full-term weighing 3200 g and had no significant medical or family history. Body weight was 10.9 kg (-2.54 SDS), height 80.4 cm (-3.38 SDS), and head circumference was 48.3 cm (-0.57 SDS). Systemic examination was normal and her pubertal development was consistent with Tanner stage 1.

The laboratory workup showed TSH 1.99 µIU/mL (0.4-5.0 µIU/mL), fT_3 2.66 pg/mL (1.57-4.71 pg/mL), fT_4 0.62 ng/dL (0.8-1.9 ng/dL), and cortisol 6.42 µg/dL (5-25 µg/dL). Celiac antibodies were negative. Pituitary MRI was reported to be normal. L-thyroxine therapy was started with the diagnosis of central hypothyroidism. While euthyroid, the stimulation tests showed insufficient GH response and normal cortisol response thus GH therapy was initiated. At the age of 12 years and 6 months (bone age 12 years), serum prolactin, follicle-stimulating hormone, and luteinizing hormone (LH) levels were found to be 2.15 ng/mL (3.8-26.7 ng/mL), 0.31 mIU/L, and 0.27 mIU/L. Results of LHRH test was consistent with hypogonadotropic hypogonadism (peak LH 0.4 mIU/L) and estrogen therapy was started. Low-dose adrenocorticotropic hormone test was performed because of low basal cortisol. Cortisol response was insufficient and hydrocortisone treatment was added with the diagnosis of central adrenal insufficiency. Repeated MRI showed a pituitary length of 4 mm. *PROP1* analysis revealed a previously reported, homozygous c.301_302delAG (p.Leu102Cysfs*8) mutation.

Mutations of the *PROP1* primarily affects thyrotroph, lactotroph, gonadotroph, and somatotroph cells. Adrenocorticotropic hormone deficiency is variable. Genetic analysis is important for identification of etiology.

(P-33)

Schmid Type of Metaphyseal Chondrodysplasia with COL10A1 Mutation

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The Schmid type of metaphyseal chondrodysplasia (MCDS) is characterized by short stature, widened growth plates, and bowing of the long bones, resulting from autosomal dominant mutations of COL10A1.

We report a patient with MCDS and COL10A1 mutation.

A 4-year 7-month-old boy was referred to our hospital because of bowing of the legs and short stature. His mother showed short stature and bowing of the legs, too. His height and weight were 75 cm (< 3th p) (-2 SDS) and 21.6 kg (75-90th p), respectively. Ca, P, ALP, PTH, and 25-OH D levels were normal. Radiographs showed findings compatible with MCDS. p.W651*(c.1952G > A) (heterozygote) mutation in the *COL10A1* gene was identified. The patient was diagnosed with MCDS.

We report this patient with MCDS and COL10A1 mutation as it is a rarely seen case.

(P-34)

A Case Report of Seckel Syndrome

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Seckel syndrome is an inherited autosomal recessive disorder characterized by short stature, microcephaly, prominent nose, and typical facial appearance. DNA damages can be detected in different genes, mainly in 3rd and 18th chromosomes. By means of its genetic heterogeneity and easily detectable morphological features, clinical diagnosis can usually be made.

A 19-year-old female patient was diagnosed with Seckel syndrome in pediatric clinic due to typical features of this syndrome at the age of 15. The patient, who already had type 1 DM, applied to our clinic in order to be followed.

On physical examination, prominent nose, flat forehead, micrognathia, high-arched palate, triangular narrow face, and large pinnae were present. On physical examination, height was 136 cm, weight 40 kg, and BMI was 21.63 kg/m^2. Clinodactyly, nail dystrophy, and mental retardation were detected. On cardiac examination, systolic ejection pulse on pulmonary focus was detected. There was no narrative of consanguineous marriage. Her treatment was metformin 500 mg 2x1, pioglitazone 15 mg 2x1, lispro insulin 3x9 units, and glargine insulin 16 units. Her menstrual period was regular. In laboratory examination, FBG was 101 mg/dL, HbA1c was 8.6%, B12 was 176 pg/mL (197-866), hypophyseal hormones were normal. Euthyroid Hashimoto thyroiditis was present. In echocardiography, ASD secundum 6-7 mm was detected. Vitamin B12 replacement was started; pioglitazone was stopped while the doses of insulin were increased. Last value of HbA1c was 6.6%.

Owing to its genetic heterogeneity, molecular prenatal diagnosis is difficult. Involvements may occur in endocrine, cardiac, gastrointestinal, and hematological systems.