The phenotype connected to *POU1F1* mutations is characterized by profound GH and PRL deficiencies, variable degrees of TSH deficiency, severe proportional short stature, atypical facies, and feeding difficulties in infancy. Patients harboring mutations within *PROP1* gene present with GH, PRL, and TSH deficiencies in addition to variable defects in luteinizing hormone/follicle-stimulating hormone and adrenocorticotropic hormone secretion. *PROP1* mutations are the most common known genetic cause of CPHD cases.

Here, we present 3 cases with *POU1F1* mutation and 1 case with *PROP1* mutation, who were molecularly diagnosed in Medical Genetics Department of Ege University.

The three cases (1 female, 2 males) carrying *POU1F1* mutations all had short stature. One male case with a novel mutation, p.K216T, also presented with micropenis in addition to short stature. The other mutations detected in *POU1F1* gene were S50A, R265W; S50A being novel. The case with *PROP1* mutation also had short stature and micropenis. Molecular analysis revealed a frameshift p.L102CfsX8 mutation in the *PROP1* gene. Biochemical testing showed PRL and GH deficiencies in all cases. Two cases with *POU1F1* defect and the case with *PROP1* defect also had central hypothyroidism.

It is considered that in patients with growth retardation together with combined pituitary hormone deficiency, *POU1F1* and *PROP1* gene mutations should be investigated. In this study, two novel *POU1F1* mutations were detected for the first time.

(P-53)

FGFR2 Gene Mutations in Patients with Syndromic or Isolated Craniosynostosis

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Craniosynostosis, premature closure of one or more cranial sutures, may occur in both non-syndromic and syndromic forms. Birth prevalence of craniosynostosis is 1/ 2,100–1/2,500. *FGFR2*, *FGFR3*, *FGFR1*, *TWIST1*, and *EFNB1* genes play a role in the syndromic craniosynostosis presenting with craniofacial abnormalities, including hypertelorism, proptosis, and midfacial hypoplasia. Limb, cardiac, central nervous system, and tracheal malformations have also been described. Mutations in the *FGFR2* gene located on 10q26 that encode fibroblast growth factor receptor 2 is responsible for a part of the syndromic craniosynostosis. The aim of this study was to determine *FGFR2* gene mutations in 85 craniosynostosis cases including Apert, Pfeiffer, and Crouzon syndromes, and isolated craniosynostosis patients who were referred to molecular genetics laboratory of Medical Genetics Department, Ege University between 2010 and 2016.

Sequence analysis was performed on 2 exons (exons 7-8) of the *FGFR2* gene in 85 cases referred for pre-diagnosis of craniosynostosis between 2010 and 2016. Sanger sequencing analysis method was used for sequence analysis.

Mutations were detected in twenty of the cases (23%). The frequency of *FGFR2* mutation in this study was 20% S252W and P253R (4 cases), 15% Y382C (3 cases), 10% Y308C (2 cases) and 5% A314S, A266P, P253A, W290C, W290R, S351C, S252P (1 case).

In syndromic and isolated craniosynostosis patients, the analysis of exons 7 and 8, which is one of the mutational hot spot of *FGFR2* gene, allows diagnosis in 23% of patients. It has been concluded that performing complete *FGFR2* gene analysis will provide larger numbers of molecular diagnosis.

(P-54)

The Mutation Spectrum of *DHCR7* Gene and Two Novel Mutations

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Smith-Lemli-Opitz syndrome (SLOS) is a rare autosomal recessive syndrome. It is one of the 46,XY disorders of sexual development. Molecular defects in *DHCR7* gene are responsible for this syndrome. In this study, the mutation spectrum of *DHCR7* gene in SLOS patients has been evaluated.

Thirteen patients from 11 families carrying mutations in the *DHCR7* gene were included in this study. Seven different *DHCR7*