Ovotesticular Disorder of Sexual Development: 31 cases followed-up in a single-center in Brazil

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Introduction

Ovotesticular Disorder of Sexual Development (OT DSD) is a rare condition characterized by histologic demonstration of both ovarian and testicular tissue in the same individual. Descriptions in literature usually have small samples and do not include patient evolution data. The aim of this study is to describe clinical, biochemical and histological findings, as well as long-term outcomes (including onset and progression of puberty) in patients with OT DSD accompanied in a tertiary center in São Paulo-Brazil.

Methods

This is a single-center retrospective study in which thirty-one patients diagnosed with OT DSD were included. The patients were accompanied between 1978 and 2018, at the Children's Institute of Hospital das Clínicas, University of São Paulo (Brazil). A systematic review of medical records was carried out to obtain clinical, biochemical and histological data and to evaluate puberty progression.

Results

The mean age of the first visit was 32.1 months (varying from 6 days of life to 17 years). The initial sex was male in 17 cases (54,8%), female in 8 cases (25,8%) and undetermined in 6 cases (19,3%). The final sex was male in 14 patients (45,1%) and female in 17 patients (54,8%).

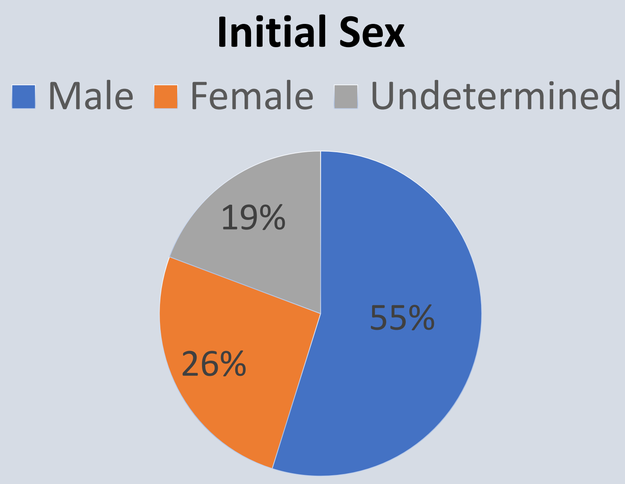
Clinical examination at the arrival evidenced mean phallus size of 2.5 cm and palpable gonads (unilateral or bilateral) in 21 patients (67.7%). The location of the urethra was predominantly perineal (74.2%), followed by topic in 6 patients (19,4%) and at the bottom of the penis in 2 patients (6,4%). Mullerian structures were observed in 23 patients (74,2%).

Twenty patients were submitted to the hCG test and 12 of them (60%) presented positive response (>150ng/dL). The mean testosterone value among all patients, considering the highest value obtained (between basal testosterone and after the hCG stimulation) was 245.4 ng/dL.

The most frequent karyotype was 46,XX, in 18 patients (58.1%), followed by mosaics in 8 patients (25.8%), 46,XY in 4 patients (12,9%) and 46,Xr in 1 patient (3,2%). The identified mosaics were 46,XX/45,X/47,XXY; 46,XX/46,XY; 46,XX/47,XXY; 46,XY/45,X; 45,X/46,Xr; 46,XX/46,XY; 47,XXY/46,XY. FISH test was performed in 5 patients, all presenting SRY negative result.

The ovotestis was the more frequent gonad (48.4%), followed by ovary (30,6%) and testicle (21%). The most common combination was ovotestis + ovary (38.7%), followed by ovary + testicle (22,6%), bilateral ovotestis (19,3%) and ovotestis + testicle (19,3%). The mean number of surgeries was 2.7 per patient.

Puberty could be evaluated in 19 of 31 patients (61.3%). Eleven presented spontaneous puberty (7 women and 4 men) and the average age of pubertal onset was 11,9 years. Eight patients needed puberty induction, 4 females and 4 males. Among them, six were agonadic.



Discussion and Conclusion

This is a significant sample of OT DSD patients, with phenotypic, biochemical, histological and genotypic data compatible with literature. In our sample, the majority of patients had female final sex (54.8%), which is not seen in most studies. The surgical option for female sex is often easier, with fewer surgical approaches and a greater chance of preserving fertility. Puberty should be monitored and appear to be spontaneous in most cases when there is preservation of gonadal tissues. OT DSD remains a challenge for clinicians and more studies are needed to evaluate these patients during puberty and in the long term.

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57ESPE 346--P3

Sex differentiation, gonads and gynaecology or sex endocrinology

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