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Undervirilization in a premature male infant: an unusual case

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Abstract: The birth of a newborn with abnormal genitalia (atypical sex) constitutes a major social emergency, and presents a difficult diagnostic challenge for the Neonatologist providing care , we present a premature male infant with undervirilized genitalia. The issue of gender assignment was discussed.
Keywords: diagnosis, male, premature, undervirilization.

INTRODUCTION

The birth of a newborn with abnormal genitalia (atypical sex) constitutes a major social emergency and presents a difficult diagnostic challenge for the pediatrician providing care, who should involves, an experienced multi-disciplinary team in the management [1-4].

The causes of ambiguous genitalia include genetic variations, hormonal imbalances and malformation of the fetal tissues that are supposed to evolve into genitalia [5-7]. We report here on a premature male infant who presented with undervirilized genitalia.

CASE REPORT

A preterm infant was born at 24 weeks of gestation to a 31-year-old Saudi G_4 P_4 A_0 mother. The infant's mother was healthy and received routine antenatal care. She was not on any medication nor exposed to radiation. He received surfactant and ventilated for 3 weeks. At one month of age the infant was seen by pediatric endocrinologist for assessment of possible ambiguous external genitalia. The infant was noted to have fused labioscrotal folds (shallow scrotum) with no palpable gonads, but there was fullness in mid-right inguinal region, and a phallus-like structure measuring 1.5 cm with an opening at the tip of the phallus. The baby did not exhibit any dysmorphic features. Chromosomal study revealed 46XY karyotype and an ultrasound of the pelvis showed no internal female organs. Serum electrolytes and glucose were normal as well as serum LH, and FSH. Serum testosterone (ST) of 2.8 nmol/L (normal 0.1-0.5). Normal thyroid function with free thyroxine (FT_4) of 15.7 Pmol/L (normal; 10-25) and thyroid stimulating hormone (TSH) of 1.7 IU/L (normal; <5 IU/L). Random growth hormone 14.3 ng/ml (normal; >10). Serum cortisol 880 nmol/L (normal; >600) and adenocorticotropic hormone (ACTH) of 8 Pmol/L (normal; 5-15). 5-α-reductase was ruled out by doing testosterone, dihydro testosterone ratio after 3 days injections of human chorionic gonadotro pin (HCG) stimulation test.

The baby was given 25 mg testosterone intramuscular. One month later the size of the phallus increased to 1.9 cm in length. At 4 months of age the right testicle was felt in the scrotum and the left one was palpable in the mid-inguinal region. The phallus size was 2-3 cm and the scrotum was still shallow.

DISCUSSION

Disorders of sex development (DSD) are defined as atypical sex. The differential diagnosis is so extensive. The appropriate sex of rearing of a very undervirilized male requires as much information as possible, as well as through multidisciplinary discussions involving the parents, urologist or pediatric surgeon, endocrinologist, geneticist and a clinical psychologist [4-7].

Male sexual differentiation is initiated by SRY gene on the short area of the Y chromosome. Under its influence the undifferential gonads forms a testis, which produces the hormonal milieu that results in male sexual differentiation. Testosterone stimulates the Wollfian structure (epididymis vas deferens, and seminal vesicles) and anti-Mullerian hormone suppresses the development of the Mullerian structure

(fallopian tube, uterus, and upper vagina). The conversion of testosterone to dihydro testosterone occurs in the skin of the external genitalia and masculinized the external structures. Most of this male differentiation takes place by about 12 weeks, after which the penis grows and the testes descent into the scrotum. In the absence of SRY, female sexual differentiation occurs. An error in genital morphogenic may occur at any step in this developmental pathway [1, 3, 4]. The hormonal results and clinical progress ruled out hypothalamic-pituitary-gonadal axis, 5-α-reductase and androgen insensitivity as a cause.

Ens *et al.* [8] had reported on a premature infant who presented with undervirilized genitalia, however, they attributed this to intra-uterine growth retardation (IUGR). Placental factors and the possibility of a role for endocrine disruptor chemical have been postulated [9-10].

Advanced in perinatal and neonatal care over the last 30-years have contributed in the survival of many premature babies [11, 12]. In approximately one-half of undervirilized males there was no specific diagnosis. It is tempting to say that immaturity in the growth like what happening with the descent of the testicles. Almost 5% of the full-term babies had undescended testicles, while the ratio can reach 30% in preterm babies [13]. Further follow-up of this baby with genetic studies are needed.

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REFERENCES

1. Hiort D; Neonatal endocrinology of abnormal male sexual differentiation, molecular aspects. Horm Res 2000; 53 (Supplement 1): 38-41.
2. Moshiri M, Chapman T, Fechner PY, Dubinsky T, Shnorharorian M, *et al.*; Evaluation and management of disorders of sex development : multi-disciplinary approach to a complex diagnosis. Radiographic 2012; 32: 1500-618.
3. Al Jurayyan NAM; Disorders of sex development: diagnostic approaches and management options. An Islamic perspective. Malaysian J Med Sci 2011; 18: 4-12.
4. Houk CP, Hughes IA, Ahmed SF, Lee PA; Writing committee for the international intersex consensus conference participants. Summary of consensus statement on intersex disorders and their management. Pediatric, 2006; 18: 753-57.
5. Morel Y, Rodolfo R, Teinturier C; Aetiological diagnosis of male sex ambiguity: a collaborative study. Eur J Paediatr 2002; 161; 49-59.
6. Ogilvy-Stuart AL, Brain CE; Early assessment of ambiguous genitalia. Arch Dis Child 2004; 39: 401-407.
7. Al Jurayyan NA, Al Issa SDA, Al Nemri AMH, Al Otaibi HMN, Babiker AM; The spectrum of 46Y disorders of sex development in a university centre in Saudi Arabia. J Pediatr Endocrinol Metab 2015.
8. Ens A, Cumming E, Sanderson S; A premature infant with ambiguous genitalia. Paediatr Child Health 2011; 16(10): 643-645.
9. De Andrade Machado Neto F, Moreno Morcillo A, Trevas Maciel-Gurrea A, Guerra-Junior G; Idiopathic male pseudohermaphroditism is associated with general growth retardation. Eur J Pediatr 2005; 164: 287-91.
10. Hussain N, Chaghtai A, Herndon CDA, Herson VC, Rosenkrantz TS, McKenna PH; Hypospadias and early gestation growth restriction ion infants. Pediatrics 2002; 109: 473-79.
11. Costeloe KL, Hennessy EM, Haider S, Stacey F, Marlow N, Draper ES; Short term outcome after extreme preterm birth in England: comparison of two birth cohort in 1995 and 2006 (the EPI cure studies). BMJ 2012; 345: 79-76.
12. Lacovidou N, Varsami M, Suggellou; Neonatal outcome of preterm delivery. Ann NY Acad Sci 2010; 105:130-134.
13. Docimo SG, Silver RI, Cromie W; The undescended testicle: Diagnosis and management. Am FAM Physician 2000; 62(9): 2037-2044.