DISORDER OF SEX DEVELOPMENT: DESCRIPTION OF 2 CASES OF MALE PSEUDOHERMAPHRODITISM

Varetti C., Burgio A., Di Maggio G., Meucci D., Garzi A., Messina M.

Division of Pediatric Surgery, Department of Pediatrics, Obstetrics and Reproductive Medicine University Of Siena

Introduction. "Genetic errors" with malformations and hormonal alterations result in a broad spectrum of disorders of sexual identity difficult to classify for etiopathogenesis and anatomy. Male pseudohermaphroditism (PEM) is the result of defects in masculinisation, persistence of mullerian ducts or genital malformations. In these patients, there is always a 46 XY karyotype, male gonads with the external genitalia and phenotype completely or partially female. The authors describe 2 cases of PEM. *Patients and Methods.* RS and HNA, 9 and 15 years old. Both had 46 XY karyotype and female phenotype with the presence of penile clitoris and small vagina. The urethral meatus was positioned at the base of the penile clitoris and upper vagina and testicles, were located at the proximal portion of the inguinal canal.Patients were subjected to MRI showing absence of uterus and ovaries. The endoscopy described a small vagina dead-end about 4-5 cm long. Complete resection of the vagina, bilateral orchiopexy, correction of perineal hypospadias with urethral tubulization, reconstruction of the scrotum, with the results of penoscrotal hypospadias were performed. *Discussion.* Sexual differentiation is the process between VII and XII weeks of gestational age leading to formation of male or female fetus from embryo potentially bisexual. Brain Code gender is bipolar: male or female. In the sexual disorders there is a discrepancy between anatomical sex and the brain code. Genital malformation with a direct mechanism (external genitalia) and indirect (prenatal pathological imprinting on all the receptors of target organs) may exacerbate the brain deterioration of the code resulting in disease patterns difficult to manage according psychological and surgical aspects. *Conclusions.* Pediatric Surgeon plays a key role in the treatment of genital anomalies. Valutation of pelvic-perineal anatomy is important for the choice of sexual phenotype and related surgical strategy. Early correction of genital abnormality may effectively prevent the child present psychological, behavioral consequences and problems in family and society.

INTRODUCTION

Intersex disorders are a very complex pathology. In order to clarify its causes it is important to revert to the genetic conditions and regularities of embryological development. The genital anomalies are mostly determined by chromosomal or endocrinic disorders or by impaired biochemical processus. Genetic errors with malformations and hormonal alterations result in a broad spectrum of disorders of sexual identity difficult to classify for etiopathogenesis and anatomy (1). Moreover psychosexual development is influenced by multiple factors such as exposure to androgens, sex chromosome genes, and brain structure, as well as social circumstance and family dynamics (2, 3). Male pseudohermaphroditism occurs when male genitalia are not developed in genetically male individuals (46 XY); the result is female phenotype.

We describe two cases of male pseudohermaphroditism.

PATIENTS AND METHODS

We report two cases, 9 and 15 years old both with 46 XY karyotype and female phenotype with the presence of penile clitoris and small vagina.

In addition to history and physical examination, the diagnostic studies included chromosomal analysis, blood steroid measurement, pelvic ultrasound, MRI, cystoscopy and laparoscopy. The patients were evaluated and treated by a team of paediatric surgeon, endocrinologist, and paediatric psychiatrist. Sexual identity of child and psychiatric evaluation were regarded as the most important factors of sex assignment.

In both the patients the history was characterized by consanguinity through the parents; in particular the patient of 9 years old had a brother with the same sex disorder development. At clinical exam, urethral meatus was positioned at the base of the penile clitoris and upper vagina; testicles were located at the proximal portion of the inguinal canal (Fig.1,2). Patients were subjected to MRI showing absence of uterus and ovaries (Fig.3). The endoscopy described a small vagina dead-end about 4-5 cm long. The endocrinologyc tests were normal for the age and sex development. Both the patients were raised as male because of developed male sex identity. Complete resection of the vagina, bilateral orchiopexy, correction of perineal hypospadias with urethral tubulization, reconstruction of the scrotum, with the results of penoscrotal hypospadias were performed. After 6 months the reconstructive surgical procedure for masculinising genitoplasty was completed for the patient of 9 years old.

DISCUSSION

Sexual differentiation is the process between VII and XII weeks of gestational age leading to formation of male or female fetus from embryo potentially bisexual. Overlooking the wide variety of congenital genital anomalies it is obvious that the pathology is complex,

Correspondence to:
Prof. Mario Messina, Division of Pediatric Surgery, Dept of Pediatrics, Obstetrics and Reproductive Medicine.
University of Siena. Policlinico "Le Scotte" - Viale Bracci - 53100 - Siena - Italy
Telefono: +39 577 586501 - Fax: +39 577 586174
E-mail: messinam@unisi.it

diagnosis is difficult and in most situations a long treatment is required (1, 4). The physicians are not always able to establish the diagnosis immediately after birth, especially if the external genitals appear normal or with a known pathology such as hypospadias or cryptorchidism. Moreover sometimes, when the family history is precisely collected, some of genital anomalies could be suspected. Brain Code gender is bipolar: male or female during gestational age. In the sexual disorders there is a discrepancy between anatomical sex and the brain code. For the management of intersex condition it is necessary evaluation and treatment by a team of paediatric surgeon, endocrinologist, and paediatric psychiatrist. Genital malformation with a direct mechanism (external genitalia) and indirect (prenatal pathological imprinting on the receptors of target organs) may exacerbate the brain deterioration of the code resulting in disease patterns difficult to manage according to psychological and surgical aspects (5). Certain examinations and tests should be performed, such as: karyotyping, ultrasound of internal genitals, and if necessary CT and MRI, urine and blood tests for hormonal levels, and endoscopy before surgery. It is important to look at the urinary tract because often there are associated anomalies (1, 6). In children sex reassignment is almost never undertaken unless the diagnosis is established early enough, usually during the first few months of life and if all possible changes are accepted by the family. The management of these pathologies became more difficult for the patient during prepuberal and puberal development. Usually the sex of rearing depends on genetical and physiological criteria and on the prognosis of life quality in the future. In all other cases surgical and medical treatment is applied to emphasize or encourage the development of the appropriate sex features to avoid anatomical discrepancy and helping to confirm psychological identity (1,7,8).

CONCLUSIONS

Paediatric Surgeon plays a key role in the treatment of genital anomalies. Valutation of pelvic-perineal anatomy is important for the choice of sexual phenotype and related surgical strategy. Many surgeons recommend surgical treatment during the first 6 months after birth (1,2). Early correction of genital abnormality may effectively prevent the child present psychological, behavioral consequences and problems in family and society.

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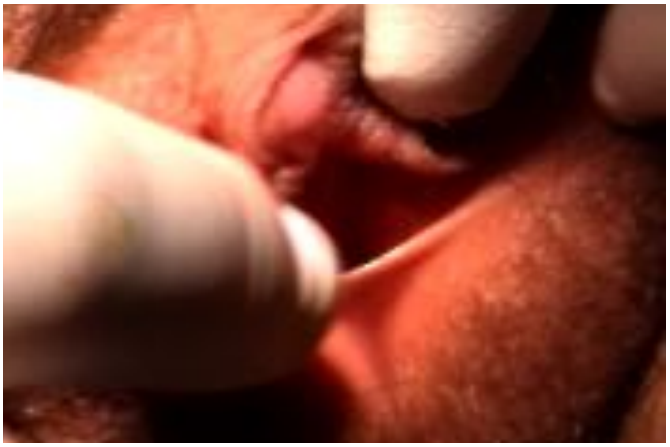

Fig.1: Presence of penile clitoris and small vagina

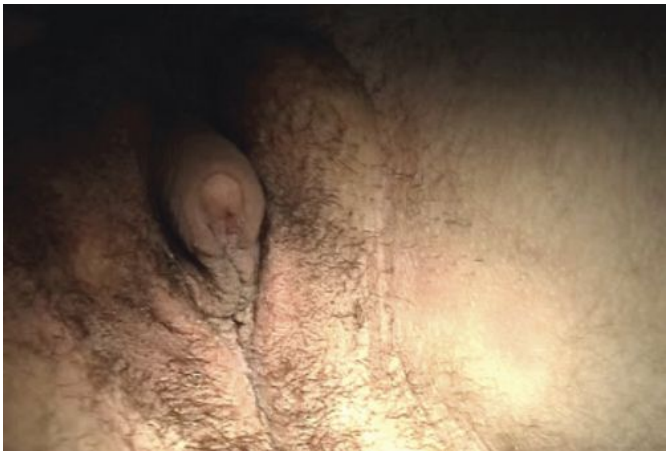

Fig.2: The urethral meatus was positioned at the base of the penile clitoris.

Fig.3:
MRI: absence of uterus and ovaries.