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 PRO1 gene present with GH, PRL, and TSH deficiencies in addition to variable defects in luteinizing hormone/follicle-stimulating hormone and adrenocorticotropic hormone secretion. PRO1 mutations are the most common known genetic cause of CPHD cases.

Here, we present 3 cases with POU1F1 mutation and 1 case with PRO1 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 microepi in addition to short stature. The other mutations detected in POU1F1 gene were S50A, R265W; S50A being novel. The case with PRO1 mutation also had short stature and microepi. Molecular analysis revealed a frameshift p.L102CfsX8 mutation in the PRO1 gene. Biochemical testing showed PRL and GH deficiencies in all cases. Two cases with POU1F1 defect and the case with PRO1 defect also had central hypothyroidism.

It is considered that in patients with growth retardation together with combined pituitary hormone deficiency, POU1F1 and PRO1 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**

Emine İpek Ceylan1, Aslı Ece Solmaz2, Hüseyin Onay1, Ayça Aykut1, Asude Durmaz2, Gözde Yeşil2, Filiz Hazan2, Aslıhan Keraz1, Beyhan Tuysüz3, Meltem Cerrah Güney3

1Ege University Faculty of Medicine, Department of Medical Genetics, İzmir, Turkey
2Bezmialem Vakif University Faculty of Medicine, Department of Medical Genetics, İstanbul, Turkey
3Dr. Behçet Uz Children’s Hospital, Clinic of Medical Genetics, İzmir, Turkey
4Kayseri Training and Research Hospital, Clinic of Medical Genetics, Kayseri, Turkey
5Istanbul University Cerrahpaşa Faculty of Medicine, Department of Pediatric Genetics, İstanbul, Turkey
6Erciyes University Faculty of Medicine, Department of Medical Genetics, Kayseri, Turkey

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 (5 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**

Ersu İskık, Hüseyin Onay, Bilçâg Akgün, Tahir Atik, Ayça Aykut, Asude Durmaz, Munis Dündar, Yaşar Bekir Kurtbay, Ercan Mihçi, Ferda Özkınay

1Ege University Faculty of Medicine, Division of Pediatric Genetics, İzmir, Turkey
2Ege University Faculty of Medicine, Division of Medical Genetics, İzmir, Turkey
3Erciyes University Faculty of Medicine, Department of Genetics, Kayseri, Turkey
4Tepecik Training and Research Hospital, Genetic Diagnostic Center, İzmir, Turkey
5Akdeniz University Faculty of Medicine, Division of Pediatric Genetics, Antalya, Turkey

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