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

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*No conﬂict of interest

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

<table>
<thead>
<tr>
<th>Karyotype</th>
<th>Case</th>
<th>Control</th>
</tr>
</thead>
<tbody>
<tr>
<td>Male Social Sex (n)</td>
<td>25</td>
<td>2</td>
</tr>
<tr>
<td>Female Social Sex (n)</td>
<td>5</td>
<td>1</td>
</tr>
</tbody>
</table>

Figure 1: Study population according to karyotype and social sex

- No signiﬁcant difference between baseline characteristics of cases and control

2. PedQOL

- 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.

- 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

- 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)

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