

# DSD

## Controversies IN DISORDERS OF SEXUAL DIFFERENTIATION

Dr E M Moshokoa

# SOCIAL IMPACT

► Intersex people and religion

Rights of inheritance

Schooling

Rights to marriage

Rights to live like any other male or female.

## ► 19th-century : RIGHT TO VOTE

« Levi Suydam was an intersex person in Connecticut whose *capacity to vote in male-only elections was questioned in 1843.* »

## ► 16th-century - laws of succession:

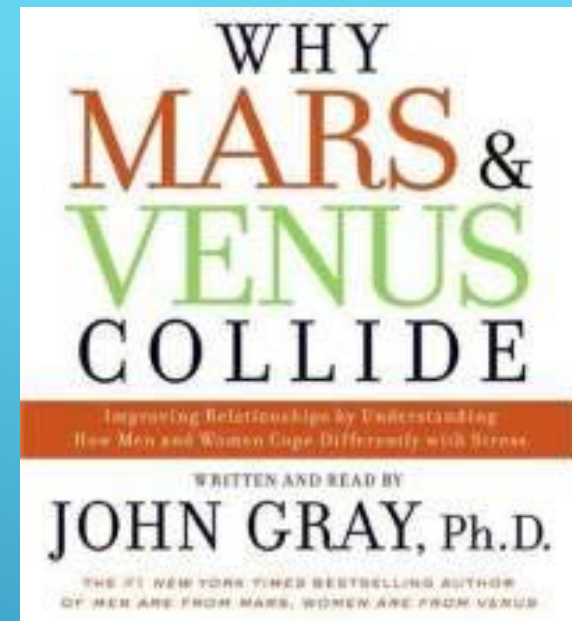
"*Every heir is* either a male, a female, *or an hermaphrodite*, that is both male and female., according to that kind of sex which doth prevail. »

English jurist and judge Edward Coke (Lord Coke)

3



- ▶ Gender assignment
- ▶ Gender identity issues,
- ▶ Management
  - ▶ Timing of Surgeries  
Parental vs patient consent)
  - ▶ Sensation and sexual function
  - ▶ Decision-making on cancer and other risks



## CONTROVERSIES

- ▶ Gender assignment
- ▶ Gender identity issues,
- ▶ Management
- ▶ Timing of Surgeries
- ▶ Parental vs patient consent)
- ▶ Sensation and sexual function
- ▶ Decision-making on cancer and other physical risks

# SEX ASSIGNMENT IS THE DETERMINATION OF AN INFANT'S SEX AT BIRTH.

- ▶ Assigned female at birth (AFAB) : "It's a girl! »

60% diagnosed prenatally, many parents are faced with the situation at birth 1 in 5,500

- Geographical variation : ovotesticular DSD in South Africa
- 5 $\alpha$ reductase deficiency in the Dominican Republic

- Assigned male at birth (AMAB): "It's a boy!"

- ▶ SEX MAY BE DETERMINED BEFORE BIRTH.
- ▶ IMPLICATIONS !

THIS DAYS ! BABY SHOWERS



Female (XX): clitoral enlargement  
(21-hydroxylase deficiency)



# Sex Has Many Levels

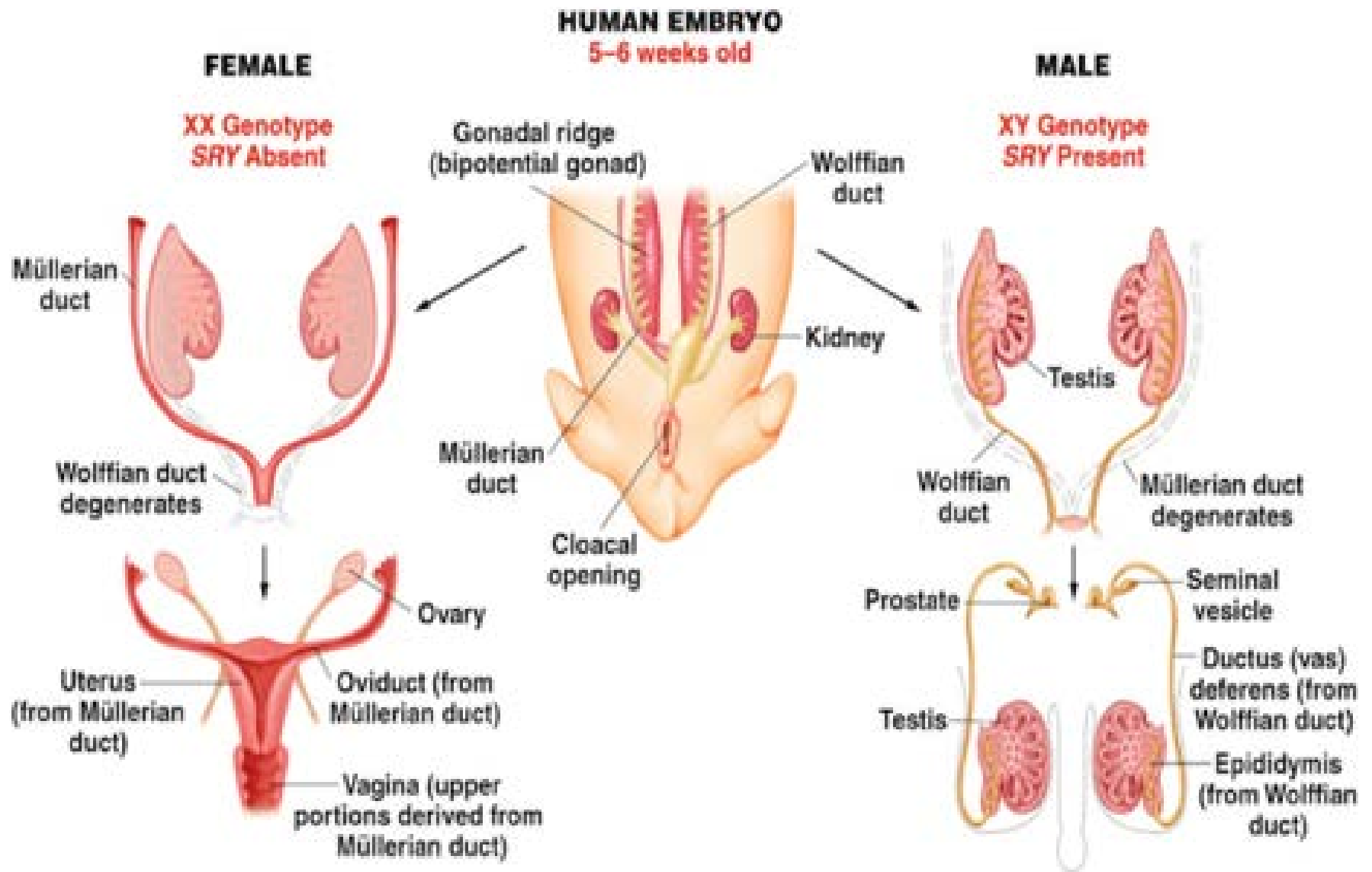
## Primary sex characteristic



- Genetic Sex – Chromosomes (X & Y in mammals only)
- **Gonadal Sex – Ovaries/Testis**
- Hormonal Sex – Estrogen/Testosterone
- Somatic Sex – Body anatomy/physiology
- Psychological Sex - Sexual identity

and then there is

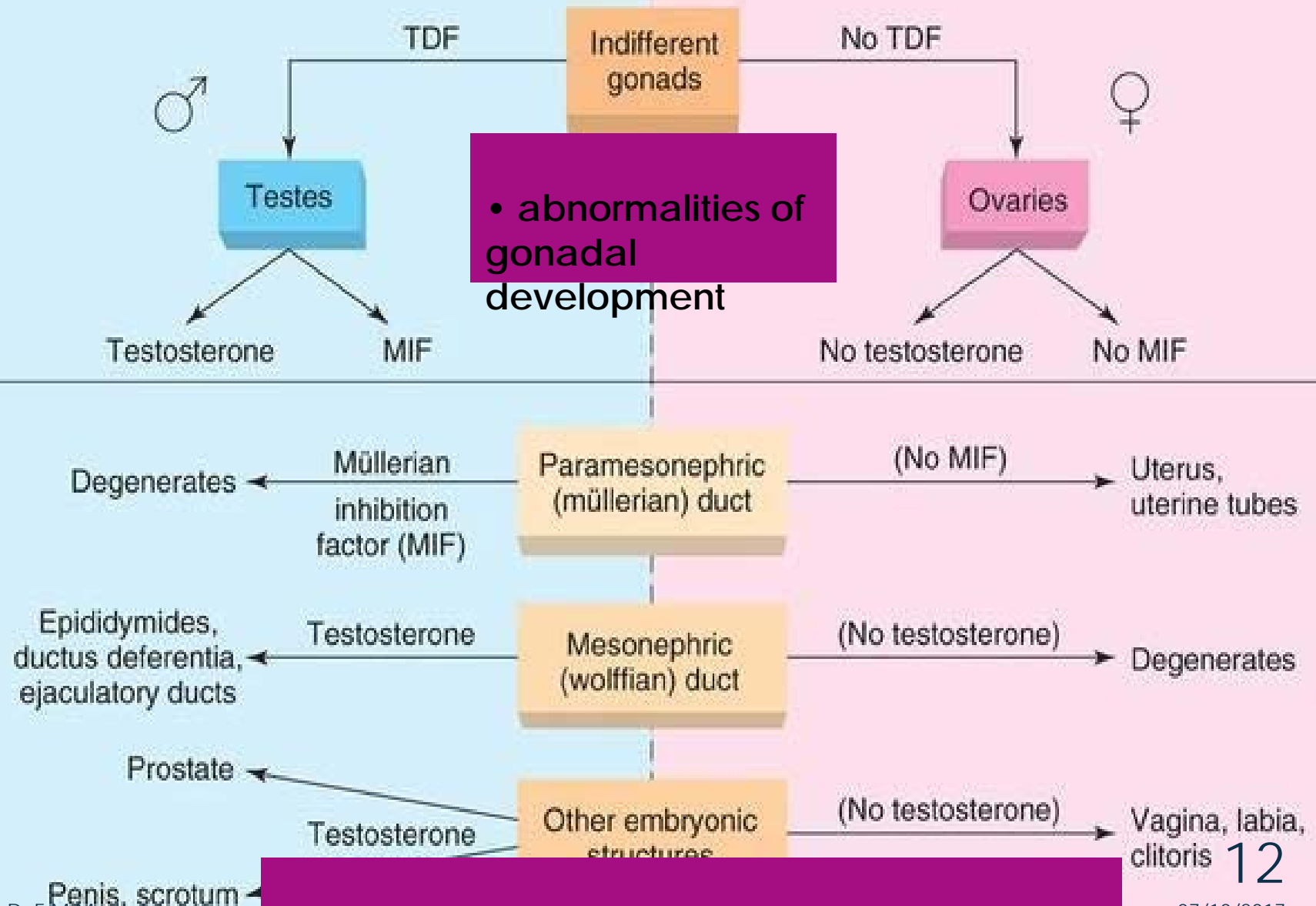
- Sexual orientation



## NORMAL PATHWAY OF SEX DIFFERENTIATION

11

## chromosomal defects



• abnormalities of gonadal development

• defects in the synthesis of sex hormones or relevant receptor



# TABLE 2

DSD classification proposed by the Chicago consensus

Sex chromosome DSD	46,XY DSD	46,XX DSD
45,X (Turner syndrome and variants)	Disorders of gonadal (testicular) development	Disorders of gonadal (ovary) development
47,XXY (Klinefelter syndrome and variants)	Complete gonadal dysgenesis (Swyer syndrome)	Ovotesticular DSD
45,X/46,XY (mixed gonadal dysgenesis, ovotesticular DSD)	Partial gonadal dysgenesis	Testicular DSD (SRY+)
46,XX/46,XY (chimeric, ovotesticular DSD)	Gonadal regression Ovotesticular DSD	Gonadal dysgenesis
	Disorders in androgen synthesis or action	Androgen excess
	Androgen biosynthesis defect (17-hydroxysteroid dehydrogenase deficiency, 5 $\alpha$ -reductase deficiency)	Fetal (21- or 11-hydroxy)
	Defect in androgen action (CAIS, PAIS)	Fetoplacental (aromatase deficiency)
	LH receptor defects (Leydig cell hypoplasia)	Maternal (luteoma, exogenous androgens)
	Disorders of AMH and AMH receptor (persistent müllerian duct syndrome)	
	Other (severe hypospadias, cloacal extrophy)	Other (cloacal extrophy, M

# CLINICAL CASE

## Dehydration

Collapse

HYPERPIGMENTATION  
SCROTUM

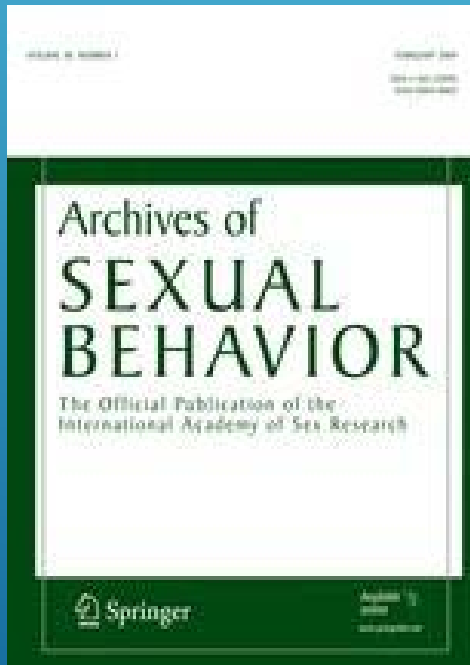
VIRILIZATION  
/CLITORIS

HYPOTHESIS ??

CAH

**Not only a DSD but a LIFE THREATENING  
CONDITION !!!!!**

# RESULTS SUGGEST THAT ADULTS WITH DSD ARE MARKEDLY PSYCHOLOGICALLY DISTRESSED ;



with **rates of suicidal tendencies and self-harming behavior on a level comparable to** non-DSD women with a history of physical or **sexual abuse**



MS L L

- 15 year old female
- 46 XX
  - Diagnosed at 3 months
  - Raised as a girl
  - Menarche at 13 with regular menses
  - Secondary sexual characteristics
    - Tanner 3 Breast
    - Tanner 2 Pubic hair

## CLINICAL CASE

### *PRESENTATION AT PUBERTY*

- Main complaint of clitoromegaly.
  - 6cm clitorus
- Clitoral reduction performed September 2016.
- Cystoscopy and vaginoscopy
  - Small vaginal introitus
  - Normal bladder and vagina





# WHEN IS DSD SUSPECTED? IN NEONATAL AGE/ CHILDHOOD ADOLESCENT/ ADULT

- ▶ Ambiguous genitalia – on external examination there is a doubt.
- ▶ Hypospadias with undescended testis
- ▶ Severe Hypospadias
- ▶ Micropenis
- ▶ Clitoral hypertrophy/ posterior labial fusion
- ▶ Inguinal hernia in a girl child

Virilization in a girl

Delayed or incomplete puberty

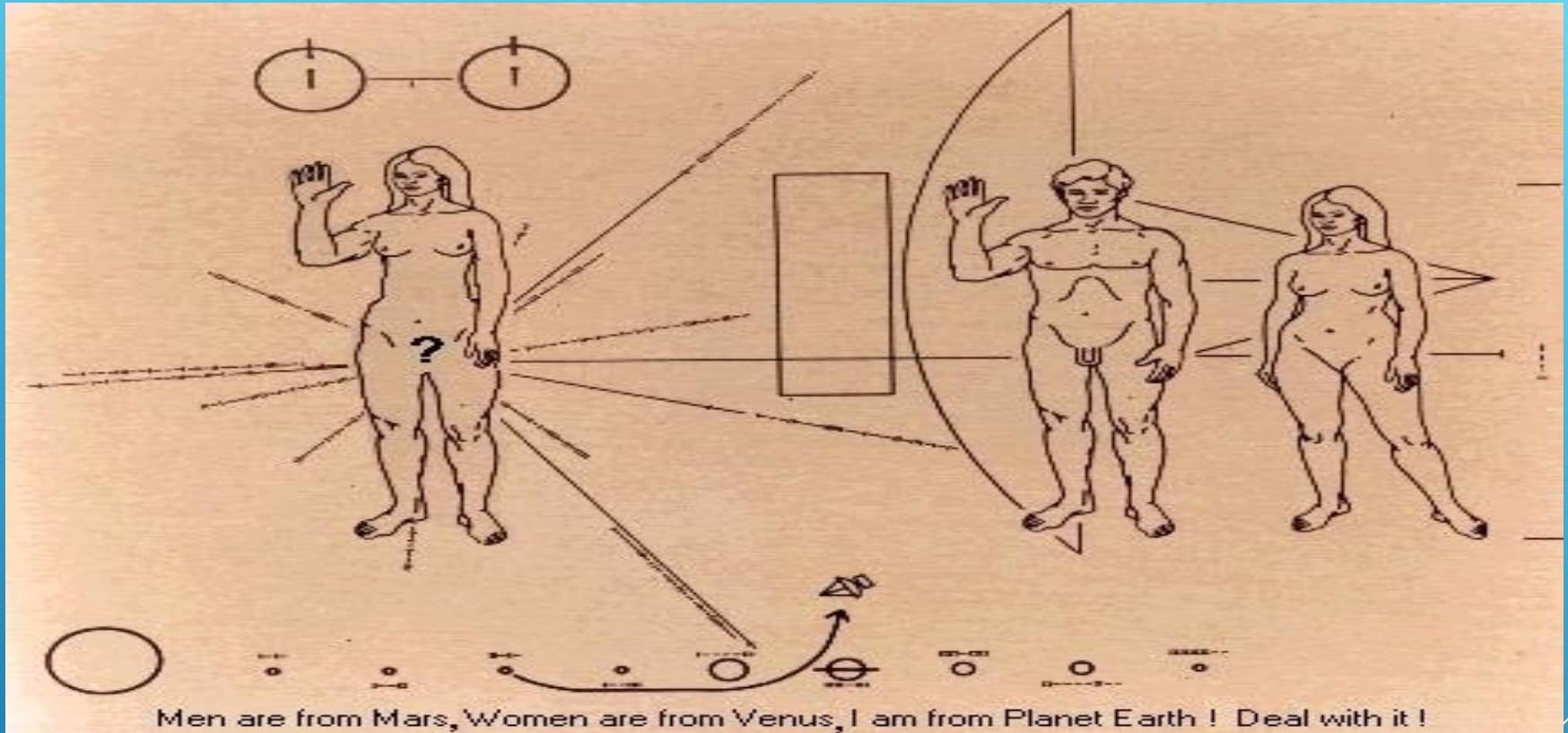
Primary amenorrhea

Infertility

Breast development in a male

Cyclical hematuria in a male

- ▶ Gender assignment
- ▶ Gender identity - gender role issues,
- ▶ Management
- ▶ Timing of Surgeries
- ▶ Parental vs patient consent)
- ▶ Sensation and sexual function
- ▶ Decision-making on cancer and other physical risks



# ME , MYSELF AND I

- ▶ Intersex people are born with sex characteristics, such as chromosomes, gonads, or genitals that,

*"do not fit typical binary notions of male or female bodies".*

« UN Office of the High Commissioner for Human Rights »

HUMAN RIGHTS

# PSYCHOLOGY

Mum why I'm I different from other children ???!!!

**I was supposed to have a couple of surgery I had twenty ....!!!**

**Sometimes I'm willing to play Jack with boys and sometimes jump with girls**

**Tiger Devore, clinical psychologist .**

Seing a pregnant women or a baby make me feel sad ....

#### **4. MAKE GENDER SEGREGATED SPACES INCLUSIVE**

Do you know how dangerous restrooms or changing rooms can be for trans or gender non-conforming youth?

**I CAN'T EVEN GO PEE  
WITHOUT EVERYONE MAKING  
A FUSS ABOUT IT!**

Trans and gender non-conforming students need access to their preferred restroom or changing room.



**It is not a caprice!**

Violence and aggressions are more likely to happen there than anywhere else, and those students are often easy targets for bullies.

- ▶ Gender assignment
- ▶ Gender identity issues,
- ▶ Management
- ▶ Timing of Surgeries
- ▶ Parental vs patient consent)
- ▶ Sensation and sexual function
- ▶ Decision-making on cancer and other physical risks



# MANAGEMENT

**1. GENDER ASSIGNMENT** ( diagnosis, genital appearance, fertility potential, therapeutic/surgical options, and familial views or circumstances relating to cultural biases )

**Alex mac farlane Australian passport with an 'X' sex marker**

**2. HORMONE-REPLACEMENT THERAPY** (to induce and sustain puberty, induce secondary sexual characteristics and pubertal growth spurt, optimize bone mineral accumulation, and for psychosocial maturation in patients with DSD )

**3. PSYCHOSOCIAL SUPPORT**

**4. SURGICAL MANAGEMENT** -There is no consensus regarding indications, timing, procedure and evaluation of outcome of DSD surgery.)

**In 2011, Christiane Völling won the first successful case brought against a surgeon for non-consensual surgical intervention.**



## LABORATORY

- ▶ 17-OH progesterone
- ▶ Karyotype – is the study of chromosomes, done via blood tests
- ▶ Hormonal assay- Testosterone/ Dihydrotestosterone/ LH/FSH/Estradiol etc

## IMAGING



## INVESTIGATIONS

# IMAGING

- ▶ Ultrasound ( internal organ,uterus, mullerian remnant )
- ▶ **Retrograde genitogram** ( before surgery , junction between urethra, vagina, mullerian structure )
- ▶ MRI pelvis
- ▶ (cystourethroscopy and vaginoscopy)
- ▶ • laparoscopy to identify internal structures
- ▶ (uterus, tubes, gonads)



Contrast sinogram (genitogram). (a) Injection of contrast through a catheter positioned too low in the first picture . Visualization of mullerian remnants with high communication with urethra

- ▶ Gender assignment
- ▶ Gender identity issues,
- ▶ Management
- ▶ Timing of Surgeries
- ▶ Parental vs patient consent)
- ▶ Sensation and sexual function
- ▶ Decision-making on cancer and other physical risks

In a study in Lübeck conducted between 2005 and 2007 ... 81% of 439 individuals had been subjected to surgeries due to their intersex diagnoses.

- ▶ Two thirds of the adult participants drew a **connection between sexual problems and their history of surgical treatment.**

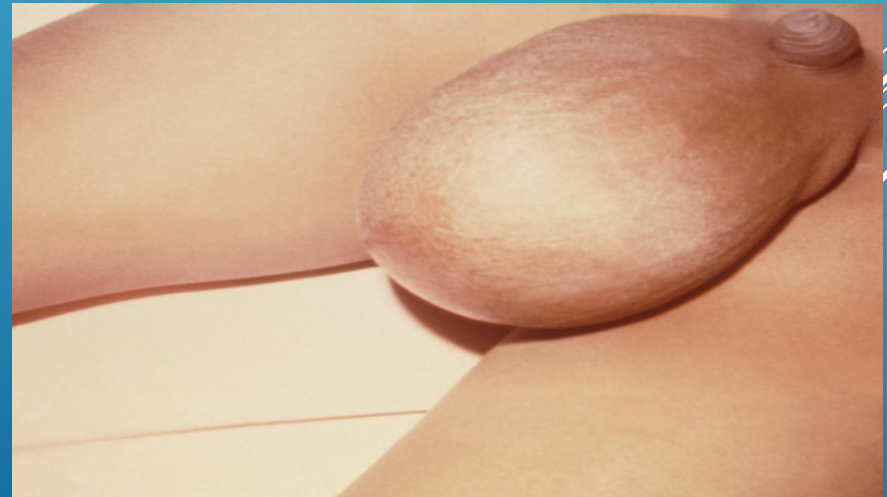
Almost 50% of participants reported psychological problems.

- ▶ The determinants of gender-related behaviour **DSD AND SEXUALITY**
- ▶ : biological factors include neuroendocrine factors
- ▶ complex sociocultural influences are less well understood

# SEXUALITY OF CAH WOMEN

- ▶ More male typical behavior at children
- ▶ Less heterosexual interest ( **J. of SEXRES** )
- ▶ Potentiel to develop bisexual or homosexual orientation
- ▶ Excess to the prenatal androgen predispose some women to same gender sexual orientation ( **BEHAVIOR GENETICS** )

- ▶ Gender assignment
- ▶ Gender identity issues,
- ▶ Management
- ▶ Timing of Surgeries
- ▶ (Parental vs patient consent)
- ▶ Sensation and sexual function
- ▶ Decision-making on cancer and other physical risks



- Timing: at **diagnosis** versus puberty versus no gonadectomy
- Healthy, functional gonadal tissue should remain in place unless the patient requests it to be removed
- Risk of gonadoblastomas:
  - CAIS: very low
  - PAIS: 30-50% if intra-abdominal testes
- Risk of **virilization** during puberty
- Consider cryo-preserving gonadal tissue for later fertility

GONADECTOMY  
CLITORIS REDUCTION  
HYPOSPADIAS REPAIR



# ACTIVISM

"all necessary legislative, administrative and other measures to ensure that **no child's body is irreversibly altered by medical procedures** in an attempt to impose a gender identity without the full, free and informed consent of the child".

## *Principle 18 on Protection from Medical Abuse*

■ Thank You

