# Multiple Pituitary Hormone Deficiency Associated with Pituitary Hyperplasia: A Case Report

Mikayir Genenş[1], Neşe Akcan[1], Zehra Yavaş Abalı[1], Firdevs Baş[1], Oya Uyguner[2], Şükran Poyrazoğlu[1], Güven Toksoy[2], Rüveyde Bundak[1], Feyza Darendeliler[1]

*[1]İstanbul University İstanbul Faculty of Medicine, Department of Pediatrics, Division of Pediatric Endocrinology, İstanbul, Turkey*

*[2]İstanbul University İstanbul Faculty of Medicine, Department of Medical Genetics, İstanbul, Turkey*

Multiple pituitary hormone deficiency (MPHD) due to the absence of pituitary transcription factors is associated with growth hormone (GH) deficiency in addition to at least one of the deficiencies of adrenocorticotropic hormone (ACTH), luteinizing hormone, follicle-stimulating hormone, thyroid-stimulating hormone (TSH), and/or prolactin (PRL). Its prevalence is 1/7000-1/10000. The aim of our presentation of the case with MPHD is to emphasize the clinical examination and hormonal findings, follow-up of radiological-clinical findings, and importance of early diagnosis and treatment. A 9-month-old male patient presented to outpatient clinics with the symptom of constipation for the last 1.5 months and diagnosed with hypothyroidism was referred to our clinic for further evaluation. Mother and father were consanguineous, first-degree cousins. At presentation, his weight was 5700 gram [-3.6 standard deviation score (SDS)], height 61.8 cm (-3.5 SDS), bone age compatible with 2 months of age. On physical evaluation, pale skin, edematous eye lids, coarse face, macroglossia, motor retardation, hypotonicity, abdominal distention, umbilical hernia, and hepatomegaly were noted; his cry was hoarse. The patient was pre-pubertal. Laboratory examination revealed anemia, elevated liver enzymes, hyperlipidemia, and central hypothyroidism. TSH response to thyrotropin-releasing hormone stimulation was blunted. Cortisol and PRL levels were normal, partial GH deficiency was detected. On cranial and pituitary magnetic resonance imaging examination, pituitary hyperplasia was determined. On L-thyroxin treatment, clinical findings of hypothyroidism and pituitary hyperplasia regressed. Clinical and laboratory findings suggested *PROP1* gene defect. However, no mutation was found in the *PROP1* gene. No *POU1F1*, *LHX3* and *HESX1* gene mutations were detected either. During follow-up, growth velocity decreased around 12 years of age. Repeated stimulation tests were compatible with GH deficiency and GH treatment was started. PRL and ACTH deficiencies did not develop and spontaneous puberty started at 12 years and 4 months of age. At his last visit, he was 14 years and 4 months old, his weight was 51.3 kg (-0.4 SDS), his height was 150.9 cm (-1.7 SDS) with progression of puberty. Genetic analysis should be done in patients with congenital MPHD to diagnose the hormone deficiencies and to start treatment if necessary.

**Key words:** Pituitary hyperplasia, central hypothyroidism, MPHD, genetic analysis, *PROP1*