A posterior-based omega-shaped flap vaginoplasty in girls with congenital adrenal hyperplasia caused by 21-hydroxylase deficiency

L.G. FREITAS FILHO, J. CARNEVALE, C.E.R. MELO, M. LAKS and M. CALCAGNO SILVA
Paediatric Urology, Hospital Infantil Darcy Vargas, São Paulo, Brazil

Accepted for publication 19 September 2002

OBJECTIVE

To describe a technical modification for constructing a vagina in girls with congenital adrenal hyperplasia caused by 21-hydroxylase deficiency, using a narrower skin base but rich subcutaneous tissue, aiming to obtain both longer and larger vaginal segments with better cosmesis of the external genitalia.

PATIENTS AND METHODS

From August 1997 to February 2001, 28 girls (aged 5 months to 17 years) had a neovagina constructed using a posterior-based omega-shaped flap. Twenty-six patients had a low vagina entering into the urogenital sinus and two had a high vagina that entered the urogenital sinus. In those with a high vagina the flap procedure was combined with the Passerini-Glazel technique. All the patients were scheduled for vaginal 'calibrations' during the first year after surgery and, according to the result, would then undergo vaginal dilatation.

RESULTS

Six children were re-operated; five had plastic surgery to correct genital folds that had regained a scrotal aspect, whereas one with a high vagina developed a urethral stricture, with urinary dribbling and infection, and had the urethra reconstructed. These six children are currently well. No hormone therapy was given to one child for 1 year who is scheduled for further surgery for a re-virilized clitoris. Two patients were lost to follow-up. Up to the last visit, 19 girls had not developed a vaginal stricture and the cosmesis of their external genitalia was deemed good.

CONCLUSION

The posterior-based omega-shaped flap enabled both the construction of wider vaginal segments with a low risk of developing stenosis in those with a low vagina, and increased vaginal dimension when associated with the Passerini-Glazel technique for those with a high vagina. However, despite good cosmesis of the external genitalia, the follow-up is too short to confirm whether this technique will meet all the expectations.

KEYWORDS

adrenal hyperplasia, congenital, genitalia, hermaphroditism, vagina

INTRODUCTION

Congenital adrenal hyperplasia (CAH) caused by 21-hydroxylase deficiency accounts for most cases of female pseudohermaphroditism. This enzyme is encoded by a gene on chromosome 6 and is inherited in an autosomal recessive manner, whereby the cytochrome P-450 enzyme is defective.

For surgical treatment three main issues must be considered, i.e. the proper timing to correct the virilization, precise knowledge of the anatomical variations and detailed technical planning. The degree of masculinization of the external genitalia is thought to be related to the time and amount of androgen production in the fetus, which can be better appreciated using pan-endoscopy to determine the extent and calibre of the urogenital sinus, the site of implantation of the vagina and urethra, and their distance to the perineum [1]. Two distinct groups are identified; a small group of extremely virilized girls, with advanced phallic development and a urogenital sinus with the meatus extending at its tip, resembling that of a neonatal boy with an undescended testis; and a larger group with different levels of virilization. In the latter the meatus of the urogenital sinus is generally at the base of a phallus featuring intense chordee. In the more virilized cases, endoscopy often shows a urogenital sinus resembling the male urethra, and a 'verumontanum' either appearing normal for a male or somewhat flattened, at the bottom of which there is a small vagina [2]. The urogenital sinus of the less virilized girls is wider, with clearly differentiated urinary and vaginal openings, where a fairly normal hymen can sometimes be present.

We report a series of 28 children with CAH caused by 21-hydroxylase deficiency who were treated using a posterior-based omega-shaped flap vaginoplasty, where a technical modification provided a narrower posterior-based skin flap with rich subcutaneous tissue, enabling the construction of longer and larger vaginal segments, and providing better cosmesis of the external genitalia.

PATIENT AND METHODS

From August 1997 to February 2001, 28 patients with CAH (aged 5 months to 17 years) were treated using the posterior-based omega-shaped flap vaginoplasty. All patients had the salt-losing form of CAH; 26 had a low vagina and two a high vagina entering the urogenital sinus. Whenever possible, as they were referred to our institution, the children underwent surgery in their first year of life, in an effort to adjust them as early as possible to their rearing sex. Nineteen children had both a clitoroplasty, as described by Mollard *et al.* [3] *and* a vaginoplasty at the same intervention. Nine children had already undergone clitoroplasty elsewhere, and six required a renewed clitoral

FIG. 1. ***a,*** *Moderate feminization (low form).* ***b,*** *Construction of a posterior-based omega-shaped flap.* ***c,*** *Skin incision.*

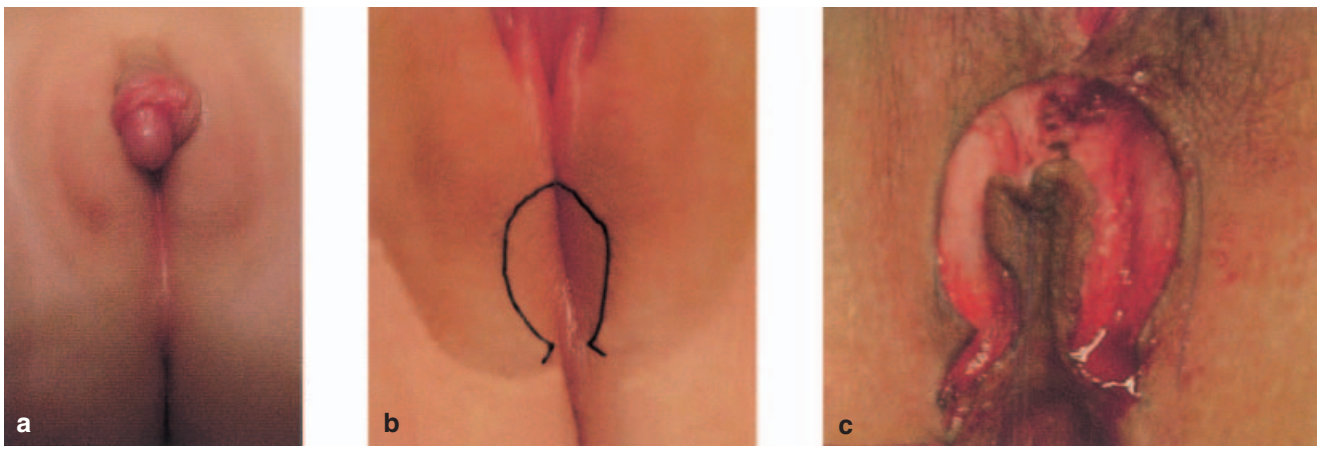
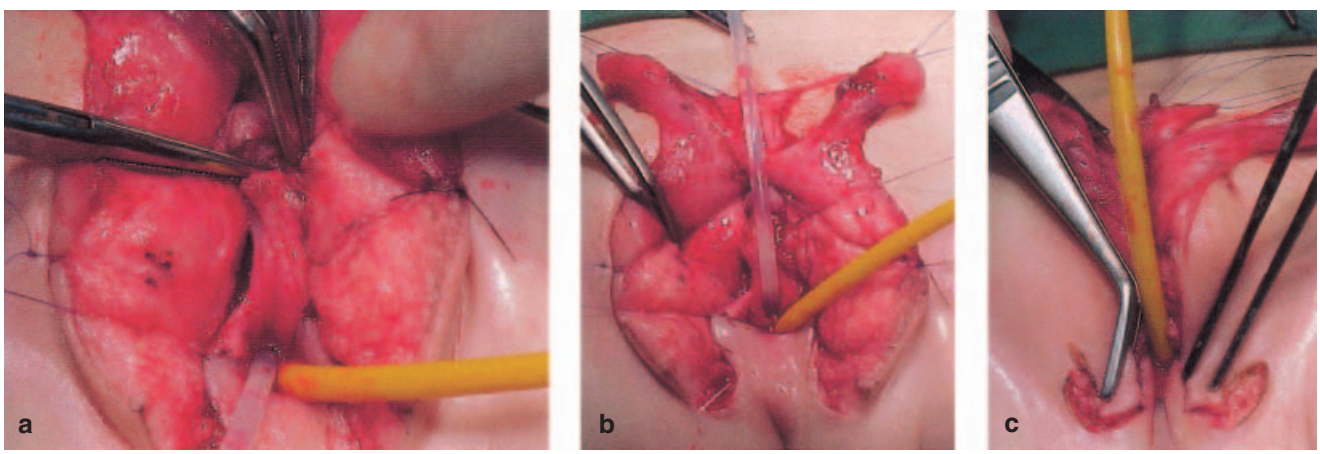


FIG. 2. ***a*** *and* ***b,*** *Urogenital sinus anastomosis with the posterior-based omega-shaped flap;* ***c,*** *Creation of the labia majora.*



reduction. Pan-endoscopy was used before surgery but at the same procedure in all patients; in the two children with the high form, the urogenital sinus showed an apparently normal 'male verumontanum'. The vaginogram in the two girls with the high form showed no vagina, not even after injection with contrast medium under high pressure; their vaginas could not be detected unless the small opening was explored using an 8 F cystoscope. All the children with the low form had a wide urogenital sinus, with the vagina and urethra clearly differentiated. In those with the high form the vaginoplasty was combined with the Passerini-Glazel technique [4]. Both children with the high form had been reared as males and although they had had dehydration episodes in the first weeks of their lives, the diagnosis was not established until they sought medical care for their 'undescended testis'. One of these children had testicular prostheses implanted elsewhere. The surgical technique in those with the low form is shown in Figs 1–3, and in the high form children in Figs 4–6.

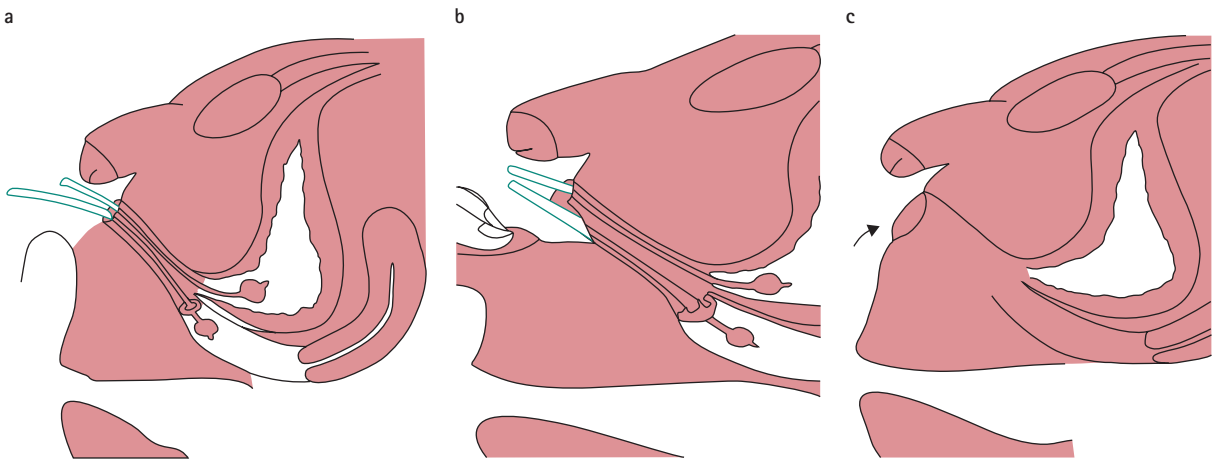
After surgery all children had systematic 'calibrations' of the neovagina under general anaesthesia, using a set of Hegar's dilators of increasing size, with the first 'calibrations' 3 or 4 weeks after surgery and further calibrations at 3, 6 and 12 months, as required. The data for all the children are shown in Table 1.

RESULTS

Six children required reoperation; one with a high form presented with urinary dribbling, incontinence and infection in the first few months after surgery. A urethral stricture was confirmed and the urethra was reconstructed by the abdomino-perineal route. She is currently well at 2 months after surgery. Five children were re-operated to reduce the labia majora, which had regained a scrotal form. A variation of the technique described by Roberts and Hutson [5] was used; currently we consider that almost all of the dark skin of the genital folds should be resected and that the subcutaneous tissue be preserved at the first intervention (Fig. 7).

One of the children discontinued treatment for 1 year and her clitoris was re-virilized because she received no adrenal hormones. She is scheduled to undergo surgery to have her clitoris corrected but interestingly, despite re-virilization of the phallus, she has developed no vaginal strictures to date. Two patients were lost to follow-up. The cosmesis of the external genitalia was deemed satisfactory in 19 children.

FIG. 3. 'Cutback' vaginoplasty with the posterior-based omega-shaped flap.



No./date of birth	Hegar, F initial	Hegar, F last	Date of surgery	re-operation
1/6-7-96	8	10	8-21-97	
2/6-4-87*	16	22	10-30-97	6-22-99
3/4-22-97	5	12	11-3-97	
4/1-11-93*	7	12	1-20-98	
5/3-27-97	5	10	4-14-98	
6/10-3-95*	8	10	5-28-98	
7/11-24-91	19	20	9-24-98	
8/12-21-83	13	20	10-6-98	
91-7-88*	10	12	10-8-98	
10/5-29-97†	–	–	10-17-98	
11/5-11-89*	9	20	10-26-98	
12/8-25-81*	20	20	1-4-99	
13/9-29-98	7	11	2-9-99	
14/10-4-98	10	11	4-20-99	
15/9-5-97	7	12	5-18-99	
16/4-18-97	10	14	7-1-99	
17/12-12-95	8	13	7-8-99	5-4-2000
18/6-16-94*	13	17	8-5-99	12-19-2000
19/12-24-90	8	18	11-4-99	11-21-2000
20/8-1-96†	–	–	11-11-99	
21/4-12-99	9	9	1-13-2000	
22/1-1-94*	10	11	2-8-2000	2-10-2001
23/2-4-97‡	20	20	3-14-2000	1-11-2001
24/7-3-90	20	20	4-16-2000	
25/1-14-89	18	22	5-4-2000	
26/6-23-91‡	11	17	6-29-2000	
27/9-25-98	11	11	8-22-2000	
28/5-26-99	8	13	11-9-2000	

TABLE 1
The clinical data before and after surgery

**Previously operated elsewhere; ‡ High form; †Lost to follow-up.*

All the children underwent initial 'calibration' under general anaesthesia. The four infants undergoing surgery in their first year of life (Table 1) were no longer examined under general anaesthesia as they showed no vaginal stricture at the second 'calibration'; the remaining patients followed the previously established schedule. Only two children have the dilatations undertaken by their parents, and are currently well. None of the children developed strictures over the follow-up. The appearance of children with the low and high form are shown in Figs 7 and 8.

DISCUSSION

CAH is an inborn metabolic error in several enzymes that convert cholesterol into the three main adrenal hormonal types, i.e. sexual steroids, and glyco- and mineralocorticoids. The most frequent cause of CAH is the CYP 21 enzyme (cytochrome P-450$_{c21}$), or 21-hydroxylase deficiency. This enzyme converts the Δ4-steroids, progesterone and 17-hydroxyprogesterone, into deoxycortisone and 11 deoxycortisol. As noted above, the disease has two main clinical presentations. In the low form the virilization is corrected by clitoroplasty associated with a 'cutback' vaginoplasty [7]. In the high form correcting the virilization might include major surgery, where a variety of techniques have been described, e.g. pull-through vaginoplasty [8,9], total urogenital sinus mobilization [10–12] and constructing a neovagina with skin flaps [4]. In both types the genital fold skin is scrotalized and dark, resembling a scrotum, because there is a high serum level of adrenocorticotropic hormone. However, the gonads are never palpable. In the 'cutback' vaginoplasty the flap-vaginoplasty described by Fortunoff *et al.* [7] is usually used, where an inverted U-flap is sutured onto the urogenital sinus previously opened in its ventral aspect. Although such a flap is conveniently vascularized, its wide base allows no imbrication of the lateral flaps that are to form the labia majora, leading to rather inadequate cosmesis. In the posterior-based

omega-shaped flap vaginoplasty the labia majora may be overlapped at the base of the omega (Fig. 4), thus consistently producing better cosmesis of the external genitalia. To maintain flap vitality it is important that a broad base of subcutaneous tissue be spared. This allows for the construction of longer and larger flaps, thus avoiding the common stenosis found at the anastomosis site between the vagina and the flap [12,13]. We consider that the posterior-based omega-shaped flap vaginoplasty, by enabling the creation of longer and wider flaps in the most distal portion, can be used with different techniques when a high vaginal form is present. This avoids major dissection of the pelvic structures and permits a more judicious use of the cutaneous lateral flaps.

Although we systematically used vaginal 'calibrations' in all children, an initial examination in the first year after surgery should suffice to define those who should or should not undergo further dilatation. However, the follow-up is still too brief to confirm whether our technical modification is stable and sufficient; the cosmesis of the external genitalia in the present patients is currently satisfactory, but a longer follow-up should confirm the stability.

Recently Creighton and Minto [14] showed that although many intersex patients have cosmetically adequate external genitalia, some feel aggrieved about not adjusting to their sex of rearing, and for not participating in the decision, as such decisions were made when they were still too young. Although we agree with these comments, we consider that only with more objective and convincing data than are currently available can a child be

FIG. 4. ***a,*** *Intense virilization ('scrotum' with prostheses implantation).* ***b,*** *Cutback vaginoplasty with the posterior-based omega-shaped flap in the high form.*

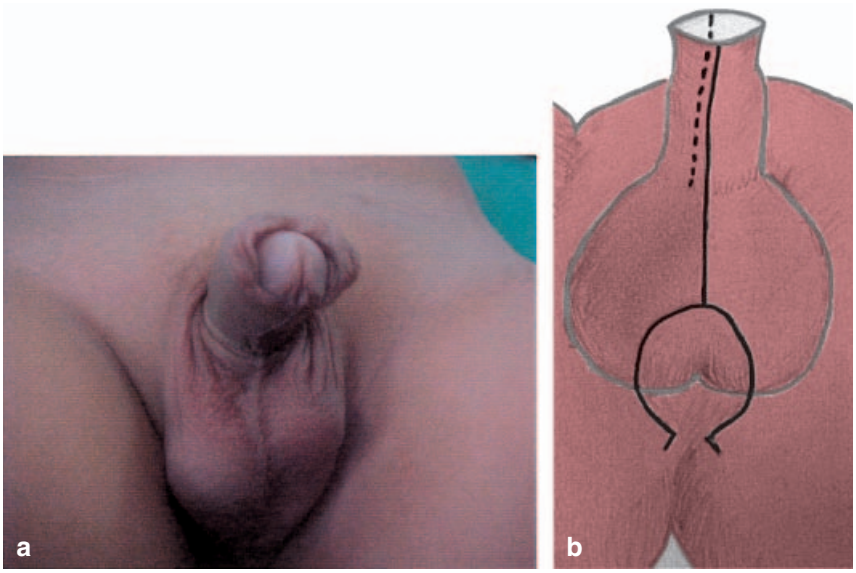
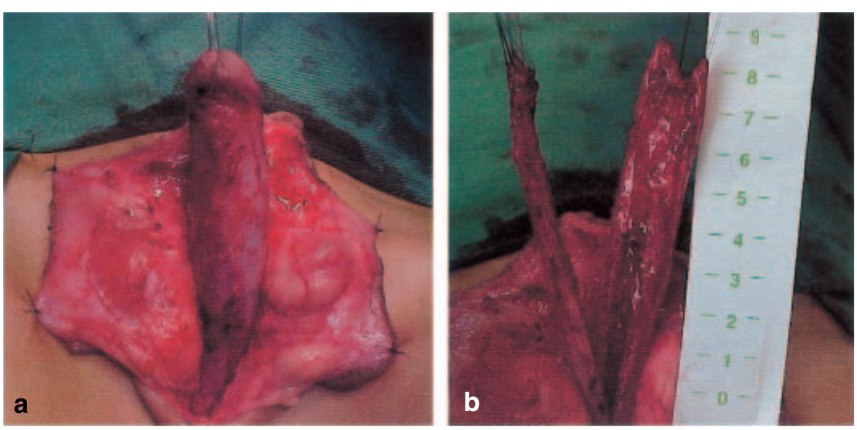


FIG. 5. ***a,*** *The phallus is ungloved.* ***b,*** *The urogenital sinus is separated from the corpora cavernosa.*



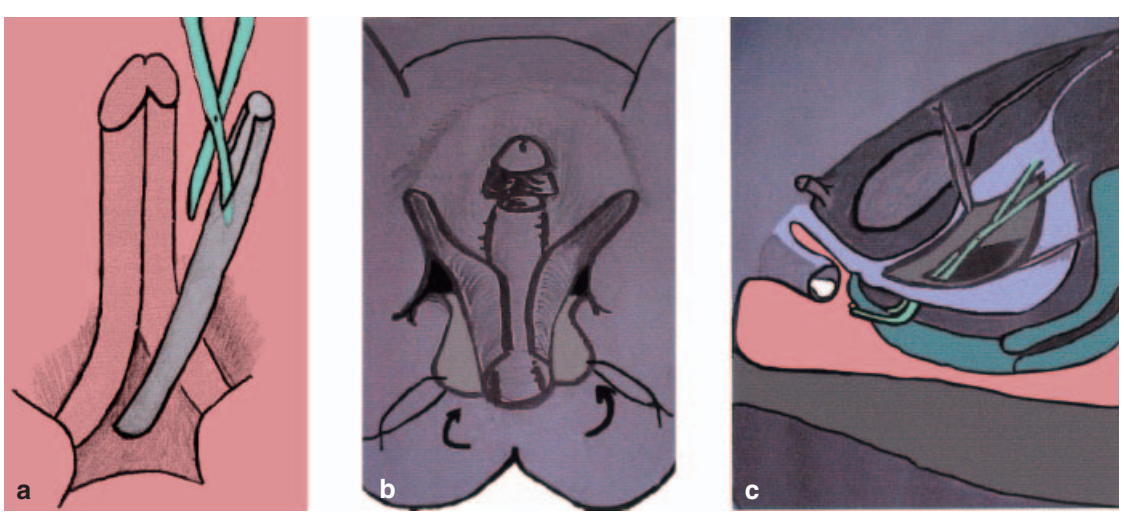


FIG. 6.
a, *The urogenital sinus is incised dorsally.* ***b,*** *After degloving the phallus, both lateral flaps are joined to the urogenital sinus and the omega-shaped flap, and inverted down the perineum.* ***c,*** *Anastomosis of the external neovagina with the vagina by a transvesical approach.*

*FIG. 7. The view before (**a**) and 16 days after (**b**) surgery in the low form.*

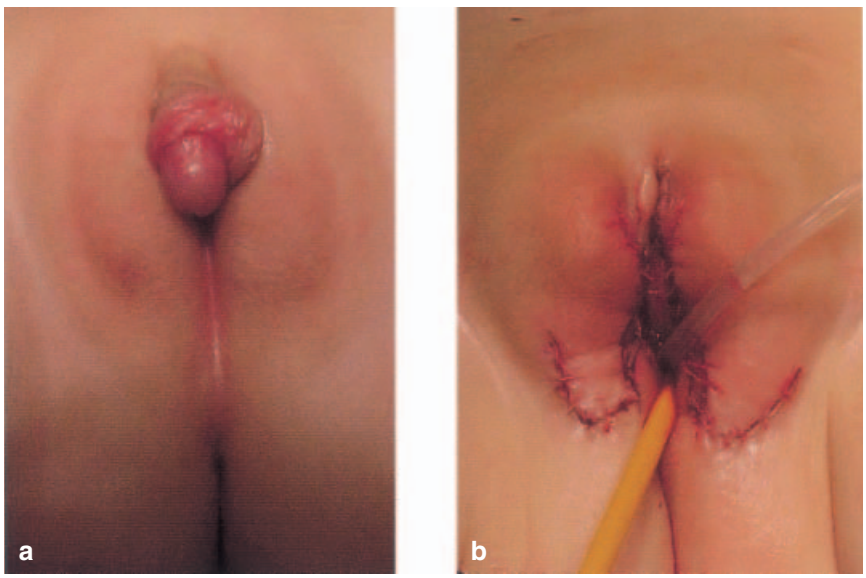
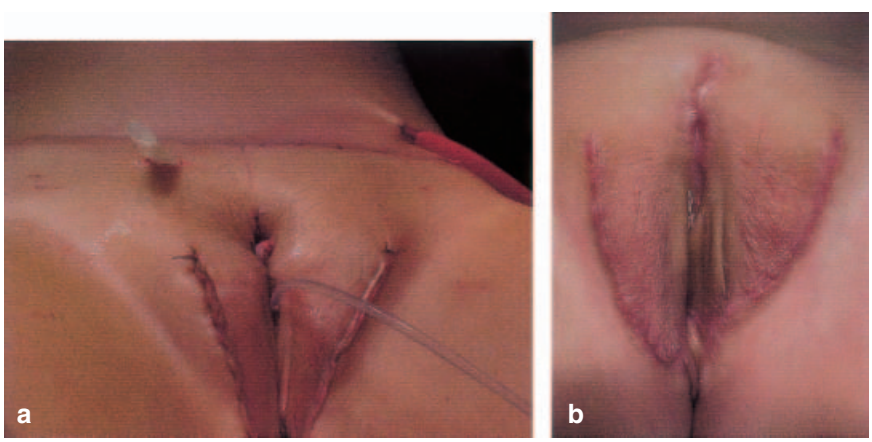


*FIG. 8. **a,** The view immediately after surgery in the patient shown in Fig. 4, and **b,** the view 1 year later.*



allowed to develop with no well-defined sexual identity.

REFERENCES

1 Josso N. Physiology of sex differentiation: a guide to the understanding and management of the intersex child, In Josso N ed. *The Intersex Child. Adolescent Endocrinology*. Vol. VIII. Chapt 1. Basel: Karger, 1981: 1–13

2 Hendren WH, Crawford JD. Adrenogenital syndrome: the anatomy of the anomaly and its repair. Some new concepts. *J Pediatr Surg* 1969; **4**: 49–58

3 Mollard P, Juskiewenski S, Sarkissian J. Clitoroplasty in intersex: a new technique. *Br J Urol* 1981; **53**: 371–3

4 Passerini-Glazel G. A new 1-stage procedure for clitorovaginoplasty in severely masculinized female pseudohermaphrodites. *J Urol* 1989; **142**: 565–8

5 Roberts JP, Hutson JM. Reduction of scrotalized skin improves the cosmetic appearance of feminising genitoplasty. *Pediatr Surg Int* 1997; **12**: 228–9

6 Sheldon CA. Intersex states. In Oldham KT, Colombani PM, Foglia RP eds, *Surgery of Infants and Children: Scientific Principles and Practice*. Vol. II. Chapt 95. Philadelphia: Lippincott-Raven, 1997: 1577–616

7 Fortunoff S, Lattimer JK, Edson M. Vaginoplasty technique for female pseudo-hermaphrodites. *Surg Gynecol Obstet* 1964; **118**: 545–8

8 Hendren WH, Atala A. Repair of the high vagina in girls with severely masculinized anatomy from the adrenogenital syndrome. *J Pediatr Surg* 1995; **30**: 91–4

9 Donahoe PK, Gustafson ML. Early one-stage surgical reconstruction of the extremely high vagina in patients with congenital adrenal hyperplasia. *J Pediatr Surg* 1994; **29**: 352–8

10 Peña A. Total urogenital mobilisation: an easier way to repair cloacas. *J Pediatr Surg* 1997; **32**: 263–8

11 Ludwikowski B, Hayward IO, Gonzalez R. Total urogenital sinus mobilization: expanded applications. *BJU Int* 1999; **83**: 820–2

12 Rink RC, Pope JC, Kropp BP, Smith ER Jr, Keating MA, Adams MC. Reconstruction of the high urogenital sinus: early perineal prone approach without division of the rectum. *J Urol* 1997; **158**: 1293–7

13 Bailez MM, Gearhart JP, Migeon C, Rock J. Vaginal reconstruction after initial construction of the external genitalia in girls with salt-wasting adrenal hyperplasia. *J Urol* 1992; **148**: 680–2

14 Creighton S, Minto C. Managing intersex: most surgery in childhood should be deferred. *BMJ* 2001; **323**: 1264–5

Correspondence: L.G. Freitas Filho, Rua Batista Cepelos 87 – Ap 61, 04109–120, São Paulo, Brazil.
e-mail: lufrei@zaz.com.br

Abbreviations: **CAH**, congenital adrenal hyperplasia.