were significantly higher. A bilateral adrenal mass was detected and bilateral surrenalectomy was performed. Plasma calcitonin level was high. A hypoechoic, coarse calcific thyroid nodule was detected. The patient underwent total thyroidectomy and neck dissection. The parathormone (PTH) level was normal. The RET mutation was positive in the patient. It was decided to screen the family. Second case: A 50-year-old male patient was called for MEN 2A family screening. Bilateral adrenal mass was detected. Bilateral surrenalectomy was performed. Calcitonin level of 267 pg/mL was detected. Hypoactive thyroid nodule aspiration was reported as AUS. Total thyroidectomy and central neck dissection were applied to the patient. Cranial involvement was also observed in the PET/CT scan for metastasis. A mass in the left cerebellum (hemangioblastoma?) was detected in brain MR. Third case: A forty-six-year-old female patient was evaluated; a mass with size of 56x64x50 mm in the left adrenal and normal right adrenal were detected. Metanephrine and normetanephrine were significantly high in the urine. Calcitonin level was significantly high. Firm thyroid nodule was detected. PTH was normal. Left adrenalectomy and total thyroidectomy were planned. The patient refused to be treated.

MEN 2A syndrome is the most common medullary thyroid cancer. Bilateral pheochromocytoma is common. Hyperparathyroidism is observed in 20-30 % of patients.

(P-58)

Investigation of Androgen Receptor Gen Mutation Spectrum in the Turkish Patients with Disorder of Sex Development

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Androgen insensitivity syndrome (AIS) is an X-linked recessive condition resulting in a failure of normal masculinization of the external genitalia in chromosomally 46,XY individuals. This failure of virilization can be either complete androgen insensitivity syndrome (CAIS) or partial androgen insensitivity syndrome (PAIS), depending on the amount of residual receptor function. Mutations in the AR gene on chromosome Xq12 cause AIS. In this study, we aimed to investigate the mutation spectrum in Turkish patients who had AR mutation analysis with suspected gender development disorder and AR insensitivity syndrome.

The *AR* gene from the DNA material isolated from the peripheral blood of patients was amplified using appropriate primers and sequenced using the new-generation sequence analysis technique on the Mi-Seq device.

In this study, molecular analysis results of 383 individuals who underwent AR genetic analysis in Ege University Medical Genetics Department between 2011 and 2016 were evaluated retrospectively. There were 44 mutations in these cases. Of the 44 cases detected in the mutation, 16 were affected and the karyotype was 46,XY. 28 of them are the 46,XX carrier mothers, carrier relatives, or siblings of the affected cases.

New mutations were detected in our studies between 2011 and 2016-L57Q, T576I, D691Y, P672R, Q739E, p.R544KfsX8, c.1745_1747delTCT, F726S, L881V, R102G, and L863F. Different mutations can be detected in AR gene in Turkish society. In cases with disorder of sex development, *AR* should be examined.

(P-59)

A Novel *HESX1* Mutation in a Case with Panhypopituitarism

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Pituitary gland insufficiency (hypopituitarism) is a clinical condition that results in inadequate production and release of pituitary hormones. The deficiency of one or more pituitary hormones is named partial hypopituitarism and the deficiency of all pituitary hormones is named panhypopituitarism. Hypopituitarism can be attributed to inherited or acquired causes. Our aim was to determine the molecular diagnosis in our panhypopituitarism patient with *HESX1* gene sequence analysis.

A 21-year-old woman was referred to our clinic with primary amenorrhea. Her medical history included use of growth hormone, thyroid hormone, and estrogen. Cranial MRI findings were consistent with empty sella syndrome. In the family history of the case, there was no consanguinity between the parents and no similar patient in the family. Based on findings and laboratory results, the diagnosis of panhypopituitarism was considered; *HESX1* gene sequence analysis from patient's peripheral blood revealed a heterozygous p.R128K mutation.