Week 8 Clinical Endocrinology

Adrenal Disorders 2

**Congenital Adrenal Hyperplasia**

* Etiology: mutations in genes encoding steroidogenic or cofactor enzymes
* Clinical Manifestations:

1. 21-hydroxylase deficiency

* GC & MC deficiency
* adrenal androgen excess

1. 11B-hydroxylase deficiency

* GC deficiency
* MC & adrenal androgen excess

1. 17a-hydroxylase deficiency

* GC & adrenal androgen deficiency
* MC excess
* Evaluation:

1. 21-hydroxylase deficiency

* Serum 17-OHP, 21-DOC
* Urine pregnanetriol, 17OH pregnanolone, pregnanetriolone

1. 11B-hydroxylase deficiency

* Serum 11-DOC, 11 deoxycorticosterone
* Urine tetraOH 11-DOC, tetraOH 11-deoxycorticosterone

1. 17a-hydroxylase deficiency

* Serum11-DOC, corticosterone, pregnenolone, progesterone
* Urine tetraOH 11-deoxycorticosterone, tetraOHcorticosterone, pregnenediol, pregnanediol
* Treatment:

1. Children:

Hydrocortisone – prevent adrenal crisis

Prednisolone – control androgen excess

1. Adult:

Prednisone – suppress excess androgen

Dexamethasone – achieve fertility

1. Mineralocorticoid and salt replacement

* Biochemical monitoring: Androstenedione, testosterone, renin
* Marker for overtreatment: 17OHP

**Pheochromocytoma**

* Clinical manifestations:
* “the great masquerader”
* headache, sweating, palpitation – triad
* tachycardia, hypertension, anxiety, panic, pallor, nausea, abdominal pain, weakness, weight loss, paradoxical response to antihypertensive drugs, polyuria, polydipsia, constipation, orthostatic hypotension, dilated cardiomyopathy, erythrocytosis, elevated BG, hypercalcemia
* Etiology:
* catecholamine producing tumors
* sympathetic or parasympathetic nervous system
* sporadic or inherited
* Evaluation:
* Plasma and urinary fractionated catecholamines and metanephrines
* CT or MRI
* MIBG, somatostatin receptor scintigraphy, dopa PET
* Treatment:
* Removal of tumor
* Preoperative preparation
  + alpha-adrenergic blocker (phenoxybenzamine)
  + liberal salt and hydration
  + prazosin or phentolamine
  + beta-blocker after alpha blockers
  + calcium channel blockers or ACE inhibitors