**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 or 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.

<table>
<thead>
<tr>
<th>Female</th>
<th>36%</th>
</tr>
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<tbody>
<tr>
<td>Male</td>
<td>64%</td>
</tr>
</tbody>
</table>

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

<table>
<thead>
<tr>
<th>Limb anomalies</th>
<th>27.2</th>
</tr>
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<tbody>
<tr>
<td>Congenital heart disease</td>
<td>45.5</td>
</tr>
<tr>
<td>Mesomelic short stature</td>
<td>100</td>
</tr>
<tr>
<td>Characteristic facies</td>
<td>100</td>
</tr>
</tbody>
</table>

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

**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.

**REFERENCES**