DISEASES OF THE GONADS

Dr. Peter Igaz PhD

2nd Department of Medicine
Semmelweis University

ns of androgen deficiency 1.

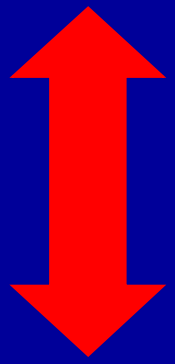
Organ	before puberty	after puberty
Bones	eunuchoid appearance	osteoporosis
Hair growth	lack of beard growth, forehead hair line, pubic hair line	diminishing sexual hair growth
Larynx	no voice change	-
Skin	decreased activity of sebaceous glands, no acnes	atrophy wrinkles
Bone marrow	anemia	anemia
Muscles	underdeveloped	atrophy
Penis	infantile	no change in size

ns of androgen deficiency 2.

Organ	before puberty	after puberty
Prostate	underdeveloped	atrophy
Spermatogenesis	not initiated	halted
Libido, Potency	no	loss

ms of hypogonadism

Central, secondary, hypogonadotrop
LH, FSH low



Gonadal, primary,
hypergonadotrop
LH, FSH high

s of central hypogonadism

- Kallmann-Syndrom
  - GnRH deficiency + Anosmia/Hyposmia
- Prader-Willi-Labhart syndrom
  - GnRH deficiency, obesity, low body height, newborn hypotony, diabetes mellitus type 1, strabismus, scoliosis, mental retadation.
- Pituitary insufficiency
- Hyperprolactinemia

y of central hypogonadism

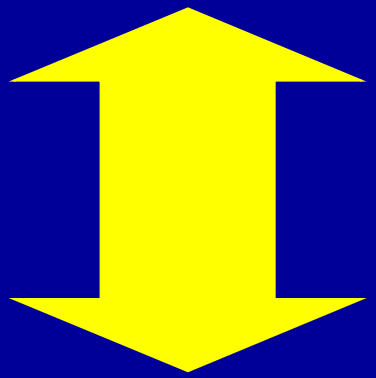
- Androgens
  - Testosteron enanthat, cypionat i.m., transdermal, per os (hepatotoxic)
- LH, FSH, hCG
  - i.v. for fertility
- GnRH, GnRH-agonists
  - pulsatile, pump for fertility

f male gonadal hypogonadism

- 1. Klinefelter syndrome (small testes, no spermatogenesis, gynecomastia, breast cancer).
- 2. del Castillo syndrome, Sertoli cell only syndrome, no spermatogenis
- 3. Noonan Syndrom (male Turner sy.)
- 4. Chemical and biological testis damage

Primary amenorrhoea

puberty



Secondary amenorrhoea

pregnancy

menopause

uses of amenorrhoea

- **Hypothalamus**
  - **Kallmann Syndrom**
  - **Anorexia nervosa**
- **Hypophyse**
  - **Sheehan Syndrom (ischemic necrosis of the pituitary)**
  - **Hyperprolactinemia**
- **Ovaries**
  - **Dysgenesis of the ovaries (Turner syndrome)**
  - **Stein-Leventhal syndrom, PCO**

Turner-syndrome gonadal dysgenesis

Primary amenorrhea
Streak gonads
Pterygium colli
Small stature (133-153 cm)
Cardiovascular Anomalies

Estrogen for secondary sexual characteristics

GH for growth promotion

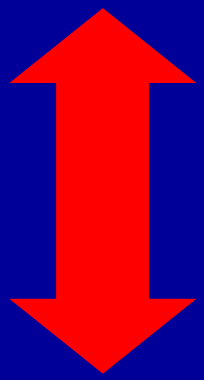
seudohermaphroditism

- Pseudohermaphroditism: chromosomal and gonadal sex is different from phenotpye
- Female pseudohermaphroditism
  - Chromosomal and gonadal sex: woman
  - Phenotype: male
- Male pseudohermaphroditism
  - Chromosomal und gonadal sex: male
  - Phenotype: female

HYPERANDROGENISM

ncreased hair growth

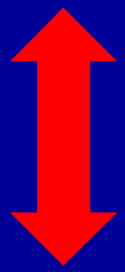
Hirsutism
androgen-dependent regions
e.g.: face, breast, belly



Hypertrichosis
Non-androgen-dependent regions
e.g. exremities

Virilisation

In utero virilisation
Intersexuality ï
female pseudohermaphroditism



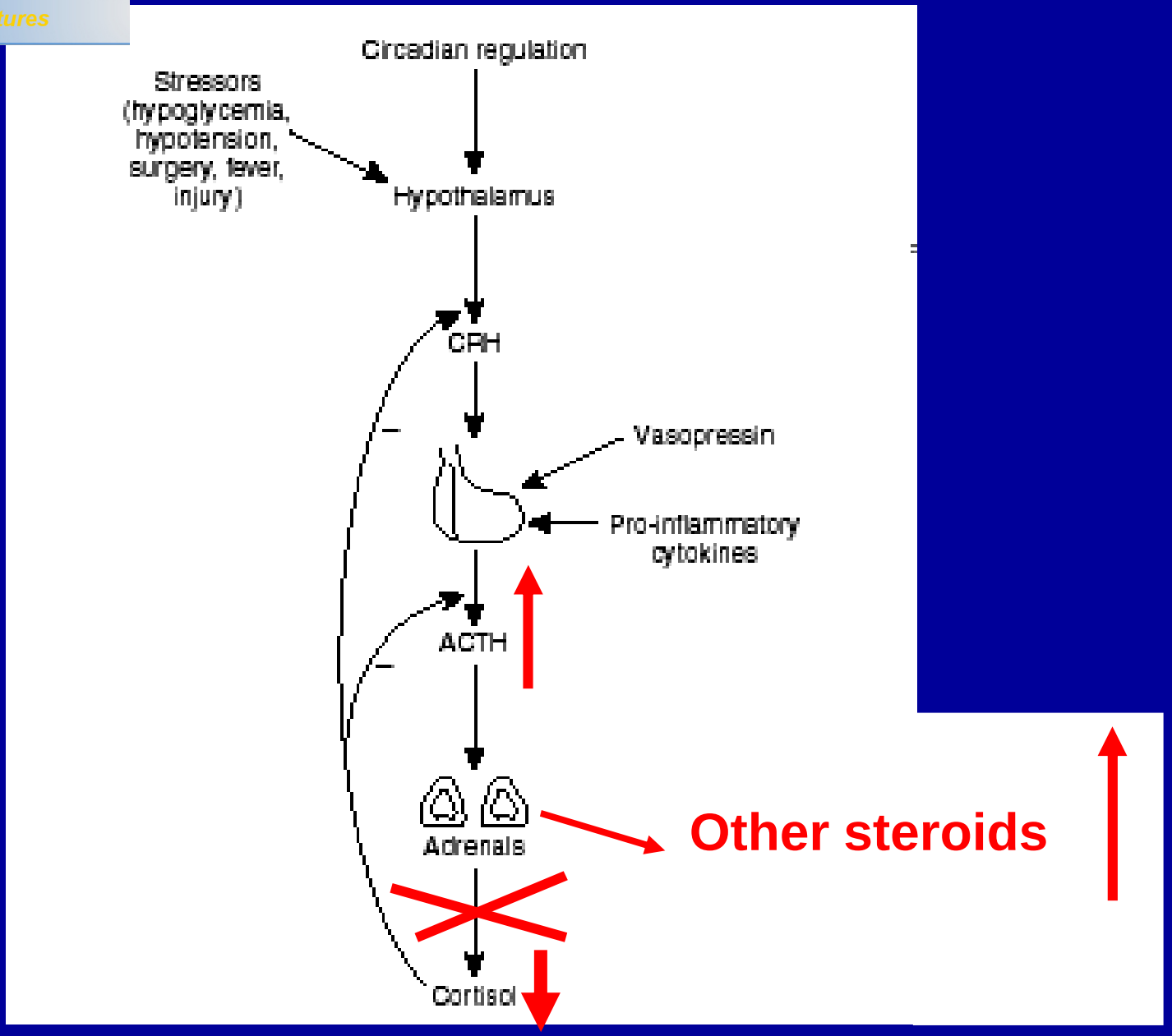
Virilisation in adults

- clitoris hypertrophy
- hirsutism
- male type baldness
- deeper voice
- secondary amennorhoea

Causes of virilisation

- 1. endocrine diseases
  - Adrenal causes
    - androgen-secreting adenoma
    - androgen-secreting adrenocortical cancer
    - Cushing syndrome
    - congenital adrenogenital syndrome
  - Ovarian causes
  - Akromegaly
- 2. Idiopathic
- 3. Drugs
- 4. Other ï e.g. anorexia nervosa

genesis of adrenogenital syndromes

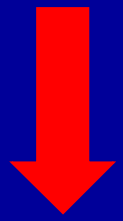


hogenital syndrome (AGS)

Deficiency of enzyme products (cortisol, mineralocorticoids)

AND

Increased concentration of steroid precursors

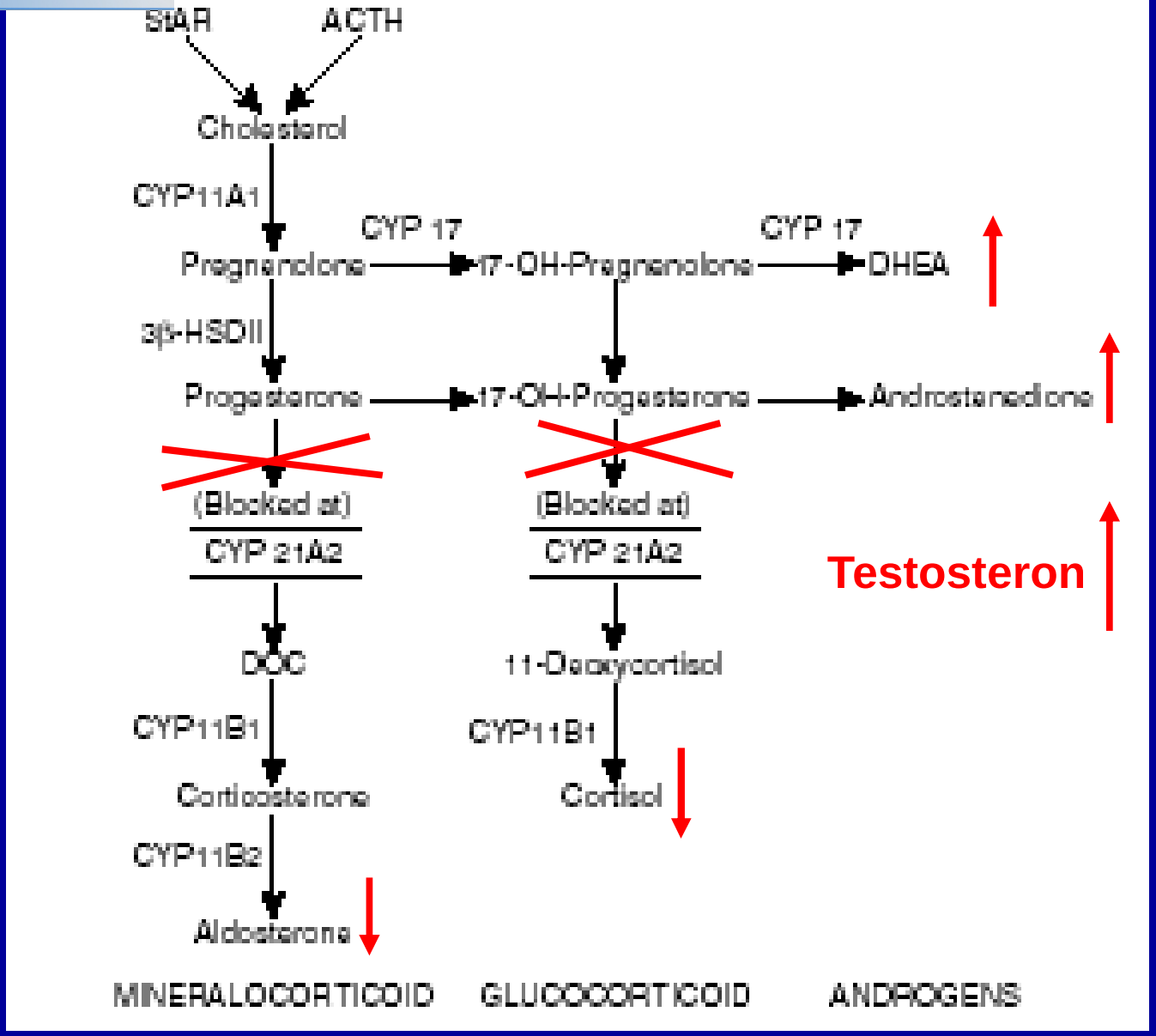


Virilisation
Female pseudohermaphroditism

Hydroxylase deficiency

- Most frequent cause of AGS
- **Major forms:**
  - Classic 21-Hydroxylase deficiency (1:14.000)
    - salt-losing form
    - simple virilising form
  - Non-classic 21-Hydroxylase deficiency (1:50-1:1000)
    - late-onset Form (postnatal virilisation, hirsutism, infertility, menstrual cycle abnormalities)
    - asymptomatic forms: only hormonal examinations can detect them

genesis of 21-hydroxylase deficiency



-hydroxylase deficiency

- **Salt-losing form:**
  - **deficiency of cortisol and aldosterone: hyperkalemia, hyponatremia, fluid loss, shock.**
- **Increased production of DHEA, androstendion and testosterone, therefore virilisation.**

iagnosis:

- **Increased serum concentraton of steroid precursors: 17-OH-Progesteron (>80 ng/ml) and urinary pregnantriol.**

Synacthen test (tetracosactid, 250 ε g i.v.)

Therapy of 21-hydroxylase deficiency

- **Glukokortikoids**
- **Hydrocortison:**
  - **Adults 20-30 mg/Tag**
- **Dexamethason**
  - **Adults: 0.25-0.5 mg am Abend**

Polycystic Ovarian Syndrome (PCO), Stein-Leventhal Syndrom

CO diagnostic criteria

- Hyperandrogenism with or without skin alterations
- Abnormalities of menstrual cycle ï raromenorrhoea or secondary amenorrhoea
- Other causes of hyperandrogenism excluded (e.g.: adrenogenital syndrome, Cushing syndrome)

ormone results in PCO

- Increased serum testosterone and DHEAS
- High insulin concentrations (oral glucose tolerance test, 75 g)
- Increased LH/FSH ratio

Therapy of PCO

- Weight reduction.
- Metformin against insulin resistance
- Contraceptive pills ï estrogen + cyproterone-acetate antiandrogenic protesterone derivative
- Induction of ovulation (Clomiphen, LH, FSH, GnRH agonists)
- Gynecologic interventions: laser coagulation, elektrocoagulation, partial wedge resection of the ovaries