups		p-value
	0-90 months	91-180 months	
Parent Self-Report mean ± SD			
Communication and innovation	4.07 ± 2.50	3.70 ± 2.05	0.329
Talking to others	14.76 ± 5.30	15.22 ± 7.00	0.013*
Future concerns	11.39 ± 4.46	10.11 ± 3.74	0.787
Medications	6.86 ± 2.55	6.59 ± 2.97	0.095
Clinic visit	12.17 ± 1.94	11.52 ± 1.39	0.045*
Surgery	24.62 ± 8.10	17.52 ± 6.99	0.006*
Experiences and reactions			
Stigma	22.20 ± 8.45	20.11 ± 4.41	<0.001*
Patient Health Questionnaire-4	**n (%)**	**n (%)**	
Normal (0-2)	33 (46.5%)	13 (48.2%)	0.758
Mild (3-5)	33 (46.5%)	12 (44.4%)	
Moderate (6-8)	3 (4.2%)	2 (7.4%)	
Severe (9-12)	2 (2.8%)	0	

**Significant by Independent samples Student's t-test.*

According to the age groups, there were 71 (72.4%) children between 0-90 months and 27 (27.6%) between 91-180 months of age. On PSR score, the differences in scores were statistically significant for the domains of talking to others, clinic visits, surgery, and stigma. Details are given in Table II.

DISCUSSION

The present study assessed the psychosocial health of the parents of female children with CAH where gender assignment was an issue. The study found variable degrees of anxiety and depression among parents, who were concerned about the future of their children having male-like physical growth. Disclosing the child's condition to others leads to the stigma and fear of being socially ostracised. This, they believed, would shield the children from potential and actual discrimination and stigmatisation. Similar concerns were reported in another study.[16]

More than half of the parents of young children and early teenagers experienced significant anxiety and/or depression. A similar pattern was reported in a study from Sri Lanka among parents of children with CAH where 59% had symptoms of depression.[17] However, in a study from 12 DSD centres across the United States, only 14% of parents reported clinically significant anxiety.[11] It may be inferred that people from the global north may have different attitudes toward patients with DSD than those from the global south. In societies where gender-related issues are discussed openly with the social support rendered by voluntary organisations, parents may find it easy to express their thoughts. In a study by Suorsa *et al*., the extent of distress that parents of DSD children experienced was akin to that of the parents whose children had a diagnosis of cancer. It is reported that 15.6% parents experienced moderate-to-severe anxiety and 18% had symptoms of depression.[18]

The gender segregated studies have reported a higher prevalence of depressive symptoms and lowered stress thresholds in women in general. Women have disproportionately more chances to suffer from stress.[19] However, in this study fathers of the children with DSD also reported stress and anxiety which was solely due to the gender-related issues of their children. The results of the current study showed that parents of older children were significantly worried about surgical procedures and clinic visits. They reported more future concerns as compared to the parents of younger children with CAH who were significantly stressed about the stigma and social interactions with peers and adults. Stigma was a significant ongoing psychological stressor that was experienced by 68% of the parents as reported in a study.[20] This is because of multiple misconceptions about the disease. Dayner *et al.* have emphasised the importance of providing accurate and comprehensible information to the parents.[21] Gender is perceived differently in many cultures and the gender of rearing once assigned at birth is hard to change at a later age. Many legal issues are also faced by the parents. These were expressed in the present study.

Many parents felt that they were responsible for the condition of their child, a type of self-blame. Some felt isolated from their relatives and few intentionally avoided visiting them or inviting others to their homes. Self-created social isolation precipitates anxiety. In a closely knitted joint family system, it was difficult for many parents to justify frequent hospital visits as they did not disclose the condition to others. Social interactions and relationships were thus affected.

The need for surgical interventions and life-long medical management of children with CAH leads to feelings of desperation and at times, inadequacy when they were not able to find support. The stigma surrounding the condition persists, presenting challenges for parents as they strive to provide the best possible care and support for their children.[20] Kraft in her doctoral thesis proposed a psychological intervention to circumvent the effect of psychosocial stressors on parents. This was comprised of psychoeducation, problem-solving activity, coping skills exercises, and social support. The intervention was aimed at empowering families to understand and manage CAH and its effects on family dynamics, communication, and emotional well-being.[22] This model can be replicated in low-resource settings.

Stigma and discrimination framework may provide a comprehensive approach to understanding and addressing the health-related stigmas that are associated with different diseases that affect mental health including chronic conditions. Educating laypeople and creating awareness in society about people born with DSD are important. However, it may be done cautiously depending upon the cultural sensitivities and religious beliefs. The complexity of individuals' realities and a responsive attitude of the people may mitigate the negative psychological impact and improve the QoL of parents as well as CAH children.[23]

This study provided substantial data about a sensitive and less-discussed issue, particularly in the context of patriarchal societies, where gender is perceived as binary. The challenges faced by the parents are equally important, especially when peers may not understand the nature of the condition.

The limitation of the study is its origin from a single institution in an urban setting. However, the strength includes the use of different validated tools to provide more comprehensive information. A qualitative study with open-ended questions as well as from parents of different geographical locations may provide deeper insight about the challenges that are faced by them and how those affect their well-being.

CONCLUSION

Parents of female children with CAH experienced clinically significant anxiety and/or depression. Mothers were more prone to the psychological distress as compared to fathers. Parents of the older children were worried significantly about the surgical procedures and clinic visits. Parents with children younger than seven years of age showed significant stress over stigma and social interactions.

ETHICAL APPROVAL:

The study was approved by the Institutional Review Board of the National Institute of Child Health, Karachi (IERB-04/2022).

PATIENTS' CONSENT:

Parents of the patients were informed about the details of the study and consent was taken to participate.

COMPETING INTEREST:

The authors declared no conflict of interest.

AUTHORS' CONTRIBUTION:

MM: Concept, study design, data collection and analysis, manuscript writing, and revision.
NZ: Conception and manuscript writing.
MNI: Manuscript writing.
JA: Conception, data analysis, manuscript writing, and revision.
All authors approved the final version of the manuscript to be published and are responsible for its content.

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