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HE OR SHE- FEMALE PSEUDO HERMAPRODITE- A CASE REPORT

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ABSTRACT

An Hermaphrodites\ Intersex individual may have biological characteristics of both the male and the female sexes and whose biological sex cannot be classified as clearly male or female and reproductive or sexual anatomy differs from the typical definitions of either sex.

The presence of intermediate or atypical combinations of physical features that usually distinguish Female from Male due to congenital involving chromosomal, morphological, genital and/or gonadal anomalies, such as diversion from typical XX-female or XY-male presentations, *e.g.*, sex reversal (XY female XX -male), genital ambiguity, or sex developmental differences.

1 in 2000 for every births(2,3). While doing supervision "Suverna Arogya Chetana" –School children health checkup programme comes under NRHM, I found this boy aged about 12yrs studying in 7th standard in governament primary school. School teacher noticed since last 4ys.on examination he has breasts like girls, ambiguous genitalia and menstruating since last 4-6 months.

This have been not only a focus of attention of anatomists but also at interest of Genetitians, Embryologists, paediatric endocrinologists, surgeons, physicians, psychiatrists, Forensic experts,socialist because it makes very sensitive issue on the boy"s family and society.

Keywords - Hermoproditism\Intersex "Suverna Arogya Chetana" Genetitians, NRHM, Embryologists, paediatric endocrinologists, psychiatrists, Forensic experts.

INTRODUCTION

An Hermaphrodites \Intersex individual may have biological characteristics of both the male and the female sexes and whose biological sex cannot be classified as clearly male or female.[1] Disorders of sexual development (DSD), formerly termed Intersex conditions, the most fascinating conditions encountered by the practioner [(5)] 1 in 2000 for every births. But some born with subtler forms of sex anatomy variation[(2,3)].

1 in 25000 for every births[(6)]

In my case study 1 in 3500 population. Analysis of worldwide infant screening of 6.5 million newborns found the incidence of CAH to be 1/ 15,000 live births. Frequency was highest in neonates of European Jewish, Hispanic, Slavic, or Italian descent.[(5)]

CASE REPORT

While doing supervision "Suvarna Arogya Chaitanaya" –School children health checkup programme under NRHM in the year 2012 . We found this boy aged about 12 yrs studying in 7th standard in government primary school. School teacher noticed since last 4ys. He has breasts like girls, Ambiguous genitalia. His new complaint is menstruating since last 4-6 months. His walking [style waddling gait] also changed like girls. Voice is also changed slow and low pitch voice but his thyroid profile normal.

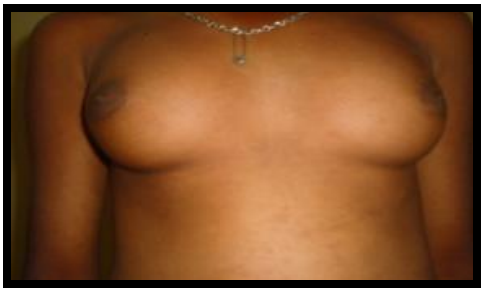
1 in 2000 for every births. But some born with subtler forms of sex anatomy variation.

HISTORY AND OBSERVATIONS

Narrator mother, father and teacher.

This boy aged about 12yrs, while doing school health examination, noticed that he has following features.

Well developing breasts as in girls.



Ambigious genitalia—appears just like female genitals externally

Penis short- Length of phallous-3.7mms

Partially fused genital fold. Hyper pigmentation and mild rugosity seen. Opening of urethra about 3 cms away from root of the phallous. Hypospadiasis, labia majora fused.

No palpable gonads. Scrotal sacs empty on both sides. Anal opening-normal,

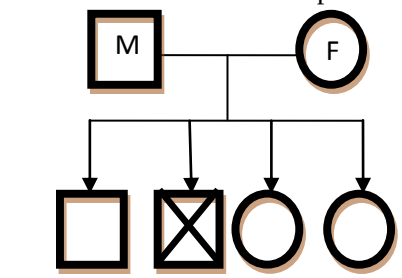
Menstruating last 4-6 months.

PAST HISTORY

Child sits and passes urine but he is not passed from the pallus but below it from the separate opening **.** NOT IMMUNISED.Studies in 7th standard, appropriate for age.

FAMILY HISTORY : Consangui'neous Marriage

NO H\O similar complaints in the family.



- 17yrs 12yrs 10yrs 19yrs

SOCIO ECONOMIC STATUS - Grade IV-Poor

General Physical Examination[GPE] poorlybuilt and nourished, very cooperative. Afebrile.

- P\R—82\mt
- BP----100\60 mm hg

Anthropometry

- Weight- 24.35kgs.
- Height—111cms
- US-LS—56.7cms;54.3cms
- HC-47cms
- CC-53cms

Growth chart

- **PROPORTIONATE SHORT STATURE----Microcephaly**
- CVS- S1 S2 heard. NO murmurs.
- P\A-soft and No organomegaly
- CNS and RS—NAD

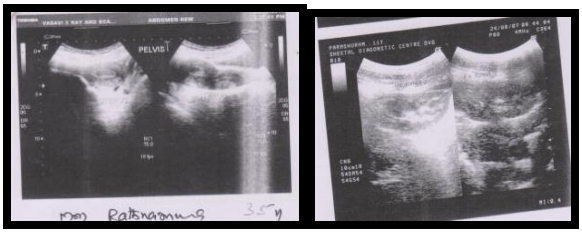
FOLLOWING INVESTIGATIONS DONE

1. Random blood sugar : 84mgs %
2. Random urine sugar : nil %
3. Serum creatinine- : 0.78%
4. Heamoglobin : 10gm.
5. Clotting time : 4’42
6. Bleeding time : 3’24
7. Serology report : HbsAg - negative
   VDRL - non reactive
8. Blood & Rh type -- A positive
9. FBS (FASTING BLOOD SUGAR) : 102mgs\dl
10. FBS (FASTING URINE SUGAR) : NIL
11. GTT-GTU-FASTNG BLOOD : G H Glucose;-Oral 75gms glucose to drinking water..RUS (RANDOM
    BLOOD SUGAR)—118mgs\d
12. RUS (RANDOM URINE SUGAR) : NIL

THYROID TESTS

13. TOTALTRIODOTHYRONINE: T3- 68ngms\dl
14. TOTAL THYROXIN: 2mgms
15 TSH [THYROID STIMULATINGHARMONE] : > 150
16 OH REGISTERONE : [1.2]

ABDOMINAL ULTRASONOGRAPHY *[USG]*



Liver : Both lobes show normal size, shape, contour and echo pattern. No focal mass/ cyst. No dilated Intrahepatic biliary radicals.

Gall bladder *:* Normal size , wall thickness and contents. NO calculus. CBD Normal.

Pancreas*:* Shows normal size and echo pattern.

Kidneys; size, shape, position, contour and echopatteren of both kidneys normal. No dialatation of calyses, pelvis and ureter . No calculus seen in kidneys

PUJ or VUJ SPLEEN: Shows normal size and echo pattern.

No obvious bowel mass . No ascites

No mass seen in the right iliac fossa.

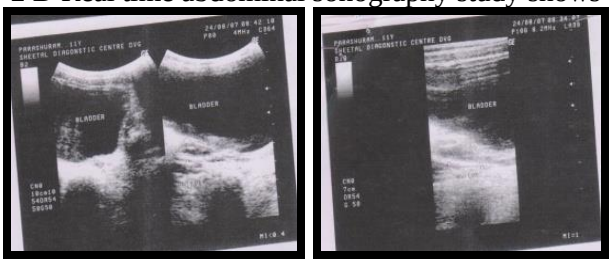
Urinary Bladder- Normal. No calculus or growth.

UTERUS—Anteverted measures 32+20mm and normal.
No fibroids seen. Endometrial thickness10mm
OVARIES – RIGHT -- measures 32+20mm and normal.
LEFT -- measures 32+20mm and normal.

No adnexal mass. No free fluid in POD.

IMPRESSION - NORMAL ABDOMINAL -PELVIC SONOGRAPHY

2 D Real time abdominal sonography study shows



Kidneys-: Both kidneys are normal in size, position and echo genicity.
No hydronephrosis or mass. No obvious renal anomalies.
Right Kidney - 62mm Left Kidney - 63mm
No ureteric dialatation on either sides.

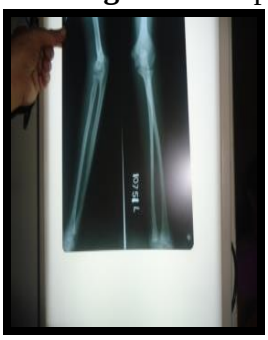
Urinary Bladder: well distended, no calculi\ mass\ diverticulino thickening of bladder mucosa

Liver, gall bladder, pancreas and spleen: are normal. No ascites or abdominal lymph adenopathy. No mass per abdomen.

Pelvis: Evidence of small uterus noted ,endometrial echoes not appreciable, uterus measures 27+9.4 + 8.4mm AP view.

Impression: Small size uterus, no obvious evidence of ovaries. No testes noted, possibilty of pseudohermoprditism and other organs are normal. Both testis are not seen in the inguinal canal or abdomen

Bone Age: Corresponds to real age



Left wrist shows-LW X-ray,

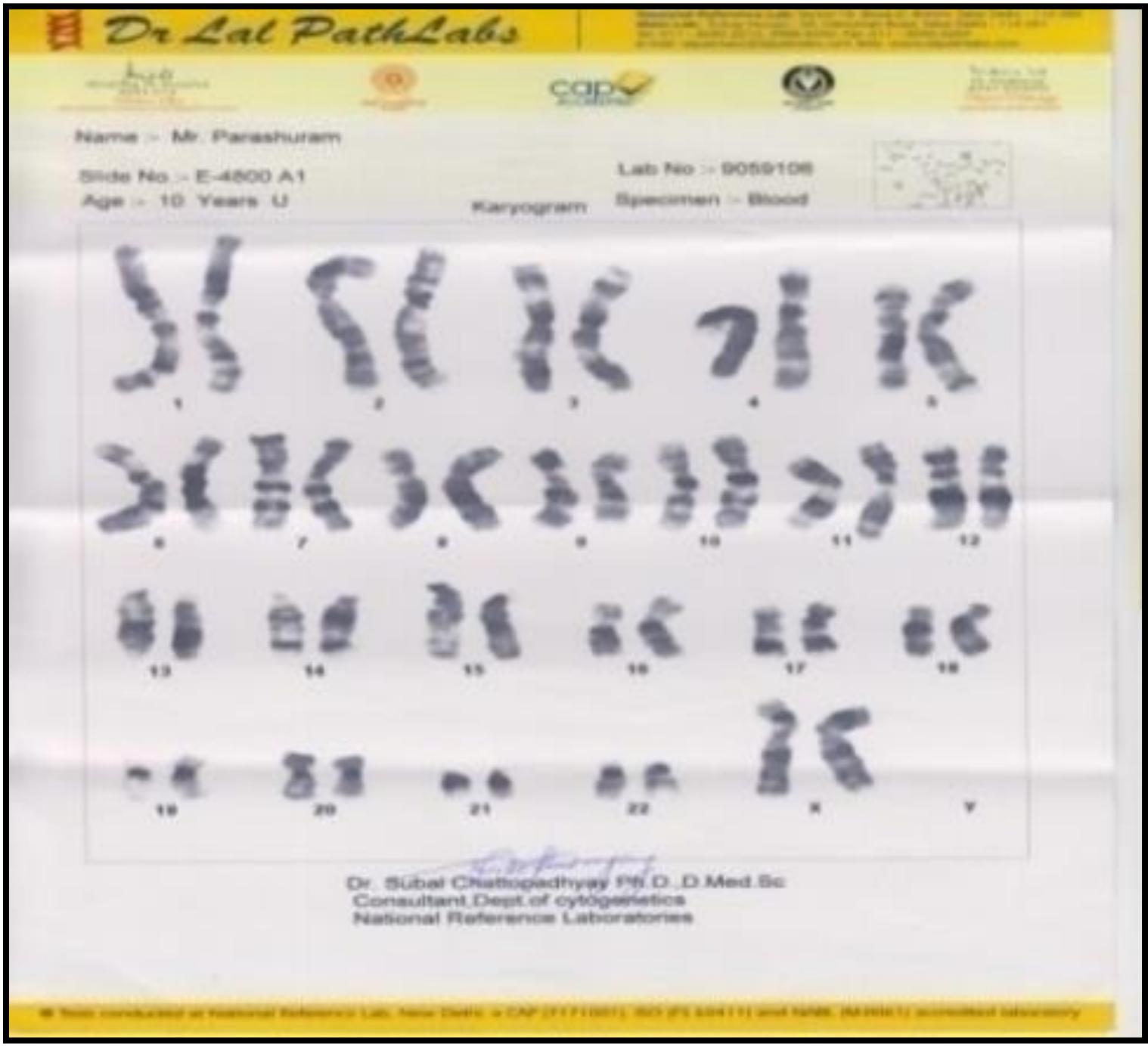
Left- Elbow shows AP X-ray

Serum cortisol (ACTH)level ; Morning -8am-CLIA—2.92micro g\dL
Eevening -9pm- CLIA—8.59micro g\dL
HIV 1 & II : NEGATIVE24. –Serum Sodium –137mEq/L

Serum Sodium – 4.09mEq/L
Serum Sodium – 103 mEq/L
Serum Sodium –137mEq/L

Karyotype: Done under the supervision of expert, - Blood sent to the National laboratory for analysis. 72hrs stimulated cultures with appropriate serum and antibiotics (Medium RPMI-1640) 450- 550 banding resolution GTG banding technique.

Metaphases-numbers---Counted---30
Analysed---30
Karyotyped--02
Photographed –02



Female chromosomal complement with no numerical or structural anomalies detected at the banding resolusion.

Note—1. Microdeletions and cryptic chromosomes deletions may not be detected with cytogenetic analysis.

INTERPRETATION-

Female pseudohermaproditism with 46xx karyotype NO "Y" CHROMOSOME

DISCUSSION

Chromosomal sex determines gonadal sex, which determines phenotypic sex. The type of gonad present determines the differentiation/regression of the internal ducts (ie, müllerian and wolffian ducts) and ultimately determines the phenotypic sex. Gender identity is determined not only by the phenotypic appearance of the individual but also by the brain's prenatal and postnatal development as influenced by the environment (5)

Gonadal differentiation

During the 2nd month of fetal life, the indifferent gonad is guided to develop into a testis by genetic information present on the short arm of the Y chromosome. Testis determining factor (TDF) is a 35–kilobase pair (kbp) sequence on the 11.3 sub band of the Y chromosome, an area termed the sex-determining region of the Y chromosome (SRY). When this region is absent or altered, the in different gonad develops into an ovary.

In my case study, genetic sex determined by the karyotype studies shows Karyotype—46xx

Female chromosomal complement with no numerical or structural anomalies detected at the banding resolusion. USG & 2 D real time abdominal sonography study shows-

Both testis are not seen in the inguinal canal or abdomen.

Impression: Small size uterus, no obvious evidence of ovaries. no testes noted, possibilty of pseudohermoprditism and other organs are normal

INTERPRETATION – Female pseudo hermoproditism with 46xx karyotype NO "Y" CHROMOSOME". absence of y chromosome along with postnatally complete absence of gonads, complete absence of both testis with development of feminine characters like breast development, since 4months regular menstruation. Etc… Correlates with my study clinically

The existence of patients with 46,XX testicular DSD, who have testicular tissue in the absence of an obvious Y chromosome or SRY genetic material requires other genetic explanations for testicular development include *DAX1* on the X chromosome, *SF1* on band 9q33, *WT1* on band 11p3, *SOX9* on bands 17q24-q25, and *AMH* on band 19q13.3. Fetal ovaries develop when the *TDF* gene (or genes) is absent 5.

Development of the internal ducts results from a paracrine effect from the ipsilateral gonad.Jost's classic research with rabbits greatly clarified the gonad's role in controlling subsequent development of internal sex ducts and external genital phenotype **(**3)

When testicular tissue is absent, the fetus morphologically begins and completes the internal sex duct development and external phenotypic development of a female.

For development of male internal sex ducts and an external male phenotype, namely, testosterone and Mülleria inhibiting substance (MIS) is produced by the Sertoli cells of the testis beginning in the eighth fetal week. The prime role of MIS is to repress passive development of the müllerian ducts (eg, fallopian tubes, uterus, upper vagina). In a male fetus with normal testicular function, MIS represses müllerian duct development, while testosterone stimulates wolffian duct development or AMH(5)

Wolffian structures located closest to the source of testosterone undergo the greatest degree of male differentiation. No wolffian development is expected in association with a streak gonad or a non–testosterone-producing dysgenetic testis. 5

Local testosterone production appears to enhance the inhibition of müllerian duct development produced by MIS, while estrogens may interfere with MIS action, resulting in a degree of müllerian duct development. This suggests that müllerian development may be more complex and the research helps explain the variable internal sex

duct anatomy that occurs in some of the complex intersex states**5**.

Differentiation of external genitalia

The external genitalia of both sexes are identical during the first 7 weeks of gestation. Without the hormonal action of the androgens, testosterone and dihydrotestosterone (DHT), helps in translation and transcription of genetic material ie external genitalia appear phenotypically female. In the gonadal male, differentiation toward the male phenotype actively occurs over the next 8 weeks is moderated by testosterone, which is converted to 5-DHT by the action of an enzyme, 5-alpha reductase, present in the cytoplasm of cells of the external genitalia and the urogenital sinus lead to normal male external genital development from primordial parts, forming the scrotum from the genital swellings, forming the shaft of the penis from the folds, and forming the glans penis from the tubercle.5

Incomplete masculinization occurs when testosterone fails to convert to DHT or when DHT fails to act on cells of the external genitalia and urogenital sinus. Testosterone-related developmental change begins at approximately 6 weeks of gestation with a testosterone rise in response to a surge of luteinizing hormone (LH). Testosterone levels remain elevated until the 14th week. Most phenotypic differentiation occurs during this period. After the 14th week, fetal testosterone levels settle at a lower level and are maintained more by maternal stimulation through human chorionic gonadotropin (hCG) than by LH. Testosterone's continued action during the latter phases of gestation is responsible for continued growth of the phallus, which is directly responsive to testosterone and to DHT(5)

CAH is the most common cause of ambiguous genitalia in the newborn. Mixed gonadal dysgenesis (MGD) is the second most common cause of DSD. Hypospadias occurs at a rate of 1 case per 300 live male births; in fewer than 1% of patients, hypospadias occurs in combination with undescended testes.

Ambiguous genitalia ; CAH is the most common cause of ambiguous genitalia in the newborn, it is inherited an autosomal recessive disorder .charcteristic by cortical enzymes so low hormonal input but increase in ACTH from the pituitary cause adrenal hyperplasia, leads to hyperplastic adrenals elaberate androgens and cause the masculisation of female fetus leading to female pseudo hermoproditism leading to hypertrophy of labia majora fused. , clitoris. His walking style and voice is changed slow and low pitch changed like girls but his thyroid profile normal.

Female pseudo hermoproditism with 46xx karyotype posess ovaries and derivatives of mullarian ducts .but masculinised by clitoromegaly ,displacement of urethra and labial fusion.results in ambiguous genitalia.Its derivatives,either endogenenously through a hormonal abnormality or HRT.leads to increase in the testosterone in affected females are adrenogenital syndrome .due to multiple inherited enzymes defficiencies 21-hydroxylase and 11b hydroxylase -] abnormalitie can occurs along the path way of cortisol productions due to [AR] produsedduring 8 and 9 week of gestational life.so increase in the testosterone level causes virulised female new born with intact Mullarian structures and ovaries. External source of norethisterone acetate and norethisterone OCP component produce this defect depending on the time of exposure and dose 7.

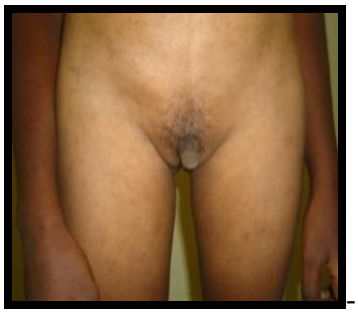
In my study shows---- Ambigious genitalia

Penis short

Length of phallous-3.7mms

Partially fused genital fold Hyper pigmentation and mild rugosity Opening of urethra about 3 cms away from root of the phallous. Hypospadiasis, labia majora fused.

No palpable gonads. Scrotal sac empty. now. Menstruating since last 4-6 months.



In the embryo, the external genital structures are originally the same in males and females. Exposure to testosterone, the male hormone, causes differentiation into a penis and scrotum, promotes formation of the prostate, and other internal male reproductive structures. Lack of testosterone, as in females causes the external genitals to differentiate in a male pattern. Common ones include partial or complete insensitivity of the tissue to testosterone, inadequate production of testosterone, lack of the testes-determining region of the Y chromosome in a male, or its presence in a female, and malformations due to exposure of the fetus in utero to certain drugs. 8

Medical Treatment

Current medical treatment for intersex individuals is in a state of flux. Infants born with obviously ambiguous genitals undergo many tests (**chromosomal, hormonal, and anatomical**) to determine the sex is based on the ability to create cosmetically unambiguous and functional genitals with the tissue present. In cases where future fertility is possible, this too is considered.

Medical aspects: Infants born with ambiguous genitalia represent a true medical and social emergency.

Salt-wasting nephropathy occurs in 75% of infants born with CAH, the most common cause of ambiguous genitalia. If unrecognized, the resulting hypotension can cause vascular collapse and death. Male infants with this syndrome may be phenotypically normal, and the diagnosis may be missed.

Surgical correction Surgery is performed before the age of 18 months to make the genitals match, the assigned sex.

- Testes \ ovo testis are absent in my patient no question of orchidectomy and malignancy. External genitalia as to be corrected to make female type and supported by oestrogen therapy to help the feminization in puberty. 4
- Scrotal cleft corrected by surgical interference with or without Hysterectomy, Tubectomy. Etc. it helps in masculization at puberty with parental testosterone.
- Vagina is too short and non capacious may requires plastic surgery- Clitarorraphy.

After surgery, 21% incidence of diverticula formation, as well as strictures and fistulas in some patients. Short-term follow-up necessary . **Average patient age in the hypospadia study was two months.** Obviously, sexual function was not one of the "functional outcomes" considered.8

Medico legal aspects of intersex

Depending upon the anatomical features of external genitalia rather on the gonads or chromosomal pattern , they rearedup as male or female.

Female pseudo hermaphrodites; reared up as females, have normal internal sex organs. Capable of developing into normal fertile woman. He having large phallus or libido scrotal fusion may require surgical correction .

In my study reared up as male, Child sits and passes urine but he is not passed from the pallus but below it from the separate opening penis short-Length of phallous-3.7mms

Partially fused genital fold Hyper pigmentation and mild rugosity Opening of urethra about 3 cms away from root of the phallous. Hypospadiasis, labia majora fused.No palpable gonads. Scrotal sacs empty.

He needs surgical correction [to create cosmetically unambiguous].

CONCLUSIONS

This have been not only a focus of attention of anatomists but also at interest of Genetitians, Embryologists, oncologists, paediatric endocrinologists, surgeons, physicians, psychiatrists, Forensic experts because it makes very sensivitive issue on the boy, family and society.

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