## ORIGINAL RESEARCH

# Quality of Life and Psychological Adjustment of Women Living with 46,XY Differences of Sex Development

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## ABSTRACT

***Background.*** Progressive care improvement for differences of sex development (DSD), regarding diagnosis communication, psychological, medical and surgical management has been claimed.
***Aim of the study.*** To assess clinical management, quality of life (QoL) and the general psychosocial adjustment of individuals with 46,XY DSD. Some differences related to age at diagnosis are investigated.
***Design.*** Cross-sectional study using standardized questionnaires.
***Population.*** Forty-three Caucasian females with 46,XY DSD (self declared diagnoses: complete androgen insensitivity syndrome, n = 34; complete gonadal dysgenesis, n = 1; 5α-reductase deficiency, n = 4; Leydig cell hypoplasia, n = 1; unknown diagnosis, n = 3; age years: 31.5 ± 9.6 [range 18–57 years]).
***Setting.*** University Hospitals.
***Methods.*** Subjects were required to fill in questionnaires (ABCL, WHOQOL, dedicated 17-item questionnaire). Academic and socioeconomic data were compared with those of the Italian population. QoL and psychological data were compared with those of a comparison group (46,XX healthy females: n = 43; age, years: 34.5 ± 9.7, range 22–51years).
***Results.*** Present sample of women living with 46,XY DSD were well adapted and were higher achievers than controls, both in educational and professional life. They showed good QoL, but they appeared less satisfied in psychological and social areas. They had borderline mean scores and statistically higher scores than the comparison group for depression, anxiety, internalizing and externalizing problems. Younger persons living with a 46,XY DSD showed better psychosocial adjustment than older ones. Younger women showed lower age at diagnosis communication. Psychological support was more often proposed at the time of diagnosis communication to younger individuals, and they undertook it more frequently than older ones.
***Conclusions.*** Italian people living with 46,XY DSD were well adapted and successful; they reported a good QoL but showed higher degree of psychological distress than the comparison group. Lower psychological distress in younger women could indicate some positive effects of changes in management. **D'Alberton F, Assante MT, Foresti M, Balsamo A, Bertelloni S, Dati E, Nardi L, Bacchi ML, and Mazzanti L. Quality of life and psychological adjustment of women living with 46,XY differences of sex development. J Sex Med 2015;12:1440–1449.**

***Key Words.*** DSD; Differences of Sex Development; Quality of Life; Psychology, Psychological Adjustment; Hormone Replacement Therapy; Diagnosis Communication

## Introduction

### Background

The term "disorders of sex development" (DSD) defines a group of congenital conditions in which the development of chromosomal, gonadal, or anatomical sex is atypical [1]. Although considered provisional, this terminology was an attempt to more correctly define a heterogeneous group of rare conditions that had stigmatizing and discriminating names in the past. However, this terminology progressively showed its limits in defining a "variant" of intrauterine human development as a "disorder" [2–5]. Thus, we will use the acronym DSD, standing for "*differences* of sex development." This term underlines the variability of the human expression of congenital conditions with genetic determinants that are not yet fully known, as well as the best psychological, medical, and surgical approaches to optimize long-term outcomes [1,2,6–12].

Indeed, relevant changes in the management of DSD took place when people with DSD broke the veil of secrecy, blamed the prejudices and paternalism that surrounded their condition [13], and pleaded for a change in its management (http://isna.org, http://accordalliance.org, http://aissg.org, etc.). New information flooded medical congresses and medical practice providing new and innovative points of view [13–15]. Support groups arose all over the world, and the Chicago Consensus started new collaborative work between DSD activists and the medical, surgical, and psychological establishment [1,16]. In Italy, AISIA (Italian Androgen Insensitivity Syndrome support group) has made a great effort to help people, to share information, and to contact medical staff, in order to promote more appropriate DSD management. Among the projects developed by AISIA, there are surveys on quality of life (QoL), emotional, and behavioral well-being, and on opinions about the medical and surgical histories of persons with 46,XY DSD (http://www.sindromedimorris.org).

These data are very difficult to collect due to the privacy that must be guaranteed and to the difficulty created by the atmosphere of secrecy and the lack of information that women with DSD suffered from in the past [17,18].

In a peer review, Wisniewski and Mazur [10] found very different data on psychological well-being and quality of life. Mazur et al. [19] assessed the QoL of five persons with 46,XY DSD and reported a general positive QoL in four of them. Johannsen et al. [20] assessed QoL and mental distress of a mixed sample of 46,XY DSD and 46,XX DSD and found that women with complete androgen insensitivity syndrome (CAIS) had a higher QoL and better psychological outcomes than a control group. Hines et al. [21] studied QoL and general psychological well-being in a group of 22 women with CAIS, showing no statistically significant differences with the matched controls in terms of sexual QoL. Warne [9] investigated a group of 50 46,XY and 46,XX DSD and did not find any differences between the DSD group and the controls, in terms physical and mental health, depression, anxiety, and other aspects of psychological well-being. Problems in sexual confidence and sexual satisfaction were found also by Fliegner et al. [22]. Krupp et al. [23] found that QoL and psychological distress in CAIS were within the average range even though a considerable percentage of individuals were over the cut-off score for clinical relevance. Slijper et al. [24] found a general an overall tendency in people with DSD to develop general psychopathologies. Schützmann et al. [25] (in their sample of 37 persons with different forms of DSD) found high rates of psychological distress. The German network of Disorders of Sex Development [12] studied 439 young, adolescent and adult males and females with different forms of DSD and planned a large clinical evaluation on QoL, gender identity, treatment satisfaction, and other factors related to DSD. In this large-scale project, 47 females with 46,XY DSD were investigated. The preliminary results indicated relevant problems in terms of desire, arousal, and dyspareunia in females with DSD [26], thereby confirming other data regarding sexual QoL [6].

Some studies [5,24,25] found that suicidal thought and suicide attempt rates were higher in people with DSD than in controls. Diamonds and Watson [27] found that secrecy, shame, and stigma caused the majority of difficulties in people with CAIS and that open diagnosis communication was still far from widespread.

## Aims

The results on psychological adjustment in adulthood remain controversial and new research to obtain updated information is considered advisable in order to better understand the specific needs for care of women living with 46,XY DSD. We planned the study described below in order to assess clinical management, QoL, and the general psychosocial adjustment of individuals with 46,XY