Case Report

Management of a 3-year-old child with ambiguous genitalia

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Abstract

True hermaphrodites have characteristics tissue both sex in the form of presence of vagina uterus, fallopian tubes and ovaries as in female and presence of testis seminiferous tubules but penis as in penoscrotal hypospadias and bifid scrotal sac confuses as well developed labia majora. Determination of sex as per parents view in our social set up. The case was provisionally diagnosed as intersex. Total abdominal hysterectomy, bilateral salphingo-oophorectomy and colpectomy were conducted. A 3-year-old child referred by a pediatrician to Nalanda Medical College, child surgical department for investigation and management of his ambiguous genitalia having following features like, urine was not passing from the tip of penis; penis along with apparently looking female genitalia was taken under treatment. Examination under general anesthesia showed a presence of both male/female internal genitalia and gonads with features of external genitalia of both sexes. After total abdominal hysterectomy and human chorionic gonadotrophin treatment growth of phallus and testicle was noted. The child was made male after extirpation of female genitals. This case illustrates that gender correction can be made as per patients consent in adult but at this stage social customs and parents consent dictated the treatment.

Key Words: Hermaphrodite, hypospadias, karyotype, orchidopexy, ovotestis

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INTRODUCTION

The true hermaphrodites have characteristics of both sexes in the form of vagina, uterus, fallopian tubes and ovaries in female, whereas; testis, seminiferous tubules and penis in male. However, penoscrotal hypospadias and bifid scrotal sac confuses as well developed labia majora. Virtually all patients have urogenital sinus and in most cases uterus is present. Approximately, 60% of the true hermaphrodites have 46, XX Karyotype, 33% mosaics with second line containing a Y chromosome (46, XX/46, XY; 46, XX/46, XXY) and 7% 46, XY. This case has karyotype 46, XY. The Incidence of gonadal tumors approximately 10% in 46, XY true hermaphrodites and 4% in 46, XX true hermaphrodites both gonadoblastoma and dysgerminoma have been described.[1] In this condition, the patient can be raised as a female and male both, but in both condition long-term gonadal surveillance ultrasonography (USG) for tumor development would be seem appropriate. Hence, the study was conducted with the aim to determine the sex as per parents view in our social set up in such hermaphrodite conditions in human beings.

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MATERIALS AND METHODS

The case was a 3-year-old child admitted in the pediatric surgical department, Nalanda Medical College and Hospital, Patna, Bihar for investigation and management of his ambiguous genitalia. The complaints depicted that urine was not passing from the tip of penis but through the perineal region since birth. There was swelling in both sides of inguinal region. Testis and scrotum were not visible and presence of penis with apparently looking vagina [Figure 1]. The findings of physical examination had shown consciousness, irritable,

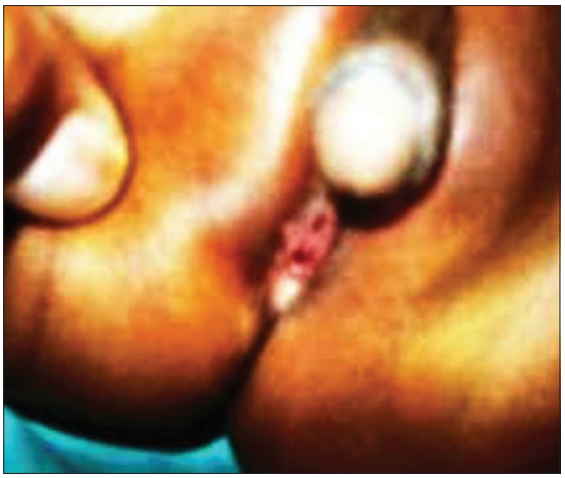

Figure 1: Presence of vagina hypospadias + small penis

aggressive and avoided genital examination. The vitals were normal. The systemic examination showed as respiratory rate-30/min, bilateral crepitations present. Cardiovascular system: S1S2 audible, No added sound. Per abdominal examination revealed – diffuse swelling in both side of inguinal region of size 3 cm × 2 cm, increases in size on head raising test and decreases on flexing and internally rotating the knee joint, impulse on coughing absent, skin over the swelling is normal. The findings of palpation showed: No organomegaly, Tenderness present on palpation of swelling, firm in consistency, no impulse on coughing. The genital examination showed small penis for his age with perineal hypospadias, scrotal rugosity absent, features of external genitalia having resemblance of male and female. Female features were – presence of labia majora, vaginal opening but absent minora. Examination under general anesthesia showed presence of both male/female internal genitalia and gonads with features of external genitalia of both sexes. The Hemogram reports were within normal range. Routine examinations of urine were normal analysis. Chest X-ray was normal. Serum testosterone (T): 91.94 ng/dl (ref range for this age: 10-30 ng/dl) and karyotype was 46, XY. USG of whole abdomen showed B/L undescended testis. The right testis measures 1.2 cm × 0.9 cm, left testis 1 cm × 0.7 cm in inguinal region. There was rudimentary penis along with uterus and without ovaries. All investigations and examinations of the case support in provisionally diagnosis; as a case of intersex. The issue of treatment was taken on principle that we treat these children the way we do (male/female), because this is the way we see all over the world. Most importantly it is the way that the children, themselves are taught to see the world.[2] The most important aspect of the management in true hermaphroditism is gender assignment. Sex assignment should be based on functional potential of external genitalia, internal ducts and gonads, according to the findings at laparoscopy or laparotomy. Unlike patients with most other forms of gonadal dysgenesis, true hermaphrodites have the potential for fertility if raised as females with appropriate ductal structures. Pregnancies have been reported in patients with true hermaphroditism, the majority with the 46, XX karyotype.[3] If the patient is to be raised as female, all testicular and wolffian tissues should be removed. For those patients with an ovary this is straight forward; if an ovotestis is present, surgical cleavage of the gonad with excision of the testicular portion has been performed successfully.[4] They recommend post-operative stimulation with human chorionic gonadotrophin (HCG) to confirm that all testicular tissue has been removed. In some settings, the cleavage plane between testicular and ovarian tissue is unclear, gonadectomy is advisable. When ovarian tissue is preserved, normal ovarian function can occur at puberty, although hormonal replacement may be necessary. Careful surveillance for potential gonadal tumors in the patients raised as females which is also advisable. If a male gender is assigned, as has been most common historically, all ovarian and mullerian tissue should be removed. Consideration should be given to gonadectomy at puberty with appropriate androgen replacement in this setting, given the high risk of malignancy and unlikelihood of male fertility. At the very least, long-term gonadal surveillance; USG for tumor development would seem appropriate. The parental concern was also considered as the social customs prevalent in our society it becomes important to us to understand parental feelings, that what they want this baby to grow as a male or female. Parents of this baby wanted, baby should be given male gender. So we left with no other option, though it was quite easy to give this baby a female gender identity. Opinion of Child Psychologist, NMCH was taken. According to him "the child has no definite orientation of either sex, but due to his external genitalia and upbringing as a male child we would consider him to be grown as a male".

Surgical treatment

Total abdominal hysterectomy, bilateral salphingo-oophorectomy and colopectomy done as a first stage procedure [Figures 2 and 3]. The histopathological examination showed feature suggestive of ovarian tissue with follicles and uterine appendages.

Follow-up

Right side orchidopexy done after 3 months period as a 2nd stage procedure. Post-operatively 3 doses of 1000 IU of injection HCG given intramuscularly. Significant growth of phallus and testicle was noted [Figures 4 and 5].

DISCUSSION

True hermaphrodites are individuals who have testicular tissue with well-developed seminiferous tubules and ovarian tissue with primordial follicles, which may take the form of one ovary and one testis or, more commonly, one or two ovotestis.

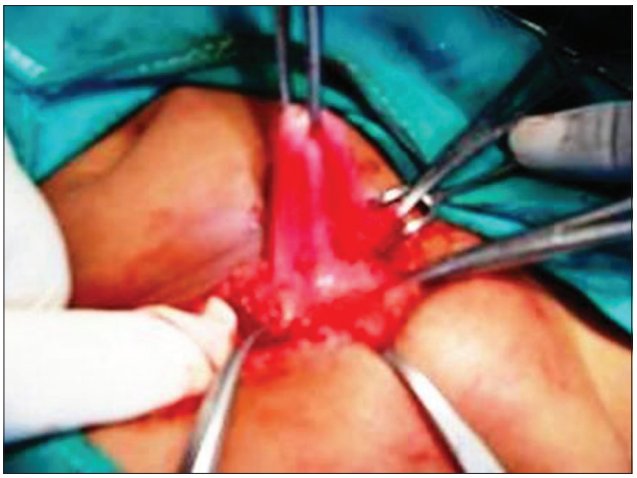

Figure 2: Picture showing ovo-testis and uterus fallopian tubes

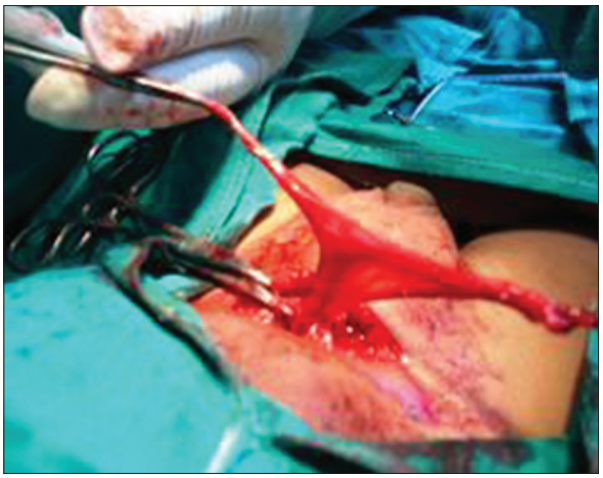

Figure 3: Tubes body of the uterus and service + vagina coming through perineum

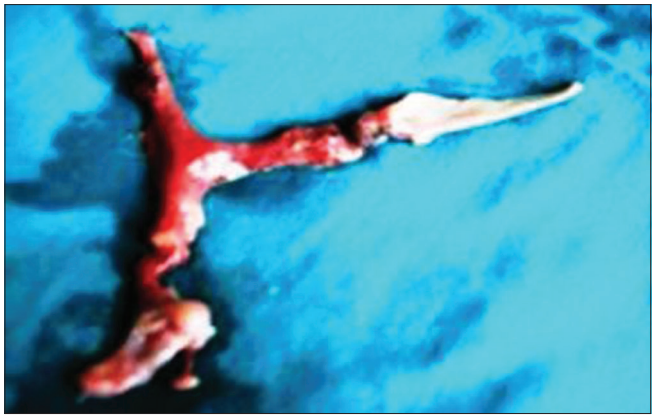

Figure 4: Resected specimen of uterus and its appendages, both ovaries and vagina

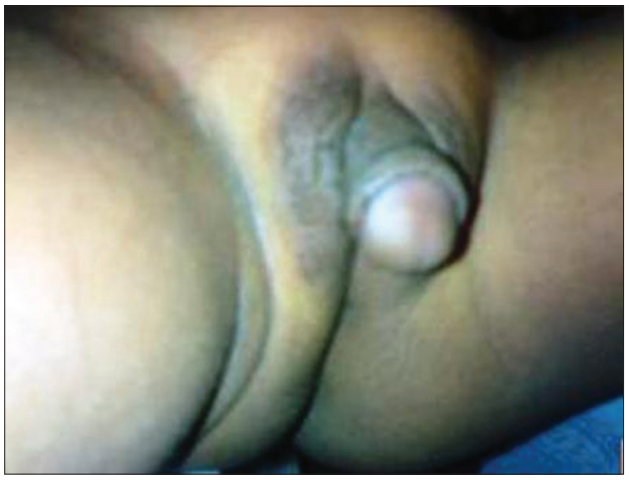

Figure 5: Penile growth after hormone therapy

Both the external genitalia and internal duct structures of true hermaphrodites display gradations between male and female. In most of the patients, the external genitalia are ambiguous but masculinized to variable degrees and 75% are raised as male. Among those raised as male, hypospadias and chordee occur in approximately 80%. Among those patients raised as females, two-thirds have clitoromegaly. Virtually all patients have a urogenital sinus and in most cases a uterus is present.[2] The ovary is found in a normal location, more commonly on the left side. The testis or ovotestis may reside at any point along the path of testicular descent. Testes and ovotestes are more commonly located on the right side. Nearly 60% of gonads palpable in the inguinal canal or labioscrotal folds are ovotestes, which may be clinically suspected on the basis of a difference in firmness at either end the gonad, consistent with polar segregation of ovarian and testicular tissue.[5] Approximately 60% of true hermaphrodite have a 46, XX karyotype; 33% are mosaics with a second line containing a Y chromosome (46, XX/46, XY; 46, XX/46, XXY) and 7% are 46, XY. Chimerism (mosaicism) has been thought to result from fertilization infusion of an ovum and its polar body, infusion of two nuclei, or double fertilization. It has also been suggested that true hermaphroditism may result from hidden mosaicism with a Y cell line. Other studies have demonstrated heterogeneity of Y-specific deoxyribonucleic acid regions detected in patients with true hermaphroditism.[6] This supports a non Y chromosome related mechanism responsible for 46, XX true hermaphroditism, such as mutation in an autosomal or X linked gene involved in sex determination. The study by Berkowitz *et al*. suggested that 46, XY true hermaphroditism may be a form of partial gonadal dysgeneis.[7] According to this theory, a partial defect in testis determination results in both testicular and ovarian development. This is supported by the finding of ovarian stroma in some dysgenetic testis. Just as the differentiation of external genitalia is variable in true hermaphroditism. Differentiation of the internal ducts is also quite variable and is related to the function of the ipsilateral gonad. Fallopian tubes are consistently present on the side of the ovary and a vas deferens is always present adjacent a testis.[7] The ovotestis, which comprises two-thirds of gonads

in true hermaphroditism, is associated with a fallopian tube in two-thirds of cases and with either a vas deferens only or both structures in one-third of cases.

The ovarian portion of the ovotestis is frequently normal, whereas, the testicular portion is typically dysgenetic. Therefore, although ovulation and pregnancy have been reported for female 46, XX true hermaphrodites and male fertility has not been clearly documented. The incidence of gonadal tumors is approximately 10% in 46, XY true hermaphroditism and 4% in 46, XX true hermaphroditism. Both gonadoblastoma and dysgerminoma have been described.[1] This case illustrates that gender correction can be made as per parents consent in adult but at this age social customs and parents consent dictated the treatment. The child was made male after extirpation of female genitals (uterus, fallopian tubes, ovaries and vagina).

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