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A Case Presentation: Sleeve Gastrectomy with Transit Bipartition as a Treatment of Type 2 Diabetes Mellitus Applied for the First Time to a Bulgarian Citizen

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Sleeve gastrectomy (SG) with transit bipartition (TB) was applied to a Bulgarian patient for the first time and no other case presentation was found in the literature using this treatment method in Bulgaria. Our aim is to introduce and disseminate this procedure in our country for the treatment of patients with type 2 diabetes mellitus (T2DM).

A 40-year-old gentlemen, height 176 cm, weight 115 kg (BMI: 37.2 kg/m^2), presented with a 3-year history of T2DM. His grandmother has T2DM. First, he was admitted in a hospital in Sofia. His Hba1C level was 9.31 % and blood glucose was 16 mmol/L. He was on treatment with metformin 850 mg morning and evening 2 times daily. Patient complaints were polyuria, polyphagia, weakness, and headache. He was informed about metabolic surgery and he referred to the clinic in Istanbul willingly to have a surgical operation.

In May 2016, he underwent laparoscopic SG with TB in İstanbul. The patient recovery was successful, and 16 kg weight loss was observed in 4 months. Hba1C value was observed in normal range -6 %. He is not on any drug treatment for his T2DM.

Surgical treatment options for diabetes mellitus are available nowadays to treat patients with obesity. The ABCD score, which comprise age, BMI, C-peptide level, and duration of T2DM (years) was reported as useful in predicting the success of T2DM treatment using metabolic surgery. SG with TB operations are getting more popular, but in our country, this is the first case of a patient treated with SG + TB.SG + TB is a simple procedure that results in rapid weight loss and remission or major improvement of comorbidities. As a conclusion of this case report, TB is an excellent complement to SG.

(P-15)

Incidentally Detected Monogenic Diabetes Case

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We present our case to emphasize that monogenic diabetes should be considered in young patients having positive family history and whom diabetes could not be classified.

A 25-year-old female was referred to our clinic due to elevated blood glucose. The patient had no symptoms of hyperglycemia. She had no chronic illness and did not take any medication. Her 55-year-old mother has been followed for uncomplicated diabetes for 16 years with oral antidiabetics, and her 75-year-old grandmother had uncomplicated diabetes for 30 years treated with basal insulin. On physical examination, vital signs were stable, height 168 cm, body weight 70 kg, BMI 24.8 kg/m^2. Systemic examination was normal, and no findings of insulin resistance were present.

Laboratory findings revealed that fasting plasma glucose (FPG) was 140 mg/dL, postprandial plasma glucose 178 mg/dL, and HbA1C 7.2 %. Blood count and biochemical parameters were normal. Fasting C-peptide was 2.32 ng/mL, urine ketone negative, anti-GAD, ICA, and anti-insulin antibodies were negative. We recommended life-style modifications and metformin treatment. Then, considering the patient age, family history of diabetes, absence of insulin resistance, negative autoantibodies, and normal body mass index, we performed genetic analysis for maturity-onset diabetes of the young (MODY). Heterozygous mutation of p.R191W (c.571C > T) was detected in glucokinase gene, and diagnosis of MODY type 2 was confirmed. She was followed with life-style modifications without metformin. FPG and glucose tolerance test results of siblings of the patient were normal. Genetic screening was recommended for the family.

It may be difficult to determine the type of diabetes in young patients. In suspected cases, genetic analysis may help to establish the definite diagnosis of MODY.

(P-16)

A Rare Cause of Insulin-Dependent Diabetes: Two Siblings with Walcott-Rallison Syndrome

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Wolcott-Rallison syndrome (WRS) is a rare autosomal recessive disorder characterized by neonatal or early infancy onset insulin-dependent diabetes and epiphyseal dysplasia. Other frequent multisystem manifestations include recurrent hepatitis, renal dysfunction, failure to thrive, developmental delay, neutropenia, and hypothyroidism. Herein, we reported two siblings with WRS.

Case 1: A 14-month-old male infant was brought to the hospital for feeding difficulty and vomiting and was diagnosed as diabetic ketoacidosis. He developed liver and renal failure after admission and was managed appropriately. Later on, physical examination showed growth failure and skeletal abnormalities, as well as dysmorphic features. Because of accompanying diabetes and skeletal abnormalities, WRS was suspected and the diagnosis was confirmed by genetic analysis which revealed a homozygous

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 an. 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.