

partial gene deletion, c.1886 (c.2817 + 1_c.2818-1)del on the *EIF2AK3* gene. The patient's parents were both heterozygous for this mutation and are therefore carriers of WRS.

Case 2: The three-month-old sister of the first case was diagnosed as diabetes. Because of her family history, the diagnosis was confirmed with genetic testing which revealed the same partial gene deletion as in her brother.

Hepatic and renal dysfunctions are typical features of this syndrome. Our first patient presented with typical symptoms and signs of WRS. Although case 2 does not have the typical signs of the syndrome, it may develop later. Children with WRS usually present in the first few months of life with diabetes, and it is recommended that any child presenting with diabetes within the first 2 years of life should be tested for *EIF2AK3* mutations.

(P-17)

Isolated Hypoaldosteronism: A Case Report

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Isolated hypoaldosteronism (IHA) is a rare (1/1.000.000) AR disorder caused by mutations in *CYP11B2* gene and may result in life-threatening salt wasting and failure to thrive. We presented this case because of the rareness of disease and our patient is only the second Turkish case with a genetically confirmed diagnosis.

A 3-day-old male first presented with jaundice and physical examination with normal findings. The parents are Turkish and consanguineous. Initial laboratory examinations showed hyponatremia (129 mEq/L) and hyperkalemia (6.9 mEq/L). Endocrinological evaluation showed low plasma aldosterone concentration of 40 pg/mL (50-900 pg/mL) and markedly elevated plasma renin activity (PRA) > 200 ng/mL/hr (2.35-370 ng/mL/hr); cortisol level after adrenocorticotropic hormone stimulation was 31.5 ug/dL. He was started a fludrocortisone treatment as 0.1 mg/daily with IHA diagnosed. Fludrocortisone dose was raised to 0.4 mg/daily. At the age of 3 years, hypertension was detected while his electrolyte levels were normal. His treatment was discontinued. At the eighth day without treatment, aldosterone was 10 pg/mL (50-900 pg/mL), PRA >10 ng/mL/hr (1-6.5 ng/mL/hr), corticosterone 1.5 ng/mL (0-3.5 ng/mL), 18-OH corticosterone 15 ng/dL (6-85 ng/dL), 18-OH corticosterone/aldosterone 15 (2.4-10.5), Na, 132 mmol/L, and K 5 mmol/L.

Genetic sequencing identified that the proband has homozygous p.I236N mutation in *CYP11B2* gene and his parents were both heterozygous. Despite this mutation was not reported in any database, another Turkish family was reported with same clinical features recently. PolyPhen-2, SIFT, and MutationTaster indicated this mutation as harmful.

Although there is no functional study of the reported p.I263N mutation which is assumed to be the cause of the disease, we present this case because two independent families were reported with the same clinical features and the mutation was predicted to be harmful by *in silico* methods.

(P-18)

Evaluation of the Response to the First Two Years of Growth Hormone Treatment in Kabuki Make-Up Syndrome

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The Kabuki make-up syndrome (KMS) is characterized by mental retardation, typical facial appearance, skeletal anomalies, joint laxity, and post-natal growth deficiency. There are limited publications on growth hormone therapy in KMS. Our aim is to present the response to growth hormone (GH) treatment in two KMS patients with GH deficiency in the first two years.

Case 1: A girl with KMS who started treatment for GH at 11.4 years old had a height of 128.6, 138.8, and 146.9 cm, a height SDS of -2.98, -2.6, and -2.01, a growth velocity of 3.2, 9.9, and 8.4 cm/year, and growth velocity SDS of -3.41, +4.14, and +3.13 at pretreatment, one-year, and two-year follow-up on treatment, respectively.

Case 2: The second case whose GH treatment was started at the age of 5.2 years had height of 94.8, 102.2, and 109.2 cm, height SDS of -3.31, -2.96, and -2.54, growth velocity of 4, 7.4, and 7 cm/year, and growth velocity SDS of -2.3, +1.6, and +1.09 at pretreatment, one-year, and two-year follow-up with treatment, respectively.

In our cases, a good response to GH treatment was obtained as in the few patients in the literature.

The post-natal growth retardation seen in 100% of patients with KMS can be accompanied by lack of GH.