

# Health-related Quality of Life & Psychological Wellbeing in Adults with Disorders of Sex Development (DSD)

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## Background:

Rare congenital conditions with incongruence of chromosomal, gonadal, and phenotypic sex have been summarized as Differences/ Disorders of Sex Development (DSD). Included in DSD are conditions with diverse genetic etiology, varying levels of prenatal androgen effects, phenotypes, and subsequently, different medical treatments. Quality of life (QoL) and psychological wellbeing are indicators of successful psychosocial adaptation to the conditions. Studies addressing these issues in individuals with DSD vary greatly.

## Methods:

This multicenter clinical evaluation study was part of a German network related to DSD funded by the German Ministry of Science and Education (BMBF 2003 to 2007). To assess health-related quality of life (HRQoL), as well as psychological wellbeing, the Short Form Health Survey (SF-36) and the Brief Symptom Inventory (BSI) were used.

## DSD conditions:

Participants (N=110) were classified into 6 groups:

This groups reflect gender, diagnosis and androgen effects: females (f), males (m) or a different gender from male or female, congenital adrenal hyperplasia (CAH), XY DSD with partial (pa) or no (na) androgen effects. Participants with gender changes and conditions not fitting the five groups are summarized in the group "Others".

### f-CAH-pa (n=44)

12 simple virilizing  
32 salt-wasting

### f-XY-pa (n=26)

13 partial/ mixed gonadal dysgenesis  
6 pAIS  
3 17beta-hydroxysteroid- dehydrogenase III deficiency  
2 5alpha-reductase II deficiency  
1 LH-receptor defect  
1 unclassified

### f-XY-na (n=13)

5 complete gonadal dysgenesis  
8 CAIS

### m-XY-pa (n=10)

3 partial/mixed gonadal dysgenesi  
1 pAIS  
3 severe hypospadias  
1 hermaphroditism verus  
1 epispadias  
1 penile hypospadias and micropenis

### other gender (n=9)

2 partial/mixed gonadal dysgenesis  
1 complete gonadal dysgenesis  
3 CAIS  
1 pAIS  
1 5alpha-reductase II deficiency  
1 17beta-hydroxysteroid- dehydrogenase III deficiency

### Others (n=8)

Females:  
1 pAIS\*  
1 penile agenesis\*  
1 Klinefelter syndrome\*  
1 aromatase deficiency (46,XX)  
1 ovarian insufficiency (46,XX)  
1 complete gonadal dysgenesis (46,XX)  
Males:  
2 CAH (46,XX)\* in one

\*with gender change in history

## Psychological Wellbeing:

Comparison of the total cohort and population based reference data

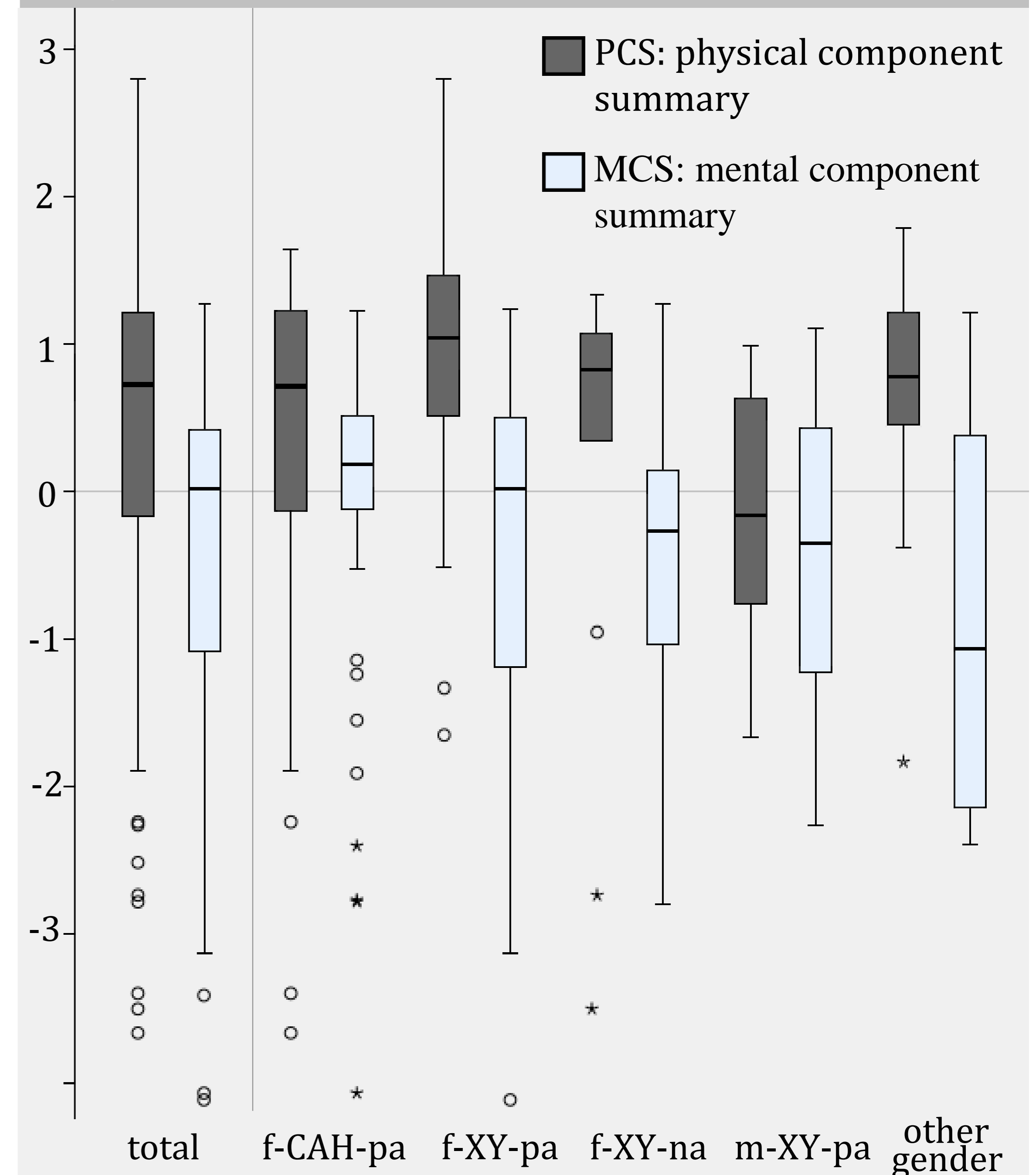
BSI	T	sd
somatization	50.68	10.76
obsessive-compulsive	<b>52.98</b>	<b>12.35 *</b>
interpersonal sensitivity	<b>53.99</b>	<b>11.72 **</b>
depression	<b>54.79</b>	<b>11.84 **</b>
anxiety	<b>52.81</b>	<b>10.84 *</b>
hostility	<b>56.10</b>	<b>11.52 **</b>
phobic anxiety	50.56	9.05
paranoid ideation	<b>54.75</b>	<b>11.37 **</b>
psychoticism	<b>54.06</b>	<b>11.63 *</b>
global severity index	<b>54.39</b>	<b>13.54 **</b>
positive symptom total	<b>54.79</b>	<b>12.04 **</b>
positive symptom distress	<b>53.45</b>	<b>12.46 **</b>
Clinical cases	47 (42.7%)	

### Group comparison; clinical cases

	Clinical cases n (%)
f-CAH-pa	14 (31.8%)
f-XY-pa	12 (46.2%)
f-XY-na	7 (53.8%)
m-XY-pa	4 (40.0%)
other gender	5 (55.6%)

## Health related Quality of Life:

Group comparison of the physical and mental HRQoL



**SF-36:** Higher scores indicate a higher HRQoL; z=0 is the mean of the norm (sd=1)

**BSI:** higher scores indicate a higher level of psychological distress; T=50 mean of the norm (sd=10); p values are calculated with the nonparametric Wilcoxon signed rank test T equals 50.00; p ≤ .05\* and p ≤ .01\*\*; Criteria for being classified as a clinical case were either a Global Severity Index of T ≥ 63 or two single subscale scores of T ≥ 63

## Conclusion :

- 9 participants (8.2%) describe themselves as a gender other than male or female. They rated their HRQoL and psychological distress as similar to the total cohort.
- Participants reported significant impaired psychological wellbeing, 42.7% can be described as a clinical case.
- Participants reported overall higher physical HRQoL and normal mental HRQoL compared to the norm
- In group comparison females with XY DSD and androgen effect reported the highest physical HRQoL; females with CAH reported less psychological distress
- Specialized interdisciplinary care with a focus in particular on psychological care is needed to improve wellbeing.