

► Subject :

Classical CAH → Ambiguous genitalia

② salt wasting

or virilization → symptoms of excess Androgens.

non classical:

↓

- mild virilization

- oligomenorrhea

Cholesterol → Pregnenolone →

(peripheral) testosterone → estradiol → ① Bone growth

② sexual characteristics

~~no enzyme~~ CAH it is ~~not~~ Cholesterol desmolase (X)

① 21 hydroxylase → ① Progesterone → 11 deoxycorticosterone

② 17 hydroxy progesterone → 11 deoxycortisol

③ all enzymes def in CAH → causes cortisol deficiency. (if)

④ if 21 hydroxylase deficient → then 11 deoxycorticosterone X

No aldosterone.

→ also no 11 deoxycortisol X

No cortisol.

Subject:

→ Shift ↓ cortisol ↓

adrenal gland ↓ androgen ↑

So in 21 hydroxylase def → - aldosterone deficiency
- cortisol deficiency
- excess androgen.

~~low~~ aldosterone → distal tubules & collecting duct.
→ reabsorption of Na & water
→ K out
→ keep B.P. & electrolyte balance.

Low aldosterone → No absorption of water so hypotension
→ hypo Na / hyper K
→ metabolic acidosis

cortisol def → hypo glycaemia
→ hypo tension (stress H)

excess Androgen → female, prenatal → ambiguous genitalia
male → at birth no problem.
→ signs of precocious puberty after

male → hypotension shock / hypo Na / hypo glycaemia / hyper K /
metabolic A

female → if she had ambiguous genitalia without TTT, same pic ↓

► Subject :

21 hydroxylase def → ambiguous genitalia plus it's female !! (as in case)

② 11β-hydroxylase.

① 11-deoxy corticosterone → corticosterone

② 11-deoxy cortisol → cortisol

- if def →
- ① No cortisol
 - ② No aldosterone
 - ③ 11-deoxy corticosterone ↑
 - ④ 11-deoxy cortisol ↑
 - ⑤ Progesterone ↑
 - ⑥ 11-hydroxy progesterone ↑
 - ⑦ androgen ↑

- So aldosterone def & cortisol def & excessive androgen.

⊗ same C/P as 21 hydroxylase def but without salt

wasting ^{crises} ~~crises~~ How? b/c 11-deoxy corticosterone has

minimal corticoid effect but weak. → b/c its high quantity ↑

aldosterone deficiency !! (b/c)

- So No signs of ald def x

- ⊗ hypotension / hypoNa / hyperK / metabolic A
- ↪ HTN / hyperNa / hypo K / alkalosis (metabolic)

Subject: _____

3- β hydroxysteroid dehydrogenase

2) hydroxylase

So aldosterone & cortisol & Androgen def.

- Salt wasting \rightarrow Pregnenolone \uparrow \rightarrow not as mineral corticoid \times

Androgen def. \rightarrow male with ~~ambiguous~~ (ambiguous genitalia)

\downarrow
DHEA $\uparrow\uparrow\uparrow$ as androgen but in female (weak Androgen) \rightarrow AG \checkmark

female normal not AG \times

So 3 β HSD def \rightarrow AG in male & female.
but more in male.

17 α hydroxylase \rightarrow if def \rightarrow No cortisol or Androgen. \times

No androgen \rightarrow male AG.

\rightarrow secondary sex characteristic problem in male.

① HTN / hypok \rightarrow hyper Na / metabolic alkalosis

② hypoglycemia

③ AG in male.

► Subject :

21-hydroxylase def \subseteq CAH $\hat{=}$ enzyme $\hat{=}$

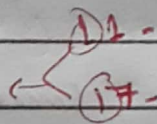
more than 90% of CAH with 21-Hydroxylase def ✓

① AG:

17- α hydroxylase def AG 1,2,4,5 female 21

17 \times 3 \rightarrow male AG.

② HTN:



3-
20- \rightarrow salt wasting

management \rightarrow all CAH \rightarrow give steroids.

- for ald. def \rightarrow give syntho with mineralocorticoid effect

- give K or Na bicarb according

- for Androgen def.

AG \rightarrow surgical correction after: ① U/S internal organ

② CT / MRI

③ Chromosomal analysis

Karyotyping

Subject :

For Dx → check B.P

→ check electrolyte

→ check glucose

→ check Blood gas

→ karyotyping

→ enzyme study → to know which enzyme deficient.

for 21 hydroxylase def → 17 hydroxy progesterone. (serum)

AG in female → ① Clitoromegaly

(external genitalia)
looks like male

② partial fusion of Labia majora.

③ overgrowth in labia majora. (scrotal like)

① 21 hydroxylase def → m.c. presentation in female? AG.

② 17 " " → " ? HTN

③ 3-β " " → " ? salt wasting.

④ 21 " in male → after 2-3m = shock in ER. ✓ No AG.

⑤ 17 → M.C
AG & HTN

* 3 β → M.C
AG & salt wasting

AG in male → ① bifid scrotum like labia majores.

② hypospadias

(AG like female or 15)

Dx

3 beta → 17 hydroxy pregnenolone ↑

17 hydroxy progesterone ↓

17-hydroxy progesterone → (screening test) for CAH

~~17-OH progesterone~~

in male & AG → low

in female & AG → low 17H → 3 beta def

in 21 & 11 B → high.

in 3B, 17a → low

① bone age for Precocious Puberty & male ^{ends} → Short stature

if AG → ① surgical repair.

② or local → estrogen cream (for female with AG & fusion)

long term = replacement ✓ hydrocortison (orally)

Subject:

Mineralocorticoid
glucocorticoid

long term → Hydrocortisone (maintenance dose)

↑ Steroids → - any infection - decrease activity - operation

⊖ no response to hydrocortisone → Fludrocortisone
↓
aldosterone

androgen production ↓ cortisol level ↓ CAH ↓

↓
negative feedback on ACTH & CRH

↓ Sex hormone production: - normal height
- no normal growth.

So hydrocortisone ① cortisol
② suppression hypothalamic pit adrenal axis

⊗ Prenatal screening test for mother of previous (CAH)

⊖ Amniocentesis ⊖ CVS

↓
⊖ 17 hydroxy progesterone level

if suspected CAH → give Steroids