

Psychological Outcomes and Quality of Life of patients with non-CAH Disorders of Sex Development

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INTRODUCTION/ OBJECTIVES

- Evidence-based treatment of patients with disorders of sex development (DSD) is challenging given the paucity of longitudinal clinical outcome studies. Furthermore, studies have tend to focus on patients with congenital adrenal hyperplasia.^{1,2}
- The objectives of this study was to evaluate the **quality of life** and the **behavioural outcomes** of patients with **DSD other than congenital adrenal hyperplasia** and to identify factors that may influence the said outcomes.

METHODS

- The study population consisted of patients with DSD other than congenital adrenal hyperplasia aged between 6 and 18 years.
- Control data was obtained from representatives of the patient's siblings matched for age and gender.
- Study tools used were the Pediatric Quality of Life (PedQOL) questionnaire and the Child Behaviour Checklist (CBCL).
- Patients were grouped by karyotype and sex of rearing/recent gender.

RESULTS

1. Background Data

- Final number of subjects were 30
- Age at study entry : 12 [8 – 14] years

	46XY	46 XX	Total
Male Social sex (n)	21	2	23
Female Social Sex (n)	6	1	7

Figure 1 : Study population according to karyotype and social sex

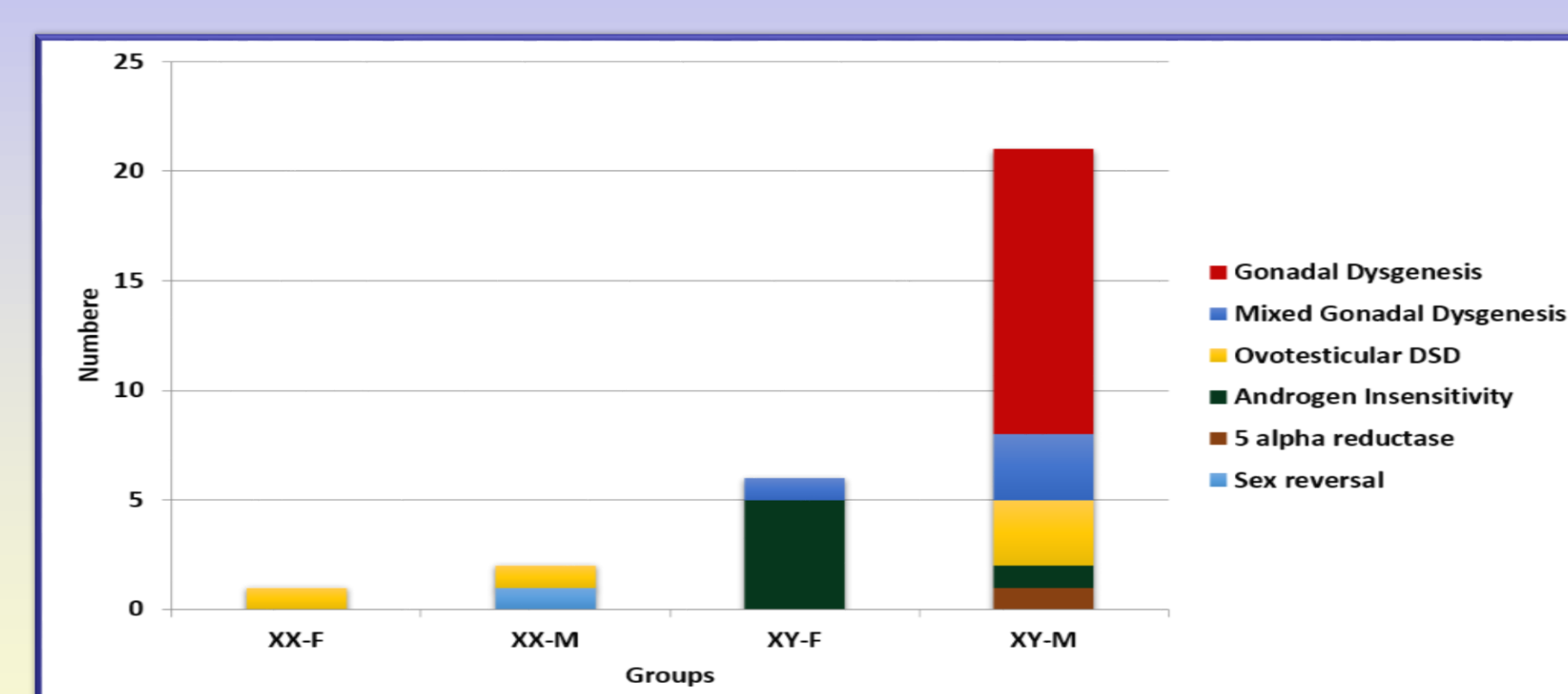


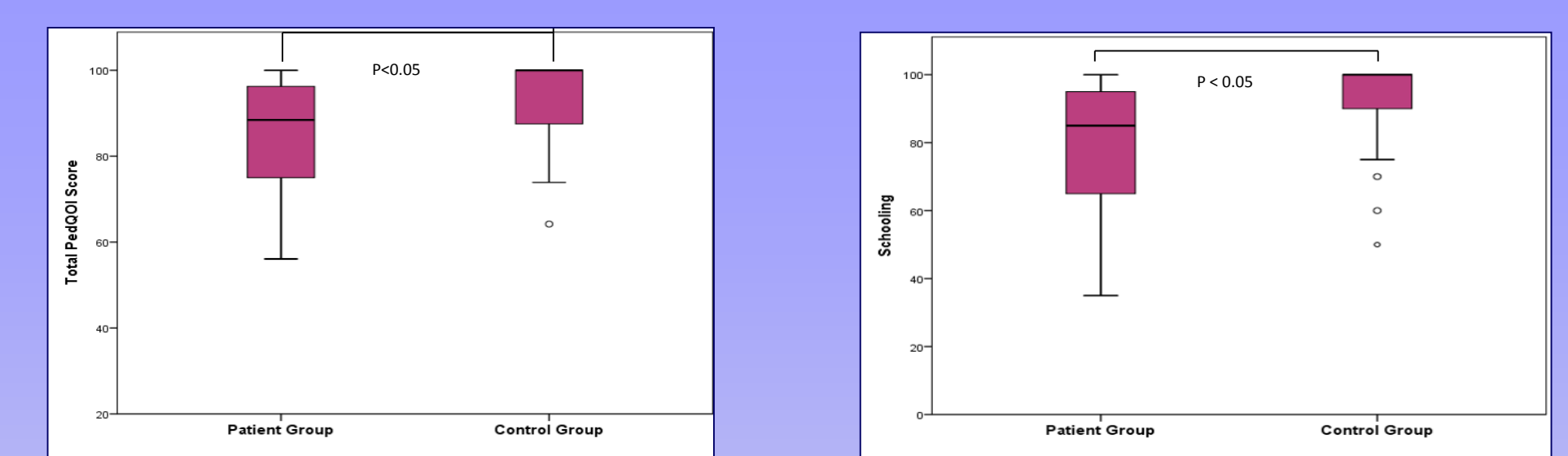
Figure 2 : Study population according to groups and diagnosis

- No significance difference between baseline characteristics of cases and control

2. PedQOL

	Case	Control	P
Physical	100[21.9]	100[6.2]	0.163
Emotional	90[32.5]	100[12.5]	0.063
Social	95[20]	100[5]	0.031
Schooling *	85[35]	100 [11.3]	0.003
Total *	90.6[24.2]	100[13.2]	0.004

Figure 3 : Table showing total and subscale scores (physical, emotional, social and schooling) from the PedQOL in the patient group as compared with the control group. Box plot showing the distribution of total and schooling scores between the patient and control group. Scores are given as median [IQR] with lower scores denoting poorer outcome. P value was adjusted using the Bonferroni-Holm method to account for multiple comparison. *Denotes significant variable.



- The total PedQOL score was significantly lower in the patient group than in the control group. In subgroup analysis this difference was reflected significantly in the schooling field.

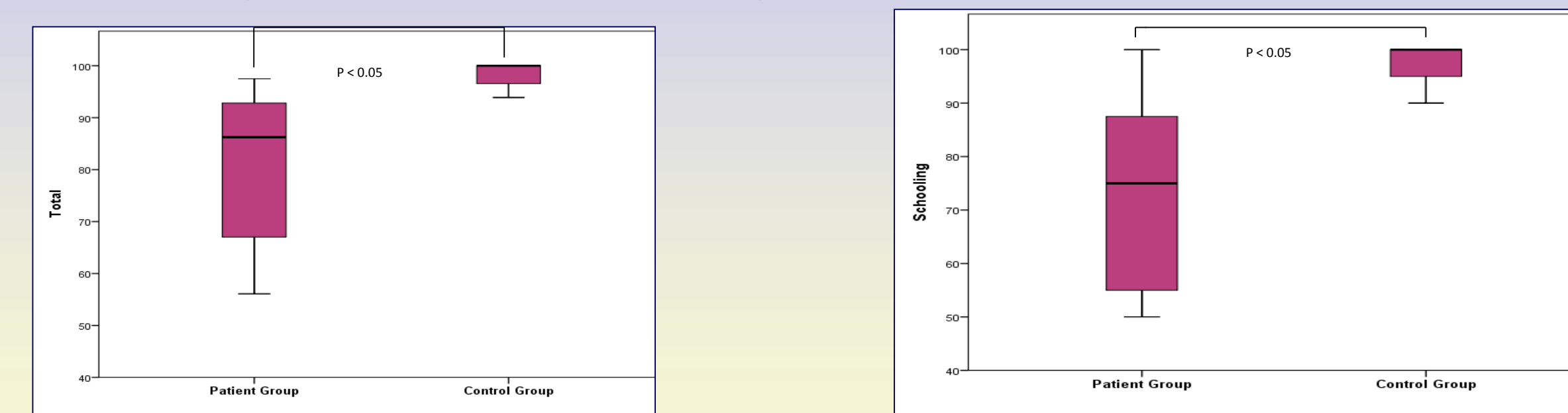


Figure 4 : Box plot showing the distribution of total and schooling scores between the female patient group and the female control group

- Upon further analysis the total PedQOL score was significantly lower in patients who had DSD who were of female social sex as compared to the controls who were females.

3. CBCL

	Case	Control	P
Internalizing Score*	50.7(12.6)	42.2(10.5)	0.007
Externalizing Score	50.5[16]	40[15]	0.026
Total	45.3(16.8)	38.5(11)	0.059

Figure 5 : Total, internalizing and externalizing scores from the CBCL in patients with disorders of sex development as compared to the control group. Scores are given as median [IQR] or mean (SD) with higher scores denoting poorer outcomes. P value was adjusted using the Bonferroni-Holm method to account for multiple comparison. *Denotes significant variable.

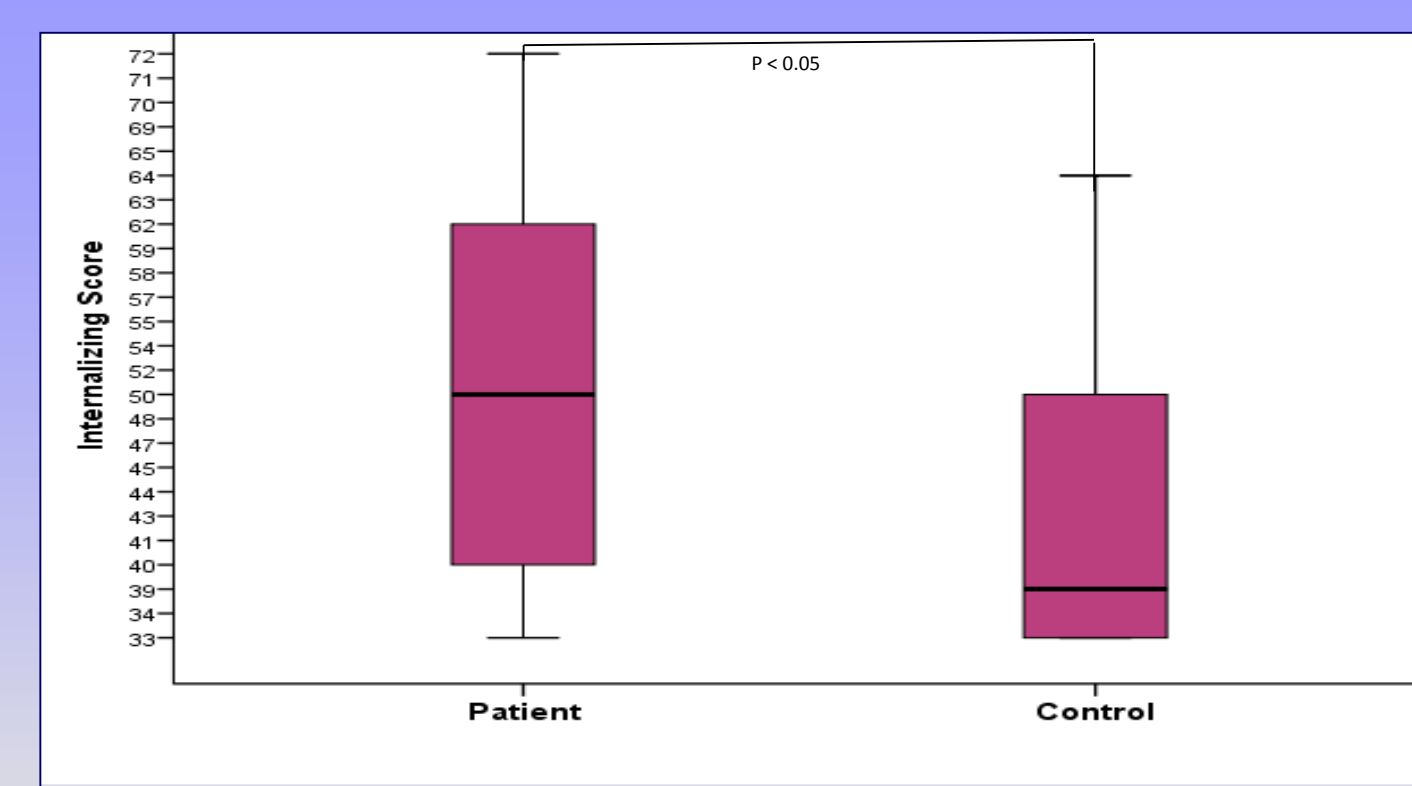


Figure 6 : Box plot showing the distribution of the internalizing scores between the patient and control group

- The internalizing score was significantly higher in the patient group (more pathological).
- Subgroup analysis revealed that the patient group had more affective, anxiety and conduct problems.
- No significant correlation between these factors and CBCL or PedQOL scores : Family income, number of surgeries, degree of virilisation (subgroup 46XY-F and 46XY-M) and mode of puberty (spontaneous vs induced)

	Case	Control	P
Affective problems*	51.5[11]	50[2]	0.012
Anxiety problems*	51[9]	50[1]	0.008
Somatic problems	50[1]	50[6]	0.396
Attention Deficit/ Hyperactivity Problems	50[6]	50[0]	0.019
Oppositional Defiant Problems	51[6]	50[1]	0.035
Conduct Problems *	50[7]	50[1]	0.001

Figure 7 : DSM-Oriented scale scores in the patient group as compared to the control group. Scores are given as median [IQR] with higher scores denoting poorer outcome. P value was adjusted using the Bonferroni-Holm method to account for multiple comparison. *Denotes significant variable

CONCLUSIONS

- This study demonstrated that the patient group had overall impaired quality of life and increased psychological distress as compared with the control group
- We also demonstrated that it was particularly female patients with DSD who had significantly lower quality of life scores as compared with the control group.
- The complexity of managing patients with DSD in a culture which is strongly male dominated needs to be addressed.
- Ultimately the findings of this study lend support to the proposal that a skilled multidisciplinary team is necessary to manage these patients and address their concerns.

References

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