

QUALITY OF LIFE IN CHILDREN AND ADOLESCENTS WITH CONGENITAL ADRENAL HYPERPLASIA

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ABSTRACT

OBJECTIVE

To assess the Quality of Life of children and adolescents with Congenital Adrenal Hyperplasia (CAH).

STUDY DESIGN

Descriptive cross-sectional study.

PLACE & DURATION OF STUDY

It was conducted at NICH, Karachi and was six months in duration, from November 2019 to April 2020.

METHOD

We enrolled all children and adolescents in the range of 8 to 18 years of age diagnosed with CAH. The World Health Organization Quality of Life-BREF (QOL-BREF) instrument was used for data collection. Four health domains, including physical, psychological, social, and environmental areas, were assessed, with higher transformed scores indicating better QOL. Data was entered on SPSS Version 23.

RESULTS

Our study population comprised of 30 children and adolescents with a mean age of 12.75 ± 3.23 years. Mean transformed scores of physical, psychological, social, and environmental health domains were 31.37 ± 12.15 , 45.13 ± 12.50 , 34.8 ± 20.49 and 37.6 ± 12.06 respectively.

CONCLUSION

The quality-of-life score in our population was relatively low in the physical and social domains of health as compared to psychological and environmental health domains. Larger community-based studies are required to further elaborate the causes and remedies of poor quality of life in children and adolescents.

KEYWORDS

Congenital Adrenal Hyperplasia (CAH), Quality of life, Endocrine disorder, Children and Adolescents.

INTRODUCTION

Congenital Adrenal Hyperplasia (CAH) is an autosomal recessive disorder predominantly caused by a defective gene that leads to a deficiency of enzymes necessary for the synthesis of hormones from cholesterol in the adrenal gland, particularly the adrenal cortex. This deficiency results in the overproduction of androgens, which can stimulate virilisation in both genders and may manifest before or after the onset of puberty.¹

Adolescence represents a transitional phase of life between childhood and adulthood, characterised by significant development in biological, psychological, physiological, and social domains. These factors collectively contribute to the formation of a positive body image, identity, and sexual attraction, which in turn reinforce one's psychological self-perception.² Research indicates that during this critical period, individuals with CAH may experience a progressive weakening of their psychological self-image, leading to a decrease in self-confidence, interpersonal challenges, and ultimately, a reduction in self-esteem.³

In the developing world, the birth of a child with developmental anomalies, including atypical pubertal progression, introduces complex biological and social challenges that permeate all aspects of life. Concerns regarding gender incongruence can provoke fear of unwarranted social scrutiny and discrimination from relatives, peers, and healthcare professionals, thereby significantly diminishing the quality of life for the affected individual.⁴

Quality of life is conceptualised as the subjective experience of a fulfilling physical, psychological, and social existence. It encompasses satisfaction with one's well-being, functional capacity, productivity, societal participation, and interpersonal relationship stability. Recent studies have shown that these aspects may be adversely impacted in individuals facing challenges related to the development of sexual identity.⁵

Data suggest that among the global youth population, one in four individuals fall within the 10–24 year age bracket,⁶ with 90% residing in low- to middle-income countries.⁷ They are disproportionately influenced by socio-cultural, economic,

and environmental risk factors due to limited resources, in view the literature related to significance of religious increased familial obligations, and an absence of supportive systems. However, most of the published literature on the quality of life for adolescents has been published in developed countries. This disparity underscores the necessity to understand the unique life challenges of youth with CAH.⁸

Evaluating the quality of life for children with CAH is essential for highlighting the importance of assessing the psychosocial domain in paediatric and adolescent populations, particularly within lower-middle-income contexts. By identifying their specific difficulties, future psychological interventions can be strategically implemented.

METHOD

A descriptive cross-sectional study was conducted at the outpatient department for children and adolescents at the National Institute of Child Health (NICH) in Karachi, Pakistan. NICH is renowned as one of the oldest and largest public sector teaching hospitals in Karachi, offering a wide range of services across various specialties, including Paediatric Neurology, Infectious Diseases, Paediatric Surgery, Paediatric Medicine, and Paediatric Endocrinology.

This study was conducted over six months, from November 2019 to April 2020. After approval from the Institutional Ethical Review Board of NICH (IERB No: 26/2019), written informed assent or consent was obtained from the caregivers of participating children and adolescents, as applicable, which included permission to publish the study results. We enrolled all children and adolescents aged 08 to 18 years diagnosed with Congenital Adrenal Hyperplasia (CAH) before the age of 8. Exclusion criteria included participants with concomitant chronic diseases such as Diabetes, Tuberculosis, or disorders of sexual development. A non-probability consecutive sampling technique was used for subject recruitment. Demographic and other relevant details of study subjects and their parents were collected on a semi-structured proforma developed for the purpose of this study. The World Health Organisation Quality of Life Brief Version (WHOQOL-BREF) was employed for data collection after obtaining permission from WHO and translating it into Urdu. The WHOQOL-BREF is a self-administered questionnaire on the individual's perceptions of their health and well-being over the previous two weeks. The scale comprises 26 questions, covering physical, psychological, social, and environmental health domains. Raw scores for these domains were calculated. The raw score of each domain was converted into a transformed score. Higher transformed scores indicate a better quality of life. The data were entered into a statistical package for Social Sciences (SPSS) Version 23. Frequencies and percentages were calculated for categorical variables, while mean and standard deviations were calculated for quantitative variables.

RESULTS

Our study population comprised 30 children and adolescents with a mean age of 12.75 ±3.23 years, the minimum being 08 years, while the maximum age of the subjects was 18 years. Mean weight and height were 37.38 kg and 122.55 CM respectively. According to education status, most of the parents were literate (66.7%), while most of the children and adolescents had primary education. Dominant languages spoken at home were Urdu and Sindhi, i.e., 23.33% of total subjects speaking each. Almost an equal number of participants belonged to nuclear and joint families. Most parents (96.7%) had consanguineous marriages (Table 1).

Table 1
Demographic Characteristics of Study Participants

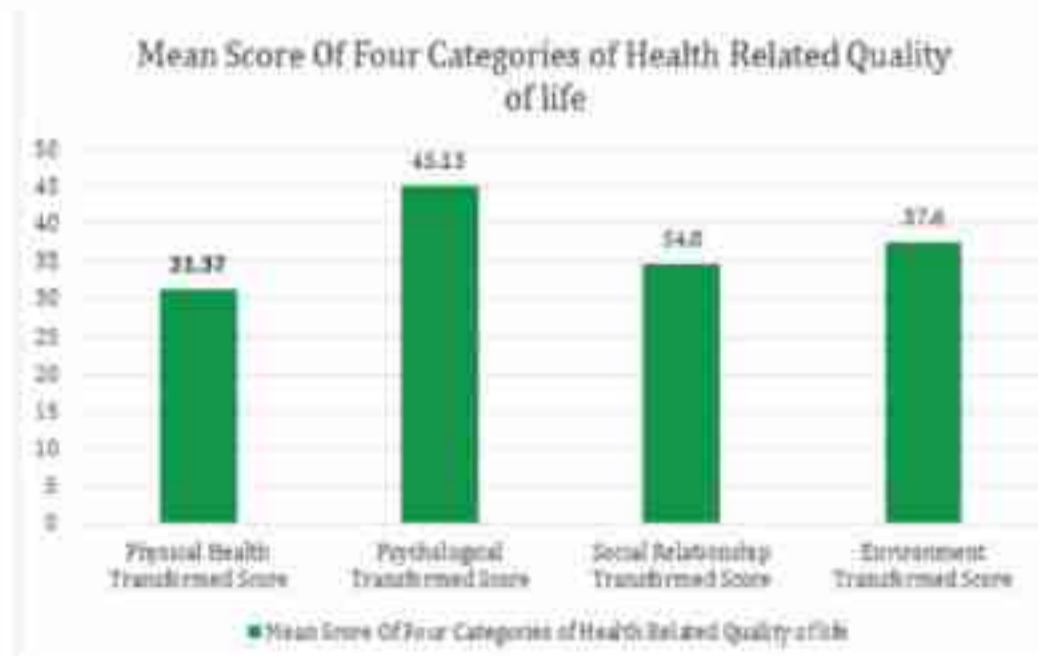
Variable	Frequency N-30	Percentage
Gender		
Male	17	56
Female	13	44
Education Status of Father		
Illiterate	10	33.33
Madrassa	4	13.3
Primary	4	13.3
Matric	6	20.0
Inter	4	13.3
Graduation and above	2	6.7
Education Status of Mother		
Illiterate	7	23.33
Madrassa	7	23.33
Primary	7	23.33
Matric	4	13.3
Inter	2	6.7
Graduation and above	3	10.0
Education status of Children & Adolescents		
Illiterate	02	6.66
Madrassa	00	00
Primary	18	60
Matric	08	26.66
Inter	01	3.33
Graduation and above	01	3.33
Language spoken at home		
Urdu	7	23.3
Sindhi	7	23.3
Punjabi	4	13.3
Baluchi	1	3.3
Pushto	5	16.7
Others	6	20.0
Family type		
Nuclear	15	50.00
Joint	15	50.00
Consanguineous marriage		
Yes	29	96.7
No	01	03.3

Mean transformed scores of physical, psychological, social, and environmental health domains were 31.37 ± 12.15 , 45.13 ± 12.50 , 34.8 ± 20.49 and 37.6 ± 12.06 respectively. The highest transformed score (45.13) was observed in psychological health followed by environmental health (37.6) and social health (34.8) while the least transformed score (31.37) was observed in the physical health domain. (Table 2, Figure 1).

Table 2
Transformed Quality of life Scores

Variable	Mean	Standard Deviation	Range	Minimum	Maximum
Physical Health	31.37	12.150	50	6	56
Psychological	45.13	12.503	44	31	75
Social Relationships	34.80	20.495	94	6	100
Environmental	37.60	12.065	50	19	69

Figure 1
Mean Score of Four Categories of Health Related Quality of Life



DISCUSSION

Our research highlights that children and adolescents with CAH in Pakistan exhibit lower scores across all domains of the WHOQOL-BREF questionnaire compared to their counterparts in developed nations from countries like France, Germany, the Netherlands, Sweden, and the UK.⁹ Specifically, psychological well-being emerges as relatively less affected amongst the four health-related quality of life domains. While scores are lower as compared to CAH children in developed countries, psychological well-being remains comparatively higher within our study population.¹⁰

High adherence to treatment regimens and younger age significantly correlate with improved quality of life among participants diagnosed with congenital adrenal hyperplasia (CAH) in the study conducted in developed countries like Sweden.¹¹ A study conducted in India revealed that the quality of life was found to be significantly reduced in all domains, just like our study. Additionally, their study reported reduced quality of social and school functioning which was not analysed in our study.¹⁷

Our study underscores the prevalence of lower levels of parental education among our population, reflecting a

disparity compared to more developed nations. Consanguinity Emerges as a significant risk factor for congenital hyperplasia,¹² further complicating the health landscape for affected children and adolescents. Addressing these socio-demographic factors is paramount in optimising the quality of life and holistic well-being of children and adolescents living with CAH. Previous research indicates a proportional increase in congenital adrenal hyperplasia (CAH) rates with consanguinity, notably prominent in Arab and Indian societies. Our study validates these findings, revealing a high prevalence of consanguinity (97%) within our study cohort.¹⁴

Equitable healthcare demands clinical treatment and psychosocial support for optimal care in congenital adrenal hyperplasia (CAH). Holistic approaches addressing both biochemical and psychosocial aspects enhance children & adolescents' quality of life. Regular psychosocial assessment is crucial to understanding its impact on functioning. Health-related quality of life questionnaires offer valuable insights, promoting collaborative partnerships and children and adolescents' empowerment in their healthcare journey. An Indian study found that congenital adrenal hyperplasia had a collective prevalence of 1 in 5762 births, revealing regional differences. They emphasised the significance of newborn screening for national programs to mitigate morbidity and mortality.¹⁵ However, a Pakistani study reported challenges with ambiguous genitalia in CAH children & adolescents and focused on the necessity for a disease registry and neonatal screening integration for early intervention and improved outcomes.¹⁸

Strengths

The study focuses on children and adolescents with CAH, which is an underserved population in research. It is conducted in a low-middle-income country, highlighting challenges specific to this population, which can lead to context-specific interventions. The study instrument is a globally recognized, validated tool, ensuring reliable measurement of quality of life. It examines multiple domains (physical, psychological, social, and environmental), providing a comprehensive view of the impact of CAH on quality of life.

Limitations

The study includes only 30 participants, limiting the generalizability of findings to broader populations. While it provides a snapshot of Quality of Life at a single time point, it lacks longitudinal insight into how Congenital Adrenal Hyperplasia impacts QoL over time. It lacks direct comparisons with healthy controls or populations from different regions. Non-probability consecutive sampling may introduce selection bias, as those seeking care at NICH may not represent the broader population of CAH patients.

Recommendations

Larger community-based studies are required to elaborate on the causes and remedies of poor quality of life in children and adolescents.

CONCLUSION

This study highlights a disparity in quality-of-life scores, with lower scores in physical and social health domains compared to psychological and environmental domains in children and adolescents with Congenital Adrenal Hyperplasia. To address this, a multidisciplinary approach is recommended. Enhanced collaboration between pediatricians and child and adolescent mental health professionals can provide crucial psychosocial support for these children and families. Ongoing liaison can be facilitated by workshops and joint academic sessions. Additionally, community-based awareness campaigns targeting caregivers, educators, and the public could promote a holistic understanding of child well-being.

CONFLICT OF INTEREST

Authors declared no conflict of interest.

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None

DISCLOSURE

None

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AUTHOR(S) CONTRIBUTION / UNDERTAKING FORM

No.	Author Name	Author Affiliation	Contribution
1	Dr. Fawad Suleman	Department of Psychiatry, Aga Khan University, Karachi.	concept mapping, data analysis and interpretation of study findings, critically reviewed the manuscript
2	Dr. Nooraba Noor	Postgraduate trainee Paediatric Medicine, National Institute of Child Health, Karachi, Pakistan	contributed to literature search of the study, worked on the initial draft and critically reviewed the manuscript
3	Dr. Samiya Iqbal	Department of Psychiatry, Aga Khan University, Karachi.	contributed to literature search of the study, worked on the initial draft and critically reviewed the manuscript
4	Dr. Zainab Sher	Department of Psychiatry, Aga Khan University, Karachi.	worked on the initial draft of the manuscript
5	Dr. Mohsina Noor	National Institute of Child Health, Karachi, Pakistan	contributed to methodology of the study, critically reviewed the manuscript

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