FEMINIZING GENITOPLASTY IN ADRENAL CONGENITAL HYPERPLASIA: ONE OR TWO SURGICAL STEPS?

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Summary.- OBJECTIVES: The best time to perform a genitoplasty in a Congenital Adrenal Hyperplasia (CAH) girl is an issue that has been discussed extensively. The purpose of this study is to find criteria that may help in the decision.

METHODS: Charts of all patients with diagnosis of CAH with 21 Hydroxylase deficit who underwent genitoplasty in our institution were reviewed (Jan 1996-Dec 2006). Demographic data, surgery performed and outcomes were analyzed.

RESULTS: In the 10 year-period, 25 patients fit the inclusion criteria; 22 had complete data. All patients were classified based on Prader’s criteria; Prader 2 (n=3), Prader 3 (n=13) and Prader 4 (n=6). Mean age at first surgery was 13.5 months (range 2-89m). In Prader 2 patients, a reduction clitoroplasty with a “cut back” vaginoplasty was performed with no complications. All patients in the Prader 3 group underwent a reduction clitoroplasty. A vaginoplasty was done in 9/13; 5/9 at the same surgery session (4 stenotic) and the other 4 in a 2nd stage with good results; vaginoplasty is still pending for the other 4 girls. In the Prader 4 group, a vaginoplasty pull-through was performed in 4/6 using the posterior sagital approach; one at the reduction clitoroplasty stage which ended stenotic and need dilatations, and the other 3 in a 2nd surgery with a good outcome. The other 2/6 girls are awaiting a vaginoplasty. 22/22 had acceptable results after a mean follow-up of 63 months (range 12-144).

CONCLUSIONS: Congenital Adrenal Hyperplasia (CAH) shows different approaches may be used for different degrees of virilization. For less severe cases (Prader <3) a cut-back may be the surgery of choice for vaginoplasty, while in the more complex cases a flap with pull-through or a posterior sagital procedure could be useful. Based on this series, we recommend performing vaginoplasty in a 2nd stage surgery, avoiding complications and further procedures such as dilatations.

Keywords: Congenital Adrenal Hyperplasia (CAH), Paediatrics, Clitoroplasty, Vaginoplasty.
Resumen.- OBJETIVO: El mejor momento para realizar una genitoplastia en una niña con Hiperplasia Suprarrenal Congénita (HSRC) es un tema que ha sido debatido ampliamente. El objetivo de este estudio es encontrar un criterio que pueda ayudar en esta decisión.

MÉTODO: Se revisaron los datos de todos los pacientes con diagnóstico de HSRC con déficit de 21 Hidroxilasa que se sometieron a genitoplastia en nuestra institución (Enero 1996-Diciembre 2006). Se analizaron datos demográficos, cirugía realizada y resultados.

RESULTADOS: En el periodo de 10 años, 25 pacientes cumplieron los criterios de inclusión; 22 tenían datos completos. Todos los pacientes fueron clasificados basándose en los criterios de Prader: Prader 2 (n=3), Prader 3 (n=13) y Prader 4 (n=6). El promedio de edad al momento de la primera cirugía fue 13.5 meses (rango 2-89m). En los pacientes Prader 2, una clitoroplastia de reducción con una vaginoplastia “cut back” fue realizada sin complicaciones. Todos los pacientes del grupo Prader 3 se sometieron a una clitoroplastia de reducción. Una vaginoplastia fue hecha en 9/13; 5/9 en la misma sesión quirúrgica (4 estenóticas) y los otros 4 en un segundo tiempo, con buenos resultados; en las otras 4 niñas la vaginoplastia está aún pendiente. En el grupo Prader 4, una vaginoplastia “pull-through” fue realizada en 4/6 usando un abordaje sagital posterior; uno al momento de la clitoroplastia de reducción, con resultados estenóticos y necesidad de dilataciones, y los otros 3 en una segunda cirugía con buenos resultados. Las otras 2/6 niñas están esperando una vaginoplastia. 22/22 tiene resultados aceptables luego de un periodo de seguimiento de 63 meses (rango 12-144m).

CONCLUSIONES: La Hiperplasia Suprarrenal Congénita (HSRC) muestra diferentes grados de virilización, por lo tanto distintos abordajes pueden ser utilizados. Para los casos menos severos (Prader <3) un “cut-back” puede ser la cirugía de elección para una vaginoplastia, mientras en los casos más complejos un calzado con “pull-through” o un procedimiento sagital posterior puede ser útil. Basado en esta serie, nosotros recomendamos realizar una vaginoplastia en un segundo tiempo quirúrgico, evitando complicaciones y procedimientos adicionales como las dilataciones.


INTRODUCCIÓN

Congenital adrenal hyperplasia is generated by a set of disorders in one of the five enzymes required for the synthesis of cortisol in the adrenal cortex (1). The most usual disorder is the 21-hydroxylase deficiency; this is present in over 90% of the cases. The deficiency generates virilization of different degrees and is the most frequent cause of genital ambiguity (1,2). Surgical objectives involve three main steps in patients with virilization due to congenital adrenal hyperplasia: Reconstruction of the clitoris, of the labia majora and vaginoplasty in search of adequate aesthetic and functional results. The vesical function must also be adequately preserved, as well as the capacity to develop an adequate sexual and reproductive life as an adult. Based on these premises, several surgery techniques have been developed, where a knowledge of anatomy, degree of virilization, length of the urogenital sinus and urethra, as well as other associated anomalies is essential (3,4).

Clitoroplasty is generally recommended in patients under six months old, but the ideal timing for a vaginoplasty to be performed is still being debated (1,2,5). When urogenital sinuses (SUG) are very short, clitoroplasty and vaginoplasty could be done at the same time (2).

However, when the vagina arrives higher up into the urogenital sinus, vaginoplasty could be done later on, thus obtaining better final results. The purpose of this report is to describe our experience in both early and later reconstruction of urogenital sinuses, and analyze the results searching for point of views which would help to reach a better decision about an adequate surgery timing in different cases.

MATERIALS AND METHODS

This study is a revision of the clinical histories of the patients with congenital virilizing adrenal hyperplasia due to the deficit of 21-hydroxylase, who were treated in our institution between 1996 and 2006.

The degree of virilization was recorded in each case, according to the Prader and the length of the urogenital sinus was measured by cystoscopy. The patients were divided in three groups, according to the degree of virilization and the length of the urogenital sinus. The age of the patients was recorded and the surgery techniques employed for vaginoplasty were analyzed. The long-term results, complications and the need for additional procedures were also recorded.

RESULTS

During this 10-year period there were 25 patients who fitted the criteria to be included in this
study, and complete data was available for 22 of them. The patients were divided in three groups, considering the Prader virilization scale and the length of the urogenital sinus. Group 1 (3 patients) included patients with Prader 2 and mean SUG length of 0.8 cm (range: 0.5 to 1 cm). Group 2 (13 patients) with Prader 3 and mean SUG length of 1.6 cm, (range 1.3 to 2 cm). Group 3 (6 patients) with Prader 4 and mean SUG length of 3.5 cm, (range 3 - 4 cm) (Table I).

All the patients had had full endocrinologic checking, urologic study, renal and urinary tract echography, uretrocystography and were examined under anesthesia.

Clitoroplasty was done in a first surgical stage in all patients, with resection of the corpus cavernosum and conservation of the vascular-nervous bundle. The mean age for this first procedure was 13.5 months in the three groups (2-89 months) and no significant differences appeared among the groups, regarding results and complications.

In the first group, vaginoplasty was carried out at the same surgical time as clitoroplasty and in the three cases it was done using the cut back technique. There were no complications in any of the cases; all the patients show an acceptable aesthetic appearance, and do not need additional procedures.

In group 2, (5/13) vaginoplasty was done at the same surgical time as the clitoroplasty. In these cases, the vaginoplasty was done using a Fortunoff inverted U flap and partial mobilization of the sinus. Of these 5 patients, 4 showed vaginal stenosis, which required dilatations, and in 3 of the 4 cases a new surgical procedure is pending execution, as the dilatation program failed. A satisfactory long-term result was achieved in one patient only, with an acceptable aesthetic result and adequate vaginal caliber. In another four patients of this group, the vaginoplasty was postponed to a second surgery stage at a mean age of 108 months (range 84-114 months). The same inverted U flap technique and partial mobilization of the sinus was employed. Adequate aesthetic and functional results were achieved in the four cases (Figure 1). Vaginoplasty is pending for 4/13 patients of this group.

In group 3, and in one of six patients, the vaginoplasty was done at the same surgery time as the clitoroplasty, using a posterior sagittal block lowering technique. (2, 3, 4). Although this patient showed an

<table>
<thead>
<tr>
<th>Group</th>
<th># Patients</th>
<th>Prader</th>
<th>Mean length sinus (cm.)</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>3</td>
<td>2</td>
<td>0.8</td>
</tr>
<tr>
<td>2</td>
<td>13</td>
<td>3</td>
<td>1.6</td>
</tr>
<tr>
<td>3</td>
<td>6</td>
<td>4</td>
<td>3.5</td>
</tr>
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adequate recovery, it has been necessary to carry out vaginal dilatations. In another 3 patients of this group the vaginoplasty was carried out in a second surgery time, with the same approach, giving satisfactory results as regards appearance, continence and vaginal caliber. Vaginoplasty is still pending for two patients of this group (Table II).

None of the patients developed urinary incontinence and all have a good aesthetic appearance. At present 6 patients are awaiting vaginoplasty in a second surgery step.

DISCUSSION

Congenital adrenal hyperplasia can cause virilization of different degrees, therefore different approaches must be employed to surgically solve these alterations, depending on their severity (1). Although many surgical techniques have been described to approach the urogenital sinus, with mixed results, the best timing to effect a vaginal reconstruction is still being debated.

Concerning vaginoplasty, our experience shows how, with different virilization degrees and length of the urogenital sinus, we have arrived at different results (Table II). Thus, for patients having short SUGs (<1 cm) the clitoroplasty and vaginoplasty were done in a single surgical step, making a posterior cut, and obtaining very good final results in this series.

In patients showing a greater degree of virilization (Prader 3, or length of the sinus >1.3 to 2 cm), approaches have included effecting a partial mobilization of the sinus associated to a Fortunoff flap (9,10). In these cases the follow-up has shown that there is a better evolution in patients on whom the procedure is done in a second surgical time, rather than at earlier ages. These results could be explained by the hormonal increase in these patients at that age, which would enhance the malleability of the tissues and their vascularization (2).

On the other hand, menstruation would help to maintain the permeability of the channel, therefore, added to the hormone changes, this would give a better final result of the procedure (2). The fact that older patients can collaborate with the post operative

<table>
<thead>
<tr>
<th>Prader/mean sinus length</th>
<th>Mean age vaginoplasty</th>
<th>Patients</th>
<th>Good results</th>
</tr>
</thead>
<tbody>
<tr>
<td>2 / 0.8 cm</td>
<td>22 months</td>
<td>3</td>
<td>3 (100%)</td>
</tr>
<tr>
<td>3 / 1.6 cm</td>
<td>9 months</td>
<td>5</td>
<td>1 (20%)</td>
</tr>
<tr>
<td></td>
<td>108 months</td>
<td>4</td>
<td>4 (100%)</td>
</tr>
<tr>
<td></td>
<td>Pending</td>
<td>4</td>
<td>-</td>
</tr>
<tr>
<td>4 / 3.5 cm</td>
<td>26 months</td>
<td>1</td>
<td>0</td>
</tr>
<tr>
<td></td>
<td>156 months</td>
<td>3</td>
<td>3 (100%)</td>
</tr>
<tr>
<td></td>
<td>Pending</td>
<td>2</td>
<td>-</td>
</tr>
</tbody>
</table>
handling, effecting dilatations if they were necessary or using vaginal tampons, could also influence the results.

However, other studies show that adequate results can be obtained at earlier ages (2, 11). Gosalbez et al. (3) carries out a mobilization of the SUG in those cases, substituting the use of the Fortunoff flap, by a flap obtained from the SUG itself. In that study, at the mean age of 3.8 years and showing good results at the time of evaluation, great importance is given to the urethral length for the procedure. As our study is retrospective, this precise data was unfortunately not recorded in all cases.

In longer SUGs (>3cm) and greater virilization degrees (Prader 4), we are using the posterior sagital approach described by Peña (12,13). Although this approach was first described for anomalies of the cloacal type, with total mobilization (12,13), since then one of the main authors (R.Z) has promoted this approach for other anomalies and urologic problems; the results on SUG in this series are comparable to those of the literature (2,3,4,10,11). This approach allows a large exposure of the surgery field and facilitates an adequate mobilization. One patient of this group, on whom vaginoplasty was effect at the same time as clitoroplasty, shows a good aspect, although it was necessary to effect vaginal dilatations as it was too narrow.

The main criticism to be made to this technique is that it could generate urinary incontinence (2, 12). This has not occurred to date in our series, besides, it has been seen that in time and with vesical training the risk of incontinence diminishes (14). Furthermore the preservation of the pubouretral ligation, both in total and partial mobilization gives better results in incontinence (10).

Thus, the long-term results show that the main problem of the patients having had early vaginoplasty is the narrowness of the vagina (5/16 in our series), this interferes with the sexual activity of the patient, therefore it has been recommended to effect it in a second phase, at the onset of puberty, to prevent vaginal stenosis and should dilatation be necessary, it can then be done adequately.

For this same type of patients, it is also possible to carry out a full mobilization of the SUG, which has been done via perineal approach although at older ages than clitoroplasty, there are data published with mobilizations at a mean age of 4 years and with good final results (4); this has also been done in patients of less than 3 years and 5.9 years (16,17).

On the other hand, and being conscious that the aesthetic aspect is difficult to evaluate, the results in our patients are of acceptance by the girls themselves the parents and the surgery team.

In spite of all the procedures carried out and the progress in the management of the pathology,
where a good aesthetic and functional result is sought, some long term studies (5, 7, 15) show that in patients undergoing feminizing genitoplasty due to congenital adrenal hyperplasia, the results are disheartening, as the patients show sexual dysfunction, diminution of sensibility and an unfavorable aesthetic result, therefore there are trends that prefer to postpone all procedures to a later stage (5).

In our practice, we prefer to carry out clitoroplasty at an early age, if possible before 6 months, and the vaginoplasty later on, when conditions are more suitable for it.

CONCLUSIONS

The best timing for a vaginoplasty in congenital adrenal hyperplasia is still being debated. Based on the results of this series, we prefer to effect clitoroplasty at an early age, together with vaginoplasty in the less severe cases, such as Prader 2 or SUGs under 1 cm.

For patients with deeper virilization degrees and/or length of the urogenital sinus >3 cm, we prefer to carry out a vaginoplasty by posterior sagital approach in a second stage, to obtain better aesthetic and functional results and avoid carrying out additional procedures.

REFERENCES AND RECOMMENDED READINGS

(*of special interest, **of outstanding interest)

4. Ludwikowski B, OeschHayward I and González R. Total urogenital sinus mobilization: expanded applications. BJU Int,1999; 83: 820-822
