**Supplemental Material**

**Case summaries according to molecular etiologies**

***MC2R-*** **Melanocortin 2 receptor gene**

Six patients from 6 unrelated families were diagnosed between the ages of 4 days to 6.25 years (median 45 days). One patient was identified during screening due to an older sister with known *MC2R* mutations at the first week of life at a pre-symptomatic stage. However, this patient had elevated ACTH and low cortisol levels at 4 days of life (ACTH: 587 pg/mL, cortisol: 1.2 µg/dL). Four patients presented with hypoglycemic seizures and hyperpigmentation in early infancy. One other patient presented hyperpigmentation, fatigue and weight loss at 6.25 years old. None of the patients had mineralocorticoid deficiency although 2 patients had mild hyponatremia (serum sodium: 126-129 mEq/L, normal range 136-146) at the presentation that resolved without fludrocortisone treatment. Four patients had mildly elevated TSH concentrations (maximum TSH level 10/7.6/6.4/6.3 mU/L) but free T4 levels were within the normal range. One female patient with homozygous c.476C>A (p.Thr159Lys) mutation had pediatric cardiology follow-up due to dilated cardiomyopathy. One year after the diagnosis of PAI, echocardiographic findings improved and cardiological treatment (digoxin, enalapril) was discontinued. Another female patient with a homozygous c.455C>A (p.Thr152Lys) mutation was diagnosed with galactose-glucose malabsorption with osmotic diarrhea and hypernatremic dehydration in the neonatal period. This patient also had precocious puberty and short stature without growth hormone deficiency.

***MRAP*- Melanocortin-2 receptor accessory protein gene**

One of the patients presented with hypoglycemic convulsion and hyperpigmentation at infancy. The other patient had inotrope resistant hypotension and hyperpigmentation at the first two days of life. None of them had mineralocorticoid deficiency.

***StAR*-** **Steroidogenic acute regulatory protein gene**

Six female patients with *StAR* defects presented with a salt-losing crisis in the first four months of life. Four patients were 46,XY females without Mullerian structures in pelvis ultrasonography and their gonads were observed in the inguinal canal. Other two patients were siblings and had 46,XX karyotypes. They also had elevated gonadotropin concentrations. The adrenal gland morphology was evaluated by ultrasonography at the time of diagnosis and did not show enlargement despite high plasma ACTH levels.

***CYP11A1*-** **Cytochrome P450 family 11 subfamily A member 1 gene**

Two patients with *CYP11A1* mutations presented with a salt-losing adrenal crisis and generalized hyperpigmentation. One 46,XY female patient who was diagnosed 10 hours after birth due to severe hypotension which is resistant to vasopressor support and generalized hyperpigmentation. She had a history of a sibling who died with a salt-losing adrenal crisis at 15. day after birth. Genetic analysis revealed novel homozygous c.461T>C (p.Leu154Pro) in *CYP11A1*. One other male patient was diagnosed at infancy. He had normal male external genitalia and his sibling had died at 1.5 years with hyponatremic dehydration. All patients had mineralocorticoid deficiency and were treated with fludrocortisone.

***NNT -*Nicotinamide Nucleotide Transhydrogenase gene**

Three patients with *NNT* defects were presented with hypoglycemic convulsion and hyperpigmentation at infancy. One female patient with *NNT* mutation [compound heterozygous for c.2507G>A p.(Gly836Asp) / c.1594C>T p.(Leu532Phe)], had mild mineralocorticoid deficiency with high renin (>234 pg/mL, normal range for age 0.8-16 pg/mL), subnormal serum sodium (133 mEq/L) and slightly high serum potassium (5.6 mEq/L; normal range 3.5-5.3 mEq/L) concentrations under hydrocortisone replacement therapy. Mineralocorticoid therapy was added to the treatment of the other female patient with homozygous c.259C>T (p.Gln87\*) variant in *NNT* because of mild hyponatremia (128-135 mEq/L) and high renin concentrations (21.8-91.5 pg/mL) under glucocorticoid therapy. The other male patient with homozygous deletion had normal aldosterone production.

***NR0B1 / DAX-1-*** **Congenital Adrenal Hypoplasia**

Three patients with *NR0B1/DAX1* defects were presented with vomiting and hyponatremic dehydration at infancy. The diagnosis of hypogonadotropic hypogonadism was established in one patient at the age of 17 by low gonadotropins and testosterone, and small testicular volumes. One patient had micropenis at the presentation. The other patient had normal male genitalia. All patients had mineralocorticoid deficiency and were treated with fludrocortisone.

***SGPL1-*** **Sphingosine-1-phosphate lyase 1 gene**

Two patients with *SGPL1* mutations were diagnosed at early infancy **(Supplemental Table 1)**. One male patient with nephrotic syndrome was diagnosed with PAI while checking for bilateral cryptorchidism. The patient had hyponatremia, hyperkalemia, and high renin concentrations. He had high gonadotropins and low testosterone (LH: 52.6 µIU/mL, FSH: 71.5 µIU/mL, total testosterone: 0.1 ng/mL). Renal insufficiency was also added during his follow-up. Second patient was a female who was being followed up for adrenal calcification detected bilaterally on prenatal ultrasonography. ACTH concentrations gradually increased and glucocorticoid therapy was initiated at 2 months of age. Proteinuria and renal failure developed at 3 months of age. Mineralocorticoid treatment was added to her treatment at the 8th month.

***AIRE*-Autoimmune Polyglandular Syndrome Type 1**

One male patient with compound heterozygote *AIRE* mutation presented with hyperpigmentation and hyponatremia at 4 years old. He had hypothyroidism due to Hashimoto's thyroiditis. At 11 years old type 1 diabetes mellitus and at 14 years old autoimmune hepatitis and optic neuritis developed. The patient had mineralocorticoid deficiency and was treated with fludrocortisone.

***ABCD1-*Adrenoleukodystrophy**

One male patient with *ABCD1* mutation was presented with hyperpigmentation and frequent infections at 8 years old. He had no neurological signs and symptoms and had normal cranial magnetic resonance imaging. Very-long-chain fatty acid (VLCFA) concentrations were in normal range before and after molecular diagnosis.

***AAAS*-Triple A Syndrome**

One female patient with *AAAS* mutation presented with hyperpigmentation and weight loss at 4 years old. The parents did not notice alacrimia. There was no family history. After getting a diagnosis of Triple A syndrome Schirmer's test confirmed alacrimia in this patient. She developed dysphagia at 5 years old.

***HSD3B2-*3β-Hydroxysteroid Dehydrogenase Type 2 Deficiency**

One female patient presented with hyperpigmentation and failure-to-thrive at neonatal period. She had hyponatremic and hyperkalemic dehydration with high renin. Her karyotype was 46,XX and she had normal female genitalia. Forty-eight hours after stopping glucocorticoid treatment, 17-hydroxyprogesterone was assessed by the LC-MS/MS method and level was found to be 1.9 ng/ml. Other female patient presented with vomiting and failure-to-thrive at neonatal period. She had hyponatremic and hyperkalemic dehydration. Mutational analysis of the patient revealed compound heterozygosity for c.939del (F314Sfs\*54) and c.745C>T (R942\*) variations in the *HSD3B2* gene. There was no evidence of genital virilization and 17-hydroxyprogesterone concentration was 3.25 ng/mL (normal range 0.2-2.3 ng/mL).