Familial Hypertension Syndromes

Abbreviations:
- GRA = Glucocorticoid Remediable Aldosteronism
- FHA-II = Familial Hyperaldosteronism type II
- CAH (11β) = Congenital Adrenal Hyperplasia
- CAH (CYP11) = Congenital Adrenal Hyperplasia
- GR = Glucocorticoid Resistance
- FHH = Familial Hyperkalemic Hypertension
- AME = Apparent Mineralocorticoid Excess
- AMR = Activating MR mutation
- HTN-Br = HTN with Brachydactyly

Synonyms:
- Familial Hyperaldosteronism type I
- 11β-hydroxylase deficiency, CYP11B1 deficiency
- 17α-hydroxylase/17,20-Lyase deficiency, CYP17 deficiency
- Primary Cortisol resistance
- Gordon’s syndrome, Chloride Shunt syndrome
- 11β-Hydroxysteroid Dehydrogenase type II Deficiency
- Geller’s syndrome, MR L810, HTN worsened in Pregnancy

References:
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Syndrome | Heritance | Aldo | PRA | K+ | Diagnostic Clues | Specific Treatment
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1. GRA | AD | ↑↑ | ↓ | - | ↑Aldo, Dex suppression, Fam. Hx., 18-OH steroids | Glucocorticoid
2. FHA-II | AD | ↑↑ | ↓ | - | ↑Aldo, No Dex suppression, Fam. Hx., adrenal nodules | Genetics unkown; MR antagonist
3. CAH (11β) | AR | ↓ | ↓ | - | ↑Cortisol, ↑Androgen, ↑DOC, ↑11-deoxycortisol | Glucocorticoid replacement
3. CAH (CYP17) | AR | ↓ | ↓ | - | ↑Cortisol, ↓Androgens, ↑DOC, ↑18OH-B | Glucocorticoid/Androgen replacement
4. GR | AD | - | - | ↓ | ↑↑Cortisol, ↑↑ACTH | MR antagonist
4. Liddle’s | AD | ↓ | ↓ | - | Amiloride response | Amiloride/Triamterene
5. FHH | AD | - | - | ↑ | Hyperkalemia, mild Acidosis, Hypercalciuria | Thiazides
5. AME | AR | ↓ | ↓ | - | MR antagonist response, ↑Cortisol/Cortisone ratio | MR antagonist
5. AMR | AD | ↓ | ↓ | - | Worsens with spironolactone | Block downstream (Amiloride)
5. HTN-Br | AD | - | - | - | - | Unknown

Cholesterol

- Cholesterol Dehydrogenase
- CYP17
- 17α-Hydroxylase
- 11β-Hydroxylase
- 18-Hydroxylase (CYP17)
- Aldosterone Synthase

- 18-Hydroxydeoxycorticosterone
- Cortisol Synthase