

INTERSEXUALITY

ABNORMAL SEXUAL DEVELOPMENT

1-Sex chromosome abnormality

Mosaicism associated with gonadal dysgenesis \Rightarrow
45X/46XY

2-Testis incapable of producing testosterone

3-End organs incapable of utilizing testosterone eg. 5 α reductase deficiency, failure of testosterone binding to receptors (androgen insensitivity)

ABNORMAL SEXUAL DEVELOPMENT

4-Deficient production of MIF \Rightarrow ♀ internal genital organs
in otherwise normal ♂

5-Muscularization of the ♀ external genitalia due to $\square \uparrow$
androgen eg. Congenital adrenal hyperplasia

6-Rarely 46XX male due to the presence of a gene the
SRY gene (Sex determining Region Y)

7-True hermaphroditism \Rightarrow the presence of testicular &
gonadal tissue in the same individual

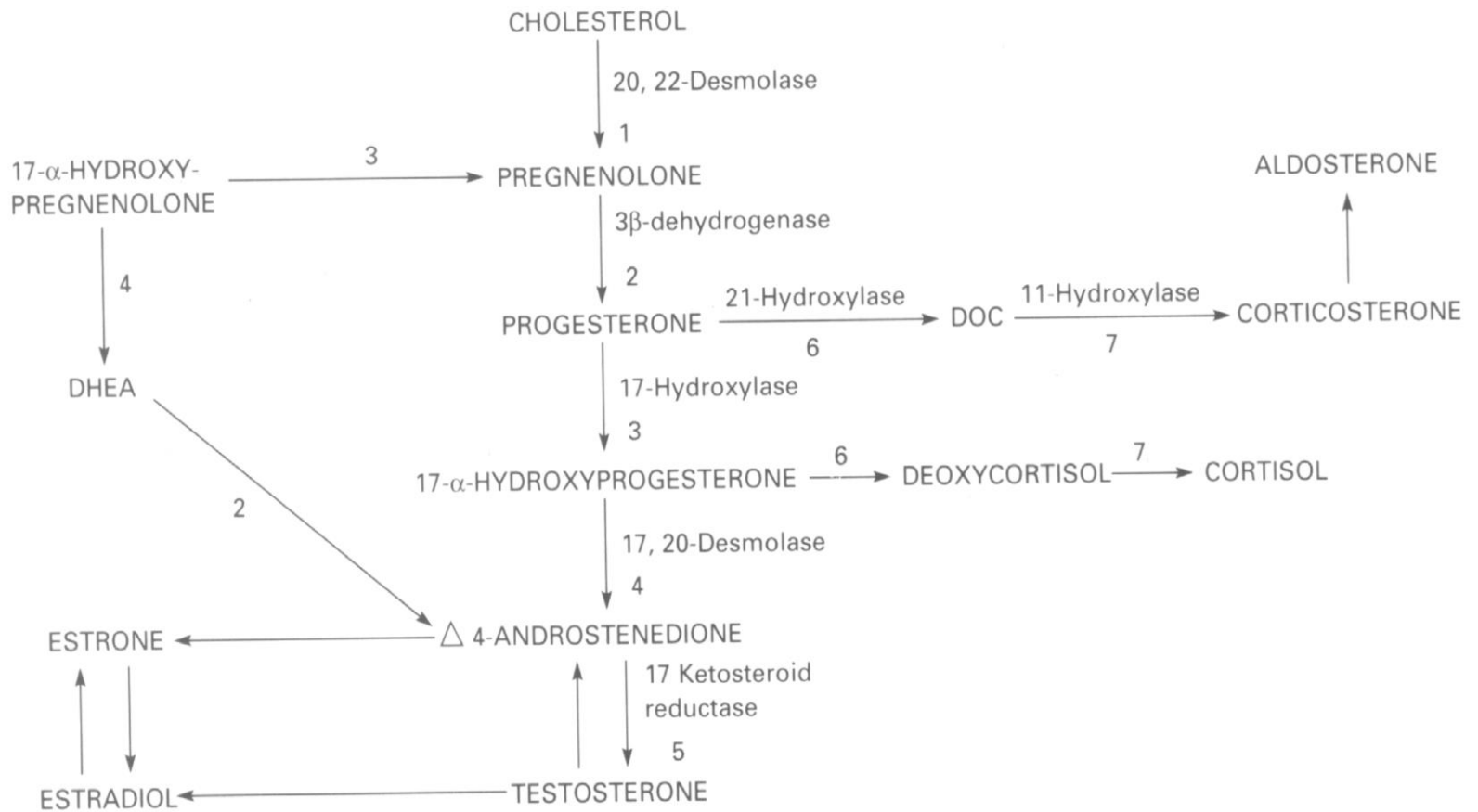
1-MUSCULINIZED ♀ ♀ PSEUDOHERMAPHRODITES

-46XX

-Exposed to androgens in utero ⇒ varying degrees of masculinization of the external genitalia

A-CONGENITAL ADRENAL HYPER PLASIA (CAH)

- The most common cause of ♀ intersex
- Deficiencies of the various enzymes required for cortisol & aldosterone biosynthesis (21-hydroxylase, 11β-hydroxylase, 3βhydroxysteroid dehydrogenase)
- 21-hydroxylase deficiency is the commonest defect 90%
- Affected ♀ may present at birth with ambiguous genitalia
 - enlargement of the clitoris
 - excessive fusion of the genital folds obscuring the vagina & urethra



Congenital Adrenal Hyperplasia



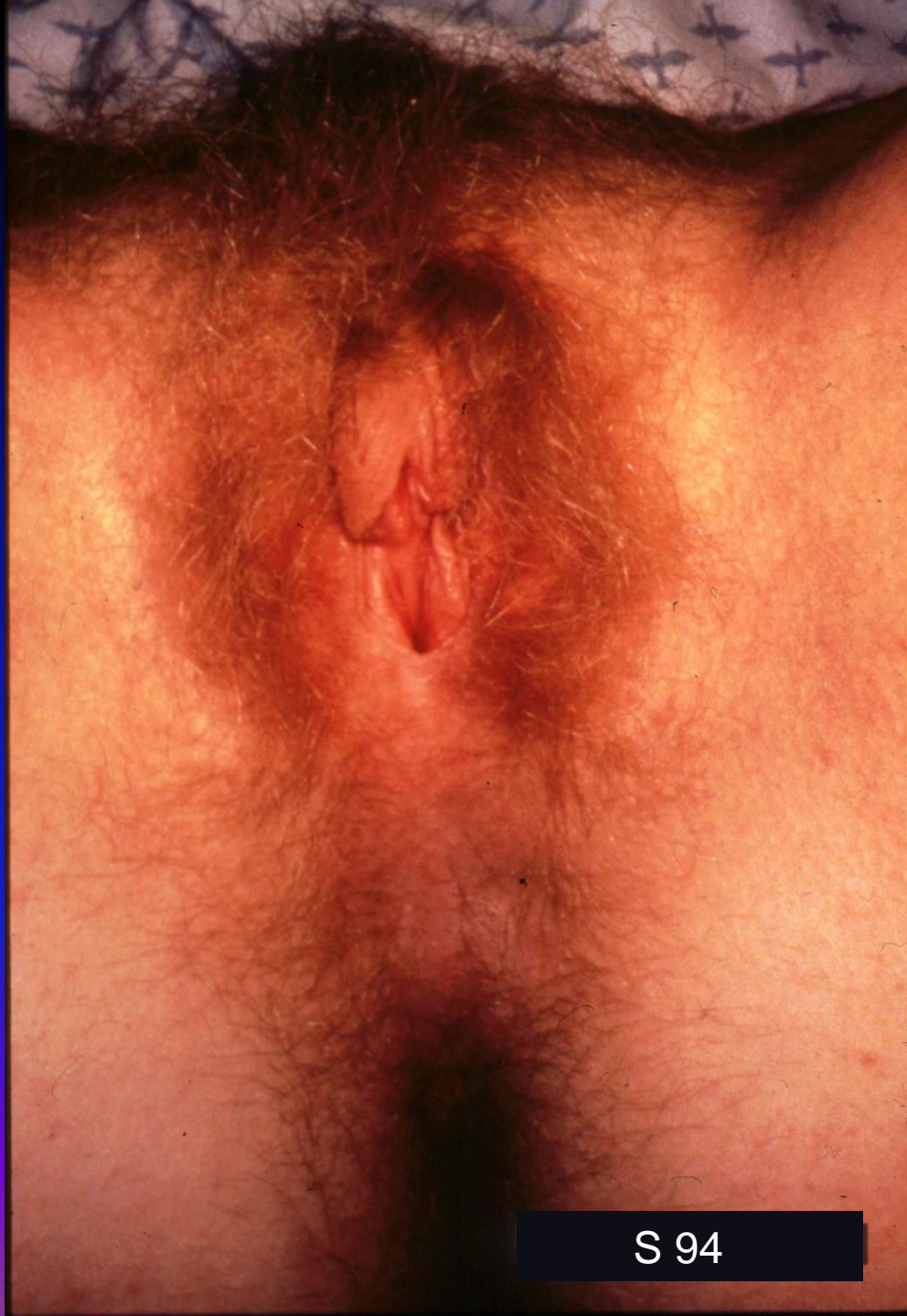
A-CONGENITAL ADRENAL HYPER PLASIA (CAH)

-thickening & rugosity of the labia majora resembling the scrotum

- A dangerous salt losing syndrome due to deficiency of aldosterone may occur in some pt
- Delayed menarche & menstrual irregularities
- INVESTIGATIONS
 - Karyotyping
 - 17- α -hydroxiprogesterone \uparrow
 - 17-ketosteroids (androgens) in urine
 - Electrolytes
 - U/S



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A-CONGENITAL ADRENAL HYPER PLASIA (CAH)

- Rx

1- Cortisol or its synthetic derivatives \Rightarrow suppress the adrenals \Rightarrow \downarrow androgen production

2-Corrective surgery

clitroplasty (neonatal period)

division of the fused labial folds

(delayed till puberty)

MUSCULINIZED ♀

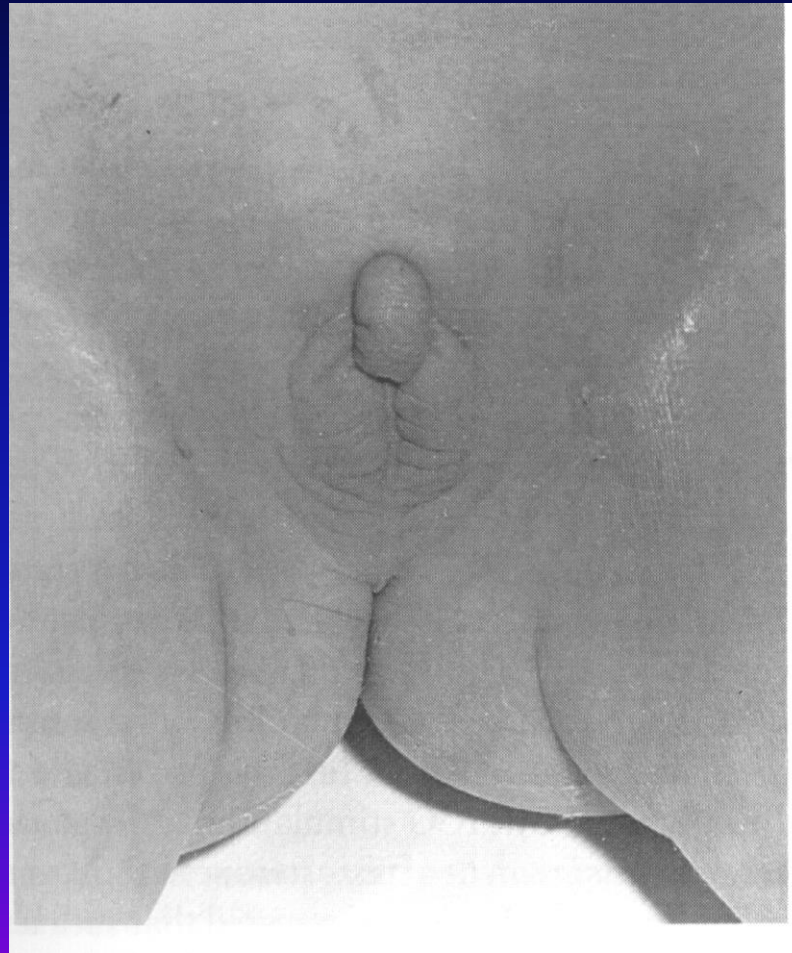
B- EXPOSURE OF THE MOTHER TO ANDROGENS

- Rare

- Androgen secreting tumours , eg. luteoma,
arrhenoblastoma

- Drugs

Muscularization of female child
Mother exposed to methyle testosterone



2-UNDERMUSCULINIZED ♂ ♂ PSEUDOHERMAPHRODITES

A-ANATOMICAL TESTICULAR FAILURE

-Pure gonadal dysgenesis

*normal chromosomes 46XY

*variable features – mild-severe

(normal ♀ , ♀ with mild masculinization)

*uterus present

-Mosaicism 45X/46XY

*Variable features

(normal ♀, ambiguous genitalia, nearly normal ♂)

♂ PSEUDOHERMAPHRODITES

B-ENZYMATIC TESTICULAR FAILURE

Enzymetic defects in the biosynthesis of testosterone

These defects are usually incomplete ⇒

Varying degrees of masculinization of the external genitalia

Uterus & tubes ⇒ absent (MIF produced by the testes)

♂ PSEUDOHERMAPHRODITES

C-ENDORGAN INSINSITIVITY

1-5 α REDUCTASE DEFICIENCY

Autosomal recessive

Formation of the ♂ external genitalia requiers

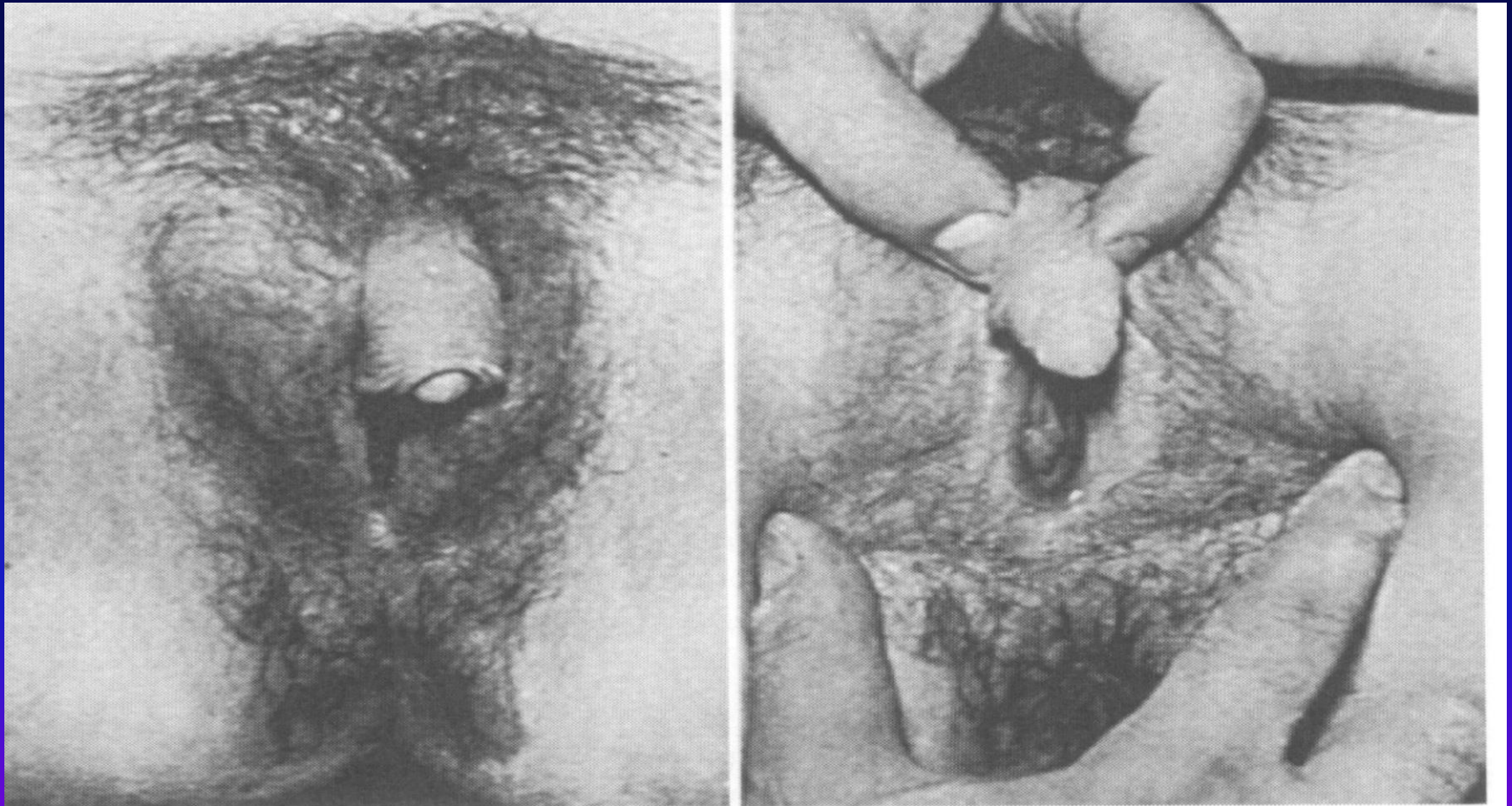
5 α REDUCTASE

testosterone \rightleftharpoons \rightleftharpoons \rightleftharpoons \rightleftharpoons dihydrotestosterone

Formation of the internal woffiane structures respond directly to testosterone

- External genitalia ♀ with mild masculinization
- Absent uterus
- At puberty \rightleftharpoons \uparrow testosterone secretion \rightleftharpoons virilization

5-alpha reductase deficiency





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C-ENDORGAN INSINSITIVITY

2-ANDROGEN INSINSITIVITY (TESTICULAR FEMINIZATION)

Etiology

- Lack of androgen receptors \Rightarrow complete (classical TF)
- Receptors are present but low in NO. or inactive
 \Rightarrow incomplete androgen insinsitivity

Clinical features of Complete Androgen Insinsitivity

Normal ♀ external genitalia with blind vagina

Absent uterus

Breast development

Present with 1ry amenorrhea

Testes found in abdomen or inguinal canal

Normal ♂ Testosterone level

2-ANDROGEN INSINSITIVITY

Rx

Gonadectomy after puberty due to ↑ incidence of malignant change (5%)

Oestrogen replacement

INCOMPLETE ANDROGEN INSINSITIVITY

Ambiguous genitalia with varying degrees

Breast development

Musculinization at puberty



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3-TRUE HERMAPHRODITES

HAVE BOTH OVARIAN & TESTICULAR TISSUE

Ovotestes on one side & ovary or testes on the other

Ovary on one side & testes on the other

Bilateral ovotestes

Varying degrees of sexual ambiguity

KARYOTYPING

46XX \Rightarrow most common

46XX/XY

46XY

46XY/47XXY

TRUE HERMAPHRODITE



Klinefelter Syndrome

47XXY

Normal male external genitalia

Tall stature

Gynecomastia

Azospermia (infertility)