(P-01)

Late-Onset Congenital Adrenal Hyperplasia Diagnosed at 53 Years of Age

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Our aim was to present a case of late-onset congenital adrenal hyperplasia (LOCAH) diagnosed at 53 years of age because of bilateral surrenal adenoma (BSA).

At 53 years of age, 11 years post-menopausal woman was referred to our out-patient clinic due to BSA. Patient's physical examination was unremarkable and Ferriman-Gallwey score was 4. There was no history of diabetes or hypertension. Adenomatous lesions were detected in the right adrenal gland corpus (10 mm in diameter) and left adrenal gland corpus (20 mm in diameter) demonstrated as out-of-phase sequence signal loss on abdominal magnetic resonance imaging (MRI). There was no significant increase in lesion diameter from previous MRI.

Hormonal function tests for Cushing's syndrome, pheochromocytoma, and Conn's syndrome were negative. The patient have two sons after spontaneous pregnancy. She has no hirsutism. Basal level of 17-hydroxy progesterone (17-OH-P) was 6.53 ng/mL thus 250 mcg adrenocorticotropic hormone stimulation test was ordered. Test results were cortisol 0' = 8 ug/dL, cortisol 30' = 9.16 ug/dL, cortisol 60' = 10.12 ug/dL, 17-OH-P 0' = 11.43 ng/mL, 17-OH-P 30' = 42.85 ng/mL, and 17-OH-P 60' > 50 ng/mL. After the test, LOCAH and adrenal insufficiency were diagnosed. Hydrocortisone (25 mg/day) treatment was started. *CYP21A2* mutation analysis revealed homozygous mutations of p.Arg339His (c.1016 G > A) in the exon 8 and p.Pro453Ser (c.1357 C > T) in the 10th exon. Test for patient's family members was ordered.

Patients followed with BSA should be investigated for LOCAH, even postmenopausal ones.

(P-02)

8Q22.3-Q24.23 Duplication: A Case Report

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We present a rare case of 8q duplication in a patient with oral frenulum history and absence of mental retardation.

A 7-year-old girl was referred to our clinic for hypertrichosis and dysmorphic facial appearance. On physical examination, hypertrichosis, upslanted palpebral fissures, epicanthus, hypertelorism, microretrognathia, high and broad nasal root, distinct glabella, fine upper lip, broad and flat philtrum, and clinodactyly were detected. She had a history of an operation for oral frenulum. Haemogram, routine biochemistry, hormone profiles, karyotype analysis, and brain magnetic resonance imaging (MRI) as well as ophthalmology, otolaryngology, and child psychiatry consultations were requested.

Hemogram, routine biochemistry, hormone profiles, brain MRI results, and the ophthalmologic evaluation were normal. Chronic otitis media was detected on otolaryngologic examination. IQ test score was reported as 95. Chromosome analysis revealed a 46,XX,der(8)add(8)(q24.1) karyotype. Karyotypes of mother, father, and sister were normal. Array comparative genomic hybridization (aCGH) was done to determine where the extra material came from. A duplication of 35.9 Mb at 8q22.3-q24.23 was detected.

Our case had similar phenotypic features to 8q duplication cases, such as hypertrichosis, hypertelorism, microretrognathia, and long philtrum. However, to our knowledge, this is the first case of 8q duplication with oral frenulum and without mental retardation.

(P-03)

Parental View on the Terminology of Disorders of Sex Development

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Disorders of sex development (DSD) is a nomenclature proposed to defeat the discomfort of families and patients. The aim of this study was to investigate the perception and usage of terminology among the parents of DSD patients in our country.

The records of the DSD council between years 2008-2015 were reviewed retrospectively. Parents were contacted through telephone inquiries focusing on the terminology the parents knew and tend to use.

In total, 121 patients were evaluated in monthly meetings of DSD council and 79 inquiries were completed. Median age at diagnosis was 1 year (0-16 years). Forty-one percent of the