

(agalsidase alfa). His daughter had flushing in her face and she could not sweat. Plenty angiokeratoma were found on her body. Eye examination was normal and there was no cardiac pathology. The same mutation was detected. Enzyme replacement therapy has been started.

Despite the fact that Fabry disease is an X-linked disorder, several female heterozygote mutation carriers have distinct clinical symptoms. Our patient does not have any major characteristics of Fabry disease, but she presented with major depression and angiokeratoma besides she had heterozygote mutation. In the literature, few mutation cases especially in men were associated with depression; however, no data found for women. It is well-known that mutation and phenotype relation is very important to predict the prognosis of the illness. It should be kept in mind that heterozygote mutation of p.G261D (c.782G>A) may be related with depression with female patients.

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A Case of MEN 2A: D631Y Mutation

Banu Şarer Yüreklî¹, Hatice Özişik¹, Nilüfer Özdemir Kutbay¹, Hüseyin Onay², Gökhan Özgen¹

¹Ege University Faculty of Medicine, Department of Endocrinology and Metabolism Diseases, İzmir, Turkey

²Ege University Faculty of Medicine, Department of Medical Genetics, İzmir, Turkey

Multiple endocrine neoplasia 2A (MEN 2A) is a hereditary disease comprising medullary thyroid carcinoma (MTC) (95%), pheochromocytoma (50%), parathyroid hyperplasia or adenoma (15-30%). RET mutations are seen generally in exon 10. Exon 11, 631 codon mutations are not common in MEN.

A 29-year-old male patient applied to our clinic. His mother was operated and diagnosed with MTC. Heterozygous D631Y RET mutation was detected in his mother. After this result, our patient was evaluated for RET mutation and MEN. Calcitonin value of the patient was normal and no nodule was detected on thyroid ultrasound. RET oncogene was positive for our patient as D631Y mutation. For the screening of MEN components, twenty-four hour urinary metanephrine and normetanephrine were high. Magnetic resonance imaging revealed adrenal adenoma 29x27x31 mm in diameter at the left adrenal. The patient underwent an operation in 2014 and pathology was consistent with pheochromocytoma. Prophylactic thyroidectomy was recommended, however, the patient did not accept this. He has been followed for development of thyroid nodule and evaluation of calcitonin level. At last visit, laboratory examination revealed PTH of 31.3 pg/mL (15-65), Ca 10.2 mg/dL (8.6-10.2), TSH 3.98 µIU/mL (0.35-5.50), fT₄ 1.47 ng/dL (0.89-1.76), calcitonin 5.77 pg/mL (0-10), and 24-hour urinary metanephrine and normetanephrine were normal.

RET 631 codon mutation is seen rarely in MEN patients. This

genetic profile might be related to the less vigorous clinical disease behavior and the late onset of MTC.

Pheochromocytoma might be the first manifestation prior to the development of MTC.

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A Case of Androgen Insensitivity Syndrome Presenting with Micropenis

Hüseyin Anıl Korkmaz

Balikesir Atatürk State Hospital, Clinic of Pediatric Endocrinology, Balikesir, Turkey

The patients with androgen insensitivity syndrome can present with various phenotypic anomalies having as a common aspect the loss of reproductive characteristics.

A boy from non-consanguineous family was admitted to pediatric endocrine department because of micropenis. A 7-year and 8-month-old boy was born with 3650 g by caesarean section. On physical examination, height was 124.1 cm (25-50p), height SDS -0.27, weight 29.7 kg (75-90p), and weight SDS was 1.09. The patient was conscious, oriented, and well-nourished with normal secondary sexual characteristics for his age. Genital examination revealed a stretched penile length of 3 cm, penile width of 0.5 cm, and no axillary and pubic hair. Right and left testis were palpated in the scrotal sac.

Karyotyping revealed a normal 46,XY karyotype. Serum follicle-stimulating hormone, luteinizing hormone, and total testosterone levels were 0.79 mIU/mL (normal reference range < 6.7), 0.06 mIU/mL (normal reference range 0.3-6.0), and 4.80 ng/dL (normal reference range < 7), respectively. Serum testosterone level was increased in response to 1500 units/dose HCG stimulation test for 3 days. No mutation was found for 5-α reductase deficiency. Androgen insensitivity syndrome was diagnosed with hemizygote p.L863F (c.2587C>T) mutation.

We emphasize the importance of genetic analysis in patients with micropenis. Routine genetic analysis to confirm androgen insensitivity syndrome may predict the long-term prognosis and management.