Best Practice & Research Clinical Endocrinology & Metabolism 24 (2010) 311–324

Contents lists available at ScienceDirect

Best Practice & Research Clinical Endocrinology & Metabolism

journal homepage: www.elsevier.com/locate/beem

10

Surgical options in disorders of sex development (dsd) with ambiguous genitalia

Isabelle Vidal, MD, Chief Resident in Paediatric Surgery [a], Daniela Brindusa Gorduza, MD, Consultant in Paediatric Surgery [b], Elodie Haraux, MD, Resident in Paediatric Surgery [c], Claire-Lise Gay, MD, Consultant in Paediatric Endocrinology [d], Pierre Chatelain, MD, Professor of Paediatric Endocrinology [e], Marc Nicolino, MD, Professor of Paediatric Endocrinology [f], Pierre-Yves Mure, MD, Professor of Paediatric Surgery [g], Pierre Mouriquand, MD FRCS(Eng), FEAPU, Professor of Paediatric Urology [h,*]

[a] *Department of Paediatric Urology and Surgery, Hôpital Mère-Enfants, 69677 Bron, France*
[b] *Department of Paediatric Urology and Surgery, Membre du Centre de Référence National sur les Anomalies du Développement Sexuel, Hôpital Mère-Enfants, 69677 Bron, France*
[c] *Department of Paediatric Urology and Surgery, Hôpital Mère-Enfants, 69677 Bron, France*
[d] *Department of Paediatric Endocrinology, Membre du Centre de Référence National sur les Anomalies du Développement Sexuel, Hôpital Mère-Enfants, 69677 Bron, France*
[e] *Claude-Bernard University, Lyon I, Department of Paediatric Urology and Surgery, Membre du Centre de Référence National sur les Anomalies du Développement Sexuel, Hôpital Mère-Enfants, 69677 Bron, France*
[f] *Claude-Bernard University, Lyon I, Department of Paediatric Urology and Surgery, Membre du Centre de Référence National sur les Anomalies du Développement Sexuel, Hôpital Mère-Enfants, 69677 Bron, France*
[g] *Claude-Bernard University, Lyon I, Department of Paediatric Urology and Surgery, Hôpital Mère-Enfants, 69677 Bron, France*
[h] *Claude-Bernard University, Lyon I, Department of Paediatric Urology and Surgery, Membre du Centre de Référence National sur les Anomalies du Développement Sexuel, Hôpital Mère-Enfants, 69677 Bron, France*

Keywords:
feminisation
masculinisation
hypospadias
urethral plate
urethroplasty
congenital adrenal hyperplasia
mixed gonadal dysgenesis
ambiguous genitalia
vaginoplasty

SUMMARY

Disorders of sexual development (DSD) include three main groups of patients: (1) The virilised 46,XX DSD essentially represented by congenital adrenal hyperplasia (CAH) ; (2) The undervirilised 46,XY DSD essentially represented by hypospadias; and (3) the chromosomic jigsaws essentially represented by mixed gonadal dysgenesis. It is in this last group that gender assignment remains a difficult decision involving various indicators, which can be split into four categories: (1) the inside sex (i.e., genes, hormones and

* Corresponding author. Department of Paediatric Urology, Hôpital Mère-Enfants, Groupe Hospitalier Est, 59, boulevard Pinel, 69677 Bron Cedex, France.
E-mail address: pierre.mouriquand@chu-lyon.fr (P. Mouriquand).

1521-690X/$ – see front matter
doi:10.1016/j.beem.2009.10.004

clitoral reduction
perineoplasty
vaginal substitution
sexual identity
individual identity
social identity
behavioural identity

target tissues); (2) the outside sex (i.e., anatomy of genitalia including size of the genital tubercle, mullerian cavity and potential adult height of the patient); (3) the functional sex (i.e., potential sexuality and fertility); and (4) and the social sex (i.e., the cultural medium in which the child is brought up). The challenge is to outline the future individual identity of the child in the post-natal period using these indicators. Current evolutions of surgical techniques of 'feminisation' and 'masculinisation' are described as well as their outcomes.

Introduction

Patients with abnormal genitalia can be divided into three main groups: (1) The first group is that of the 46,XX patients who are virilised and are essentially represented by patients with congenital adrenal hyperplasia (CAH). All 46,XX CAH patients are brought up as females in Western countries. There are few situations where the individual is 46,XX, not CAH, for whom gender assignment is a thorny issue. It is essentially the ovotesticular DSD and the 46,XX SRY-negative individuals with asymmetrical genitalia (46,XX testicular dysgenesis). (2) The second group is the undervirilised 46,XY patients. The vast majority of these is represented by unlabelled (the so-called 'idiopathic') hypospadiac patients with various degrees of hypoplasia of the penile ventral tissues (ventral radius). Few of them have an identified partial androgen insensitivity syndrome (PAIS). Those with a complete androgen insensitivity syndrome (CAIS) have a female phenotype and are raised as females. (3) The third group is represented by patients with a chromosomic mixture, essentially the mixed gonadal dysgenesis 45,XO/46,XY for whom gender assignment is discussed. In practical terms, the most difficult cases to handle are those where gender assignment is an issue, knowing that no decision taken can be fully satisfactory.

The team challenge during the neonatal period is to use the tools available to make the least bad choice in order to select a gender able to match the individual identity (II) of the child (which is invisible during this period), the social identity (SI) (which is the way the 'society' looks at the individual and the only tangible identity approachable after birth) and the behavioral identity (BI), which is not formed yet. The team is composed of endocrinologists, geneticists, biologists, surgeons, psychologists and, above all, the parents. The tools we have can be categorised into four groups: (1) The 'inside sex', which is represented by the genes, gonads, hormonal machinery and tissue targets of the patients. These indicators are well established and play a major role in the gender assignment. (2) The 'outside sex', which is the visible part of the iceberg defined by the dimensions of the genital tubercle and the possible presence of a retro-urethral mullerian cavity (ultrasound scan). The potential adult height is considered by some as a useful criterion, which may tip the scales towards the feminine gender if the future height is expected to be short. As a general rule, it is surgically easier to create a penetrative conduit than a penetrating organ, although techniques of phalloplasty have recently made considerable progress. (3) The 'functional sex' is the potential ability of the individual to have sexual relations and to procreate. (4) The 'social sex' is also essential and involves the cultural medium in which the child is going to be raised. The parents and especially the mother's feelings about the situation are of utmost importance to take the ultimate decision. A few years ago it was said that gender assignment could be delayed until later in childhood[1] when the child expresses more clearly his or her II. This attitude has raised a lot of controversies, and it is currently accepted by most that the assignment should be done early in life to avoid unbearable situations for the parents and the child.

Surgical options in the virilised female 46,XX

The anatomy of the virilised 46,XX individual needs to be briefly described (Figs. 1 and 2): The vaginal cavity is connected to the posterior wall of the urethra, which is open near the base of the