

# HORMONAL ASSESSMENT OF MALFORMATION SYNDROMES ASSOCIATED WITH DISORDERS OF SEX DEVELOPMENT: CASE SERIES

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## INTRODUCTION

- Disorder of sex development (DSD) is a challenging condition confronting the patients, their relatives and the clinicians. It is more challenging when this disorder is associated with other extra-genital malformations. This makes their overall management more complex than if they just had DSD needing a multidisciplinary approach involving pediatricians, specialists in the field of endocrinology, genetics, surgery and psychiatry in order to reach a prompt and accurate management of such patients.
- To be considered, malformations associated with DSD may be either with defective hormonal function or with normal hormonal function but aberrant morphogenesis of the internal or external genitalia.

## OBJECTIVES

- This work aimed at clinical review of 9 cases with malformation syndromes associated with DSD and evaluation of their testicular hormonal status. Moreover, this study aimed at highlighting the importance of genital and extra-genital examination and its relation to the hormonal assay.

## SUBJECTS & METHODS

- Nine patients with syndromic DSD attending the Endocrinology clinic in Alexandria University Children's Hospital were assessed on emphasis on detailed history taking, thorough clinical examination (genital and extra-genital examination), reproductive hormonal assessment, karyotype analysis and the appropriate imaging study as needed. Hormonal assessment included basal FSH, LH, AMH, inhibin B, and testosterone measurements and hCG stimulation test by giving hCG (1500 IU/day) intra-muscular injections for three consecutive days.

## RESULTS

- 9 patients having malformations associated with DSD were assessed. All patients were diagnosed at the neonatal period except one patient diagnosed at one year reflecting awareness among parents about DSD.
- All patients were initially assigned as males.

Table (1): Distribution of the studied cases according to associated malformations

Associated malformations	No.	%
Robinow syndrome	2	22.2
Multiple congenital anomalies (cardiac, skeletal, anogenital, dysmorphic features, ophthalmological)	2	22.2
Congenital hypothyroidism	2	22.2
Steroid resistant nephrotic syndrome	1	11.1
Arrested hydrocephalus and learning disabilities	1	11.1
Skeletal dysplasia	1	11.1

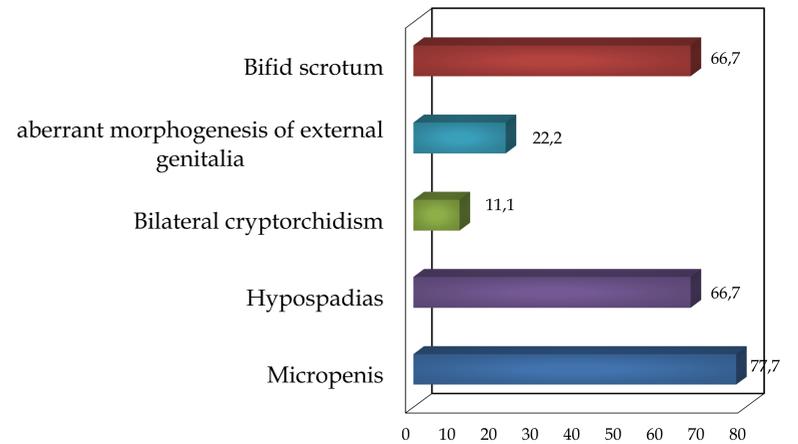


Figure (1): Physical examination of the external genitalia among the studied cases

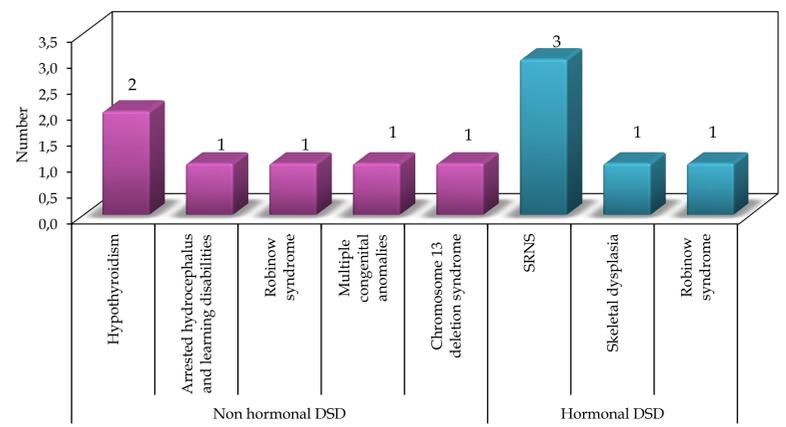


Figure (2): The studied cases according to their hormonal profile

## CONCLUSION & RECOMMENDATIONS

- Malformation syndromes with XY DSD are more challenging conditions either in work up to reach the definite diagnosis or in counselling for families about their affected children or other siblings.
- Collaborative hormonal assay should be done to reach the final diagnosis in all patients having DSD even those with other malformations.
- Genetic testing is very essential to illustrate the picture in the management of XY DSD especially in those with other malformations.

## REFERENCES

- Hutson JM, Grover SR, O'Connell M, Pennell SD. Malformation syndromes associated with disorders of sex development. *Nat Rev Endocrinol* 2014; 10(8):476-87.
- Shojaei A, Ebrahimzadeh-Vesal R, Ahani A, Razzaghy-Azar M, Khakpour G, Ghazi F, Tavakkoly-Bazzaz J. Genetic Screening of Iranian Patients with 46,XY Disorders of Sex Development. *Rep Biochem Mol Biol*. 2017 Oct;6(1):59-65.
- Soman C, Lingappa A. Robinow Syndrome: A rare case report and review of literature. *Int J Clin Pediatr Dent* 2015;8(2):149-52

