Etiologic Classification of 46, XY Disorders of Sexual Differentiation According to Chicago Consensus: Single Center Results

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Objective

The aim of the study was to describe the etiologic diagnosis, clinical characteristics in children with 46,XY disorder of sexual development (DSD).

Methods:

The 125 46, XY patients were included the retrospective study. The definitive diagnosis was made by presentations and clinical findings, gonadal morphology and genital anatomy of patients, basal and stimulated hormone results, imaging methods and molecular genetic analyzes. All data obtain from hospital records.

Results:

Types and ratios of each presentation of the 125 patients with 46,XY DSD were as follows (Table) Disorders of testicular development (8%), disorders of androgen synthesis or action (16%), other causes (57.6 %) and associate with syndromes (%). Among the other causes etiology were as hypospadias (43.2%), undescended testis (16.2%) and micropenis (36.1%). Hypospadias was detected in a patient with *CYP21A2* mutation. Sixteen patients were raised as girls. 55 patient' parents were consanguineous. Brothers of thirteen patients and cousins of five patients' also have similar disorders.

	No	Raised gender as
46, XY DSD (n:125)		
A-Disorders of testicular development		
1-Complete gonadal dysgenesis		
9p del	1	F
WT1	1	F
2-Testicular regression syndrome	5	M
3- Ovotesticular DSD	3	1F, 2M
B-Disorders of androgen synthesis or action		
1-Androgen synthesis defects		
a-Smith-Lemli-Opitz syndrome	1	M
b-20,22 Desmolase deficiency	1	F
c-3 BHSD deficiency	2	M
d-17 Hydroxylase deficiency	1	F
e-5a Reductase 2 mutation	4	2F, 2M
f-21 Hydroxylase deficiency and hypospadias	1	M
4-Disorders of androgen action		
a-CAIS	9	9F
b-AMH-R defect	1	M
III-Others		
a-Hypospadias	29	M
b-Hypospadias and Micropenis	2	M
c- Hypospadias and undescended testis	1	M
b-Epispadias	2	M
c-Undescended testis	12	M
d-Micropenis	23	M
e-Micropenis and undescended testis	3	M

Conclusion:

The most common etiological diagnosis in 46 XY DSD was hypospadias. Defects in androgen synthesis and action as etiological causes of DSD were at the same frequency with associate with syndromes in this study. Ovotesticular DSD was rare.