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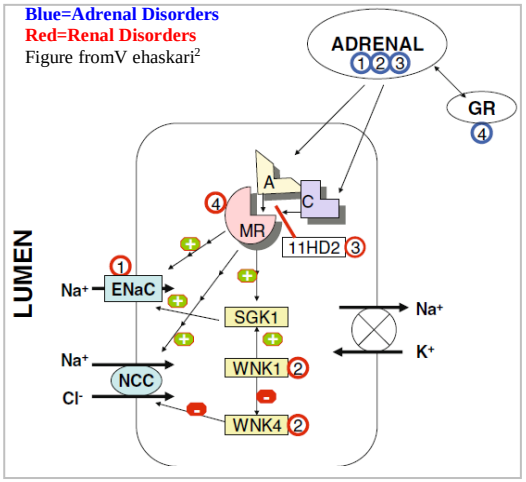
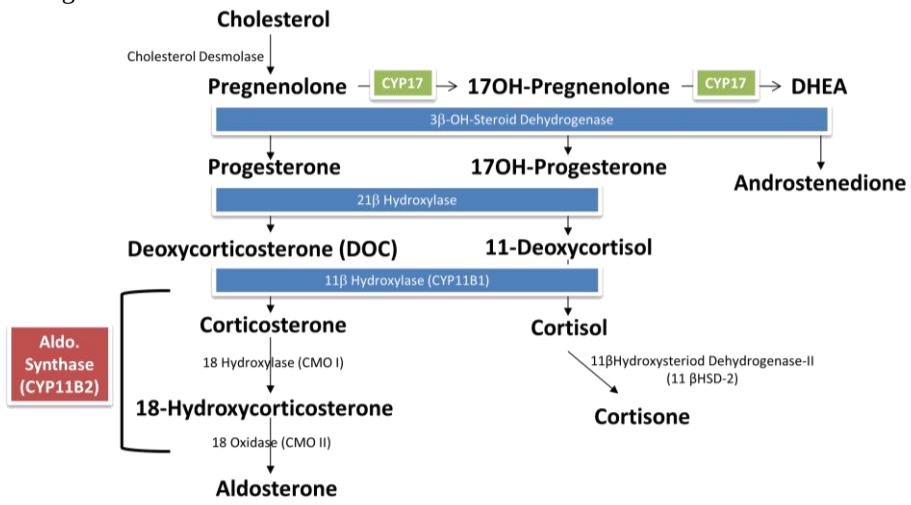
Familial Hypertension Syndromes

Abbreviations:	Synonyms/Mutation	References[1-4]
GRA = Glucocorticoid Remediable Aldosteronism	Familial Hyperaldosteronism type I; CYP11B2/B1 cross-over	3, 4
FH-II = Familial Hyperaldosteronism type II	*CLCN2* mutation	3-6
FH-III = Familial Hyperaldosteronism type III	*KCNJ5* mutation	3, 4, 7, 8
FH-IV = Familial Hyperaldosteronism type IV	*CACNA1H* mutation	3, 4, 9
PASNA = Primary Aldosteronism with Seizures and Neurologic Abnormalities, *CACNA1D* mutation		3, 4
CAH (11β) = Congenital Adrenal Hyperplasia	11βhydroxylase deficiency, *CYP11B1* deficiency	10
CAH (*CYP17*) = Congenital Adrenal Hyperplasia	17α-hydroxylase/17,20-Lyase deficiency, *CYP17* deficiency	10
GR = Glucocorticoid Resistance	Primary Cortisol resistance	11, 12
FHH = Familial Hyperkalemic Hypertension	Gordon's syndrome, Chloride Shunt syndrome	13, 14
AME = Apparent Mineralocorticoid Excess	11βHydroxysteroid Dehydrogenase type II Deficiency	15
AMR = Activating MR mutation	Geller's syndrome, MR_{L810}, HTN worsened in Pregnancy	16
HTN-Br = HTN with Brachydactyly		

Syndrome		Heritance	Aldo	PRA	K^+	Diagnostic Clues	Specific Treatment
1.	FH-I/GRA	AD	↑↑	↓	-/↓	↑Aldo, Dex suppression, Fam. Hx., 18-OH steroids, *CYP11B2/B1*	Glucocorticoid
2.	FH-II	AD	↑↑	↓	-/↓	↑Aldo, No Dex suppression, Fam. Hx., adrenal nodules, *CLCN2*	MR antagonist
3.	FH-III	AD	↑↑	↓	-/↓	↑Aldo, childhood onset, *KCNJ5*	MR antagonist
4.	FH-IV	AD	↑↑	↓	-/↓	↑Aldo, incomplete penetrance, *CACNA1H*	MR antagonist
5.	PASNA	sporadic	↑↑	↓	-/↓	↑Aldo, incomplete penetrance, *CACNA1H*	MR antagonist
6.	CAH (11β)	AR	↓	↓	-/↓	↓Cortisol, ↑Androgen, ↑DOC, ↑11-deoxycortisol	Glucocorticoid replacement
7.	CAH (*CYP17*)	AR	↓	↓	-/↓	↓Cortisol, ↓Androgens, ↑DOC, ↑18OHB	Glucocorticoid/Androgen replacement
8.	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





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