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Sunday, November 5, 2017

## Steroid Hormone synthesis pathway (Clinical aspect)

Hello Awesomites :D

I was reviewing the corticosteroid synthesis pathway and its applied.

Let us begin. :))

Adrenal gland consist of two parts :-

1. MEDULLA

2. CORTEX

Adrenal dysfunction includes hyperfunction / hypofunction of medulla and cortex.

1. MEDULLA

**A) HYPERFUNCTION - pheochromocytoma  
-Neuroblastoma**

2. CORTEX

**A) HYPERFUNCTION -Conn's disease  
-Cushing's syndrome (Primary tumors)  
(excess ACTH -pituitary hypersecretion, ECTOPIC)**

**B) HYPOFUNCTION -ACTH deficiency (Iatrogenic , pituitary insufficiency)**

**C) CONGENITAL ADRENAL HYPERPLASIA (from partial enzyme deficiencies due to mutation in genes)**

Clinical features of CAH :-

1. DUE TO DECREASED ALDOSTERONE :-

-Sodium wasting (hyponatremia+dehydration+shock)

(early presentation)

-increased potassium

-acidosis

2. DUE TO DECREASED CORTISOL:-

-Hypoglycemia

-increased ACTH

3. DUE TO INCREASED TESTOSTERONE :-

In female , virilization.

In male, No symptom, increased size and pigmentation of penis.

Q. What is the difference between 21-hydroxylase deficiency and 11beta hydroxylase deficiency ?

Ans. In 21-hydroxylase deficiency, hypotension occurs due to salt wasting.

Accumulation of 11-deoxycorticosterone as a result of 11 beta hydroxylase deficiency leads to "HYPERTENSION".

Q. Most common form of CAH is due to mutation or deletion of which gene?

Ans. *CYP21A resulting in 21-HYDROXYLASE DEFICIENCY* .

Q. Which Enzyme deficiency showing virilization in females?

Ans. -21 hydroxylase

-3beta HSD

-11 Beta hydroxylase.

Q. Two hypertensive form of CAH.

Ans. *11beta hydroxylase and 17hydroxylase deficiency*.

Other points :

- Females with 17-hydroxylase deficiency appear phenotypically female at birth but do not develop breasts and menstruate in adolescent because of INADEQUATE ESTRADIOL PRODUCTION(17 hydro Pregnenolone is also a precursor of estrogen). They may present with **hypertension**.

-CAH is a type of enzyme deficiency. So it can be partial or complete. There is a severity spectrum.

More severe form shows salt wasting.

Milder form shows "NON CLASSICAL TYPE of CAH".

Diagnosis:-

- 17hydropregnenolone with or without ACTH test
- CYP21A2 panel, sequencing, deletion
- Carrier screening test (Preconception test)
- Karyotyping ( In case of ambiguity of sex)
- Hormones and electrolytes

Treatment:-

- Counsel the parents.

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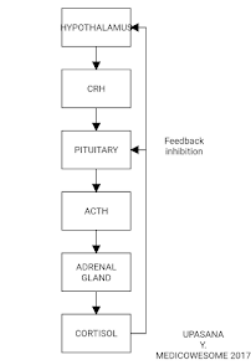
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What others are reading :)

- Protect from Adrenal insufficiency ( Give mineralocorticoid and glucocorticoid)
- Avoid salt wasting crisis during illness, stress, etc. ( Increase dose of glucocorticoid, Give IV fluids and sodium and dextrose)
- Surgery ,sex assignment.

(Note :- There are two more variants of CAH 1. Lipoid CAH

2. POR deficiency ( P450 oxidoreductase enzyme deficiency) - also involved in both sterol and steroid synthesis pathway).



**VARIANTS OF CONGENITAL ADRENAL HYPERPLASIA**

**21hydroxylase deficiency :-**  
 Aldosterone - decreases  
 Cortisol - decreases  
 Testosterone- increase  
 Ambiguous genitalia- 46+XX

**17 hydroxylase /17,20 lyase deficiency :-**  
 Aldosterone - increases (+hypertension)  
 Cortisol -decreases / normal  
 Testosterone-decreases  
 Ambiguous genitalia- 46+XY

**11Beta hydroxylase deficiency :-**  
 Aldosterone - decreases (DOC increases causes hypertension)  
 Cortisol- decreases  
 Testosterone- Increases  
 Ambiguous genitalia- 46+XX

**3Beta-hydroxysteroid dehydrogenase:-**  
 Aldosterone - decreases  
 Cortisol -decreases  
 Testosterone- In male, low levels.  
 In females, normal levels.  
 Ambiguous genitalia-46+XY

UPASANA Y. MEDICOWESOME 2017

Study hard.  
-Upasana Y. :)

Posted by Upasana Y. 7:03 PM

Reactions:  Interesting (0)  Kissable (0)  Awesome (0)  Nice (0)



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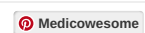
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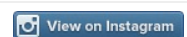


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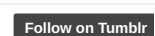
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