CASE REPORT

AMBIGUOUS EXTERNAL GENITALIA OF AN ADOLESCENT REARED AS FEMALE

Ranabir Pal*, Ankur Barua*, Pradip K Mohanata, Amrita Gosh*****

Departments of Community Medicine, Surgery, ***Pharmacology*

Sikkim Manipal Institute of Medical Sciences 5th mile, Tadong, Gangtok, Sikkim-737102. India

Abstract: *A 12-year-old adolescent, with ectomorphic innocent feminine appearance, presented with ambiguous genitalia with associated congenital absence of right thumb, scoliosis, severe anaemia, primary amenorrhea. She had normally developed breast without any other secondary sexual charactenshcs. She complained of recurrent episodes of abdominal pain and burning micturition with fever for the last 3 years. There was no vaginal orifice. In the genital area there was a pair of ill developed labia majora fused at upper end with a whirling pattern containing inside the urethral opening. The lower end of genitalia had loose skin folds containing adequate amount of rugae with a raphe mimicking the texture of scrotal skin. Ultrasound imaging studies confirmed that she had structures like ovary and uterus with hematometra. Renal scan confirmed agenesis of the left kidney and hypo-plastic right kidney with parenchvmal insufficiency.*

INTRODUCTION

Ambiguous genitalia is a congenital physical abnormality where the outer genitals do not have the typical appearance of either sex; it is difficult to classify the infant (by physical examination) as male or female. So, inter sex disorders associated with phenotypic males and females may be diagnosed much later. In very rare instances, the physical appearance may be fully developed as the opposite of the genetic sex (e.g., a genetic male may have normal female appearance).[1]

The Mayer-Rokitansky-Kuster-Hauser Syndrome refers to a group of associated findings that include unilateral renal agenesis or ectopia, ipsilateral mullerian defects and vaginal agenesis. Approximately 15% have contralateral vesicourethral reflux. Here, the müllerian agenesis second in frequency only to gonadal dysgenesis as a cause of primary amenorrhea. It can be caused by mutation in the genes encoding anti-müllerian hormone (AMH) or its receptor (AMHR). Women with this syndrome have a 46XX karyotype, secondary sexual characteristics of female and normal ovarian function, including cyclic ovulation, but have absence or hypoplasia of the vagina. The uterus usually consists of only rudimentary bicomuate cords, but if uterus contains endometrium, cyclic abdominal pain and accumulation of blood may occur. One third of men with this syndrome have abnormalities of urogenital tract and one tenth with skeletal anomalies, usually involving the spine.

CASE HISTORY

The patient Nazma Akhter, 12 years old adolescent was brought to kolkata for consultation from a remote village of Chittagong in Bangladesh during January 2006 with the chief complaints of recurrent and sever abdominal pain since last four years, few episodes of buming pain during passing urine since last three years, occasional bleeding from single opening in the genital area as her genitalia was not of quite normal shape and size. There were no pubertal changes. She has no feminine looks with axillary and pubic hair along with smaller breasts in comparison to her other daughters. There was no history of menarche or regular periods except occasional trickling of blood was present along with severe pain in loin that made her bedridden and she had recurrent bouts of low grade fever.

Birth history: Her mother recalled that she had no illness or had not taken any form of medication during this pregnancy which ended with a full-term delivery. The birth history was uneventful as home delivery with no proper ante-natal, intranatal and post-natal cares. This child was not given any vaccine till date and no growth monitoring was done.

Family history: The patient was the third child among six siblings- 2 older and 2 younger sisters and 1 younger brother. There was no history of similar complaints among other family members. In the paternal and maternal tree also there was no similar problem. The mother had reported fetal wastage once but there was no neonatal death in family. There was no history of maternal virilisation, suggesting an androgen—producing maternal tumor. Parents of this patient have never practiced any form of contraceptive methods till date.

On clinical examination the girl appeared scared, anxious, severely pale, and thin built, of medium stature with ectomorphic features, in general with an infantile feminine look. Her weight was 26 Kgs with height of 127 cms, There was absence of right thumb, cyanosis, jaundice, oedema, clubbing. There was scoliosis to left in dorsal region and to the right in lumber region. The abdomen was scaphoid shape with definite tenderness in the hypogastric region. Though the patient gave a history of reeling acute pain in the loin and lower abdomen, but there was no such tenderness or severe pain or mass or organomegaly on superficial and deep palpation in the abdomen. There was no abnormal finding in systemic examination. Rectal examination did not reveal the cervix and uterus to confirm the internal mullerian structures. The anal aperture was apparently normal in appearance.

Secondary sexual character examination: No age specific secondary sexual feature was present viz, no adolescent change in voice, adolescent increase in width of girth of pelvis and buttock, no axillary or pubic hair (Tanner stage 0). There was well developed breast with areola (tanner stage 3).

External genitalia examination: A pair of ill developed labia major like structure was visible. The size and degree of differentiation of the phallus was noted, since variations may represent with an irregular clitoromegaly or hypospadias. A protruding mass similar to hypertrophied clitoris of 1 cm long was overhanging the external genitalia. The external genitalia of this age without any pubic hair and infantile pelvic girth were unusual for this age in an adolescent (Figure-1).

On lithotomy posture, a single aperture was found clinically in the region between mons pubis and anal opening which was clinically noted as external urethral meatus. The latter was almost wholly covered by a structure similar to be called labio-scrotal folds. This could be separated

Correspondence: **Dr. Ankur Barua**, Assistant Professor, Department of Community Medicine, Sikkim – Manipal Institute of Medical Sciences (SMIMS), 5th Mile Tadong, Gangtok – 737 102 India Fax.: 03592-231496, e-mail : ankurbarua26@yahoo.com



Fig. *A pair of ill developed labia majora like structure seen with a protruding mass similar to hypertrophied clitoris with urethral opening.*

apart to note the lone aperture seated deeply. The fold of skin contained adequate amounts of rugae with a raphe. This clinically was similar to texture of scrotal skin has given an appearance of a scrotum without any solid structure like testes. This was unlike fatty smooth feelings of labia majors or minor. Rugose scrotal or labioscrotal folds with increased pigmentation suggested the possibility of increased corticotropin levels as part of adrenogenital syndrome. This fold of skin was fused at upper end with a whirling pattern apparently resembling clitoris. On pressing the meatus, a few drops of blood oozed out with dribbling of urine.

Investigations: Routine hematological studies revealed that the hemoglobin level of the patient was 4.2 g m/dl, MCHC 14.4 percent. Her serum creatinine level was O.7mmol/L.

Imaging Studies: Congenital absence of right thumb and scoliosis to the left in dorsal region and to the right in lumber region was confirmed through radiography. A Renogram with ^{99}mTc-DTPA, dose 4mCi was done and the report revealed; poor uptake in the right kidney and nil uptake in the left kidney.

Renal scan report confirmed that this patient had an agenesis of left kidney along with a hypoplastic right kidney, consistent with insufficiency ' of renal parenchyma.

Abdominal ultrasound was performed and structures resembling uterus and ovaries were noted. Uterus was of normal size with evidence of collection inside (haematometra). Genitography, CT scanning, MR and diagnostic laparoscopy, exploratory laparotomy, gonadal biopsy were advised, but due to financial constraints did not give consent.

Follow-up: This patient was treated for endstage renal failure dialysis was initiated, but could not be continued because of financial constraints; renal transplant was not possible due to lack of donor and financial constraints, the patient expired during the 8th month of follow up.

DISCUSSION

Intersex conditions are among the most fascinating conditions encountered by the clinician. In most cases today, clinicians can promptly make an accurate diagnosis and counsel parents on therapeutic options. However, the paradigm of early gender assignment has been challenged by the results of clinical and basic science research, which show that gender identity development begins in utero. While the techniques of surgical genital reconstruction have been mastered, the understanding of the psychological and social implications of gender assignment is poor.[23,4]

Intersexual individuals are treated in different ways by different cultures. In some cultures intersexuals were induded in larger third gender or gender-blending social roles along with other individuals. In most societies, intersex individuals have been expected to select one sex, and adhere to its gender role. Since the rise of modem medical science in Western societies, intersexuals with ambiguous external genitalia have their genitalia surgically modified to resemble either male or female genitals. But there are increasing calls for some critics had condemned the practice of performing corrective surgery on the genitals of intersexuals as an act of female genital mutilation. Corrective surgery is done purely for aesthetic or social purposes. It may lead to negative consequences for sexual functioning in later life, but many intersex individuals had resented the surgical intervention. Some so had opted for sexual reassignment surgery later in life. [14,5]

Medico-legal-Pitfalls: Treatment for intersex states is controversial; the controversy revolves around issues of gender reassignment. Though some intersex activists and health care professionals have called for a moratorium on gender reassignment and genital surgery until studies have been completed on the long-term effects of such surgery, but many had opposed the proposed moratorium.

Infants born with ambiguous genitalia present a true medical and social emergency. Salt-wasting nephropathy occurs in many of the infants born with ambiguous genitalia. If unrecognized, the resulting hypotension can cause vascular collapse and death. Male infants with this syndrome may be phenotypic ally normal and hence, the diagnosis might be missed. Modem treatment of infants with ambiguous genitalia involves a team-oriented approach. The goal is to provide appropriate medical support and counseling regarding care and therapy.[4,5] Evaluation of ambiguous genitalia requires a multidisciplinary.

Hence, from the medico-legal standpoint, the best approach to managing these cases of ambiguous genitalia is to provide parents with adequate information and guide them to make rational decisions.

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LITERATURE REVIEW

Recovery of renal function after 90 d on dialysis: implications for transplantation in patients with potentially reversible causes of renal failure

Samira Siddiqui, Michael Norbury, Sue Robortson, Alison Almond and Chris Ises. Clinical Transplant 2008,2, 133-137

Late recovery of renal function in patients requiring dialysis is a well recognized but uncommon phenomenon. Moves to increase the number of live donor transplants and the recognition that early transplantation is associated with better graft survival means it is possible that patients who are going to recover renal function may be transplanted unnecessarily. Prospective survey of patients receiving dialysis for more than 90 d in south west Scotland from 1 January 1994 to 31 December 2005. Routine measurement of residual renal function by combined urea and creatinine clearance allowed us to detect late recovery whenever this occurred. Eight of 202 (4%) patients recovered sufficient renal function to stop dialysing after 90-d treatment. The likely cause of the renal failure in five of these patients was atheroembolism. One with atherosclerotic renovascular disease had been stented and would have received a live related renal transplant had his sister not had second thoughts about the procedure. It may be sensible to postpone transplantation in patients with certain types of renal failure, perhaps particularly patients with renovascular disease who have recently undergone a failed revascularization procedure.