Original Study

Virilized External Genitalia in Young Girls: Clinical Characteristics and Management Challenges in a Low-Resource Setting

S.O. Ekenze MBBS, FWACS [1,*], C.O. Adiri MBBS, FWACS [2], I.O. Igwilo MBBS [1], O.O. Onumaegbu MBBS, FWACS [3]

[1] Sub-Department of Paediatric Surgery, University of Nigeria Teaching Hospital, Enugu, Nigeria
[2] Department of Obstetrics and Gynaecology, University of Nigeria Teaching Hospital, Enugu, Nigeria
[3] Plastic Surgery Unit, Department of Surgery, University of Nigeria Teaching Hospital, Enugu, Nigeria

A B S T R A C T

Objective: Virilization of the external genitalia in young girls (VEG) manifests mostly as ambiguity of the genitalia and elicits concerns and uncertainties especially in settings with poor awareness. This study evaluates the profile and challenges of VEG in southeast Nigeria.
Methods: We analyzed 23 children with VEG managed in 2 referral centers in southeast Nigeria from June 2005 to January 2013.
Results: They presented at median age of 13.3 months (interquartile range [IQR] 3 months-3 years). The cases included 3 (13%) of Prader type 1, 6 (26%) of type 2, 11 (48%) of type 3, and 3 (13%) of type 4. Five of the Prader type 3 and all 3 cases of Prader type 4 were reared as male prior to presentation. Following evaluation, all the cases were assigned female gender at a mean age of 2.7 years (range 2 months-10.5 years). Appropriate feminizing genitoplasty was undertaken in all the cases and after a follow-up period of 3 months to 5 years (mean 2 years), 2 patients developed vaginal stenosis, and 3 cases had surgical wound infection. Poor awareness, delayed presentation, inadequate facilities, and lack of trained manpower were the challenges in the management of the cases.
Conclusion: VEG in our setting is associated with delayed management. Focused health education and public awareness programs, and improved healthcare funding may improve outcome and minimize the need for gender reassignment.
Key Words: Virilized genitalia, Girls, Intersex, Challenges, Developing country

Introduction

Virilization of the external genitalia in female children (VEG) results mostly from in-utero exposure to supra-physiologic level of androgens.[1] The recognized causes of this condition include congenital adrenal hyperplasia (CAH), luteoma, polycystic ovary disease, Krukenberg tumors, and exposure to progestins.[2–4] CAH is the commonest cause of virilization of the female child. It defines a cluster of inherited deficiency of 1 of the enzymes necessary for the synthesis of cortisol from cholesterol.[3] The virilization causes ambiguity of the genitalia which phenotypically manifest as varying degree of clitoromegaly, labioscrotal fusion, rugation (folded, wrinkled, or creased appearance of), and pigmentation of the labia.[3,5] The severity of virilization varies with the degree of exposure to the androgen and it is classified clinically according to Prader's staging[6] into 1 to 5 (1, isolated clitoromegaly, 5, complete masculinization but no palpable testes). Management of VEG aims to facilitate the best possible quality of life for the patient and critical to this goal is consideration for cosmetic appearance, sexual function, fertility, and psychological well-being.[3,5,7] Surgical procedures to repair the genitalia in VEG may range from simple procedure in mild cases to daunting vulvovaginal reconstruction in more severe cases.[8] Over the past decades, outcome of management of VEG has improved, probably as a result of early diagnosis, multidisciplinary team management, improvements in diagnostic testing, and timely genital repair.[5,7] Controversies however still exist in the gender assignment and timing for vaginal repair in cases with very severe virilization.[9]

In many developing countries, delayed presentation and lack of diagnostic facilities, among other factors, have continued to challenge the management of VEG with resultant indecisions in gender assignment, delay in intervention, and poorer outcome.[10,11] In this setting, it may be imperative to understand the profile of VEG and factors that underlie these challenges in order to develop initiatives to address the challenges.

This study evaluates the clinical characteristics and the challenges of managing VEG at the University of Nigeria Teaching hospital, and Mother of Christ Specialist Hospital, Enugu, in south-east Nigeria.

Materials and Methods

Cases of ambiguous genitalia with suspected virilization of the external genitalia are referred to University of Nigeria Teaching hospital Enugu (UNTH) from secondary healthcare facilities in southeast region of the country for management. In addition to this, some cases are referred to Mother of Christ Specialist Hospital, Enugu (MOC) where the lead author provides pediatric surgical services. On presentation,

The authors indicate no conflicts of interest.

* *Address correspondence to:* Dr. S.O. Ekenze, Sub-Department of Paediatric Surgery, University of Nigeria Teaching Hospital, Enugu 400001, Nigeria; Phone: 234-803-777-3831
E-mail address: sebekenze@gmail.com (S.O. Ekenze).

1083-3188/$ - see front matter
http://dx.doi.org/10.1016/j.jpag.2013.07.004

the patients are evaluated with the available facilities for gender confirmation: buccal smear for Barr body estimation (positive if >5% of the cells have Barr body); pelvic ultrasonography to identify the uterus, fallopian tubes, ovaries; and pelvic minilaparotomy to visualize the nature of the internal ducts if the preceding investigations are inconclusive. In the past 2 years serum chemistries and routine electrolyte tests have been undertaken. Facilities for chromosomal analysis and laparoscopy are not available in the 2 centers. Following evaluation, appropriate gender is assigned after discussion with parents, and consent obtained for genital surgery. The cases were managed in conjunction with the clinical psychologists and gynecologists.

From June 2005 to January 2013, 23 cases were managed in the 2 centers (15 from UNTH, 8 from MOC). These cases were retrospectively analyzed. Data on given names, gender at presentation, age at presentation, possible reason for delayed presentation (presentation after 1 month of noticing the anomaly), and clinical presentation were collected from the case notes, discharge summaries, and theatre records. Other data collected include results of investigation, age at definitive gender assignment, treatment, outcome, and follow-up.

Statistical Package for Social Sciences (SPSS 15.0 version, SPSS Inc, Chicago, IL) was used for data entry and analysis. Results were expressed as percentages, median or mean.

Results

The median age of the cases at presentation was 13.3 months (IQR 3 months-3 years). Of the patients, 13 (57%) had feminine names, 8 (35%) had masculine, and 2 (88%) had unisex names at presentation. Overall, 15 (65%) were reared as female, and 8 (35%) as male prior to presentation (Table 1).

Presentation

Three (13%) patients had isolated clitoral hypertrophy (Prader type 1), 6 (26%) had clitoral hypertrophy with urethral and vaginal orifices present but near each other (Prader type 2), 11 (48%) had clitoral hypertrophy with single urogenital orifice and posterior fusion of labia majora (Prader type 3; Fig. 1), and 3 (13%) had penile clitoris with 'penoscrotal hypospadias' and complete fusion of the labia majora (Prader type 4; Fig. 2). Five cases of the Prader type 3 and all 3 cases of Prader type 4 were reared as male prior to presentation.

Table 1
Prader Type and Gender at Presentation among the 23 Patients with VEG

Prader Type	n	Gender at Presentation	
		Female	Male
Type 1	3	3	-
Type 2	6	6	-
Type 3	11	6	5
Type 4	3	-	3
Type 5	-	-	-
Total	23	15	8

Fig. 1. A 4-year-old with clitoral hypertrophy, single urogenital orifice, and posterior fusion of labia majora managed during the study period.

The genital anomalies were noticed at a mean age of 1 month (range 2 days-4 months). Only 3 (13%) patients presented within 1 month of noticing the anomalies. The reason for delayed presentation among the remaining 20 patients was delayed referral in 14 (70%), and ignorance of the problem in 6 (30%).

Evaluation

Barr body estimation was positive in 11 (48%) patients and negative in 12 (52%). Pelvic ultrasonography identified the uterus in 10 (44%) cases, but did not identify uterus, fallopian tubes or ovaries in 13 (57%). Ultrasonography did not demonstrate adrenal enlargement in any of the 23 patients. Overall, 13 (56%) patients had pelvic minilaparotomy that showed normal sized uterus, fallopian tubes and ovaries. Blood electrolytes revealed normal values for sodium, potassium, chloride, and bicarbonate in all cases. Only 4 patients had estimation of serum ketosteroids which showed elevated 17-hydroxyprogesterone. The rest of the cases could not undertake it because the facilities were not available when they presented. Genitography was required to further define the anatomy of the vagina in 5 patients.

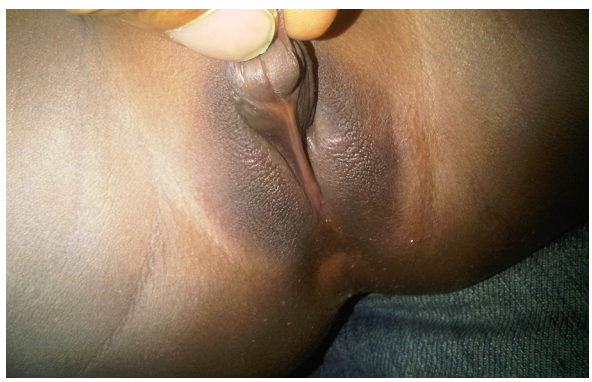

Fig. 2. A 3-year-old with penile clitoris, 'penoscrotal hypospadias' and complete fusion of the labia majora (Prader type 4) managed during the study period. The patient was initially reared as a male before presentation.