VANDERBILT School of Medicine

James M. Luther, MD MSCI

Familial Hypertension Syndromes

Abbreviations:

GRA = Glucocorticoid Remediable Aldosteronism
FHA-II = Familial Hyperaldosteronism type II
CAH (11β) = Congenital Adrenal Hyperplasia
CAH (*CYP17*) = Congenital Adrenal Hyperplasia
GR = Glucocorticoid Resistance
FHH = Familial Hyperkalemic Hypertension
AME = Apparent Mineralocorticoid Excess
AMR = Activating MR mutation
HTN-Br = HTN with Brachydactyly

Synonyms	References[1, 2]
Familial Hyperaldosteronism type I	[3]
	[4, 5]
11βhydroxylase deficiency, *CYP11B1* deficiency	[6]
17α-hydroxylase/17,20-Lyase deficiency, *CYP17* deficiency	[6]
Primary Cortisol resistance	[7-10]
Gordon's syndrome, Chloride Shunt syndrome	[11-14]
11βHydroxysteroid Dehydrogenase type II Deficiency	[15]
Geller's syndrome, MR_{L810}, HTN worsened in Pregancy	[16]
	[17]

	Syndrome	Heritance	Aldo	PRA	K^+	Diagnostic Clues	Specific Treatment
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 uknown; MR antagonist
3.	CAH (11β)	AR	↓	↓	-/↓	↓Cortisol, ↑Androgen, ↑DOC, ↑11-deoxycortisol	Glucocorticoid replacement
3.	CAH (*CYP17*)	AR	↓	↓	-/↓	↓Cortisol, ↓Androgens, ↑DOC, ↑18OHB	Glucocorticoid/Androgen replacement
4.	GR	AD	-	-/↓	↓	↑↑Cortisol, ↑↑ACTH	MR antagonist
1.	Liddle's	AD	↓	↓	↓	Amiloride response	Amiloride/Triamterene
2.	FHH	AD	-	-/↓	↑	Hyperkalemia, mild Acidosis, Hypercalciuria	Thiazides
3.	AME	AR	↓	↓	↓	MR antagonist response, ↑Cortisol/Cortisone ratio	MR antagonist
4.	AMR	AD	↓	↓	-	Worsens with spironolactone	Block downstream (Amiloride)
5.	HTN-Br	AD	-	-	-	brachydactyly, Turkish/German heritage	Unknown