SPECTRUM OF GENITAL ABNORMALITIES IN ROBINOW SYNDROME: CASE SERIES

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INTRODUCTION

- Robinow syndrome is a rare genetic disorder inherited either in autosomal recessive or dominant patterns. It is characterized by mesomelic dwarfism, characteristic facial features, skeletal abnormalities, multiple sets of teeth and external genital abnormalities.
- In males, the characteristic pattern is micropenis with or without cryptorchidism, webbed penis or hypoplastic scrotum. In females, the anatomical defect is not always evident. There is hypoplastic clitoris and labia minora.
- Human mutation in WNT5A or ROR2 that are involved in Robinow syndrome is a candidate mechanism for cryptorchidism in many patients with that syndrome.

OBJECTIVES

- This work aimed at defining the spectrum of genital abnormalities as a part of the full picture of the syndrome in 11 patients with Robinow syndrome following up in Alexandria University Children's Hospital.

SUBJECTS & METHODS

- Eleven patients having Robinow syndrome were subjected to full history taking, detailed clinical examination and anthropometric measurements including height, weight, and head circumference. Furthermore, parents and available siblings were examined. X-ray studies, echocardiography, and chromosomal analysis, done by G-banding technique using peripheral blood sample, were performed for these patients.

RESULTS

- The study included 11 patients with Robinow syndrome. They included 7 boys and 4 girls. Their age ranged from 3 months to 66 months. History of consanguinity was found in 63.6% of these patients. Two patients had history of similar condition in their families.

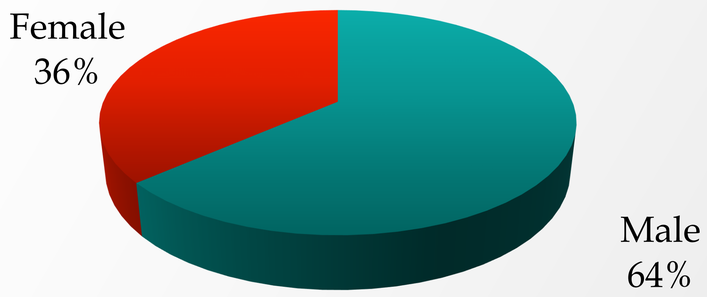


Figure (1): Gender distribution of the studied patients.

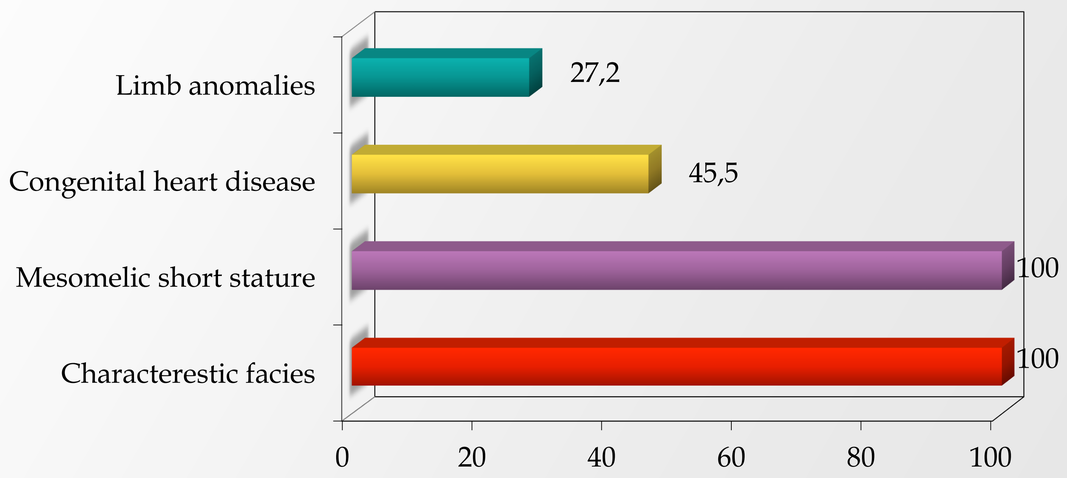
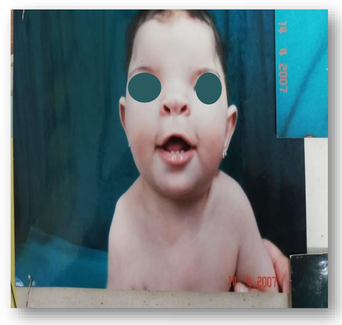
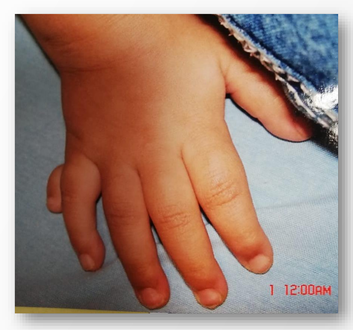
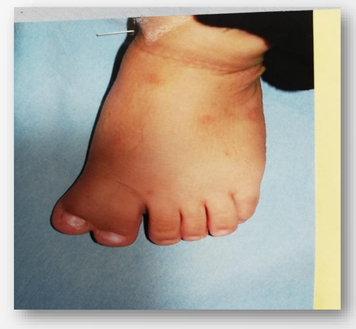


Figure (2): Physical characteristics in Robinow syndrome of the studied patients







- As regard genital abnormalities, all girls had no genital abnormalities. However, 90.9% of boys had genital abnormalities. These anomalies were found either isolated or in combination in the form of disorder of sex development.

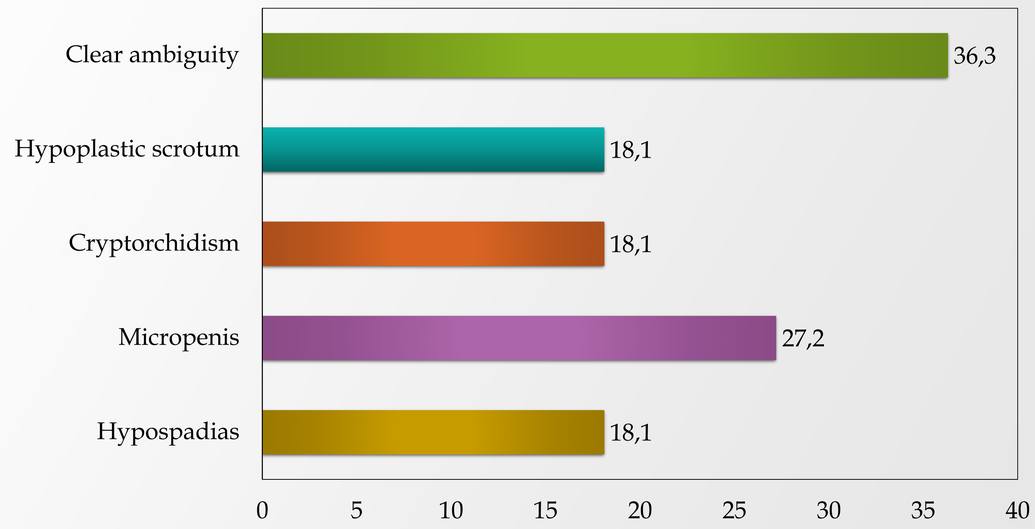


Figure (3): Spectrum of genital abnormalities in boys with Robinow syndrome

Conclusions & Recommendations

- Robinow syndrome is diagnosed based on clinical and radiological findings.
- Genital abnormalities were very evident among male population in our cohort. These abnormalities include hypospadias, micropenis, hypoplastic scrotum, cryptorchidism or ambiguous genitalia.
- Robinow syndrome is one of the rare aetiologies of disorders of sex development.

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Sex differentiation, gonads and gynaecology or sex endocrinology

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