

OP-01 Primary microcephaly cases with molecular genetic basis

Ümit Taşdemir¹, Oya Demirci¹

¹ Zeynep Kamil Women and Children Diseases Training and Research Hospital

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Objective: Primary microcephaly (MCPH, for “microcephaly primary hereditary”) is a disorder of brain development that results in a head circumference more than 3 standard deviations below the mean for age and gender. Our aim is to evaluate ultrasonographic findings, clinical features and molecular analysis of two cases of primary microcephaly

Case: Two cases at 26 and 23 weeks referred to our clinic due to small BPD and HC measurements for gestational age at second trimester screening ultrasonography

Discussion: The ongoing discovery and research on MCPH genes and their animal models will increase our knowledge in this rare non-progressive neuropsychiatric disorder. MCPH genes might play an essential role during evolution, and therefore, they are suitable candidates for studying normal brain development

Conclusion: All cases of microcephaly should be referred to the medical genetics service to confirm or rule out the diagnosis and to provide genetic counseling.

Keywords: Brain development, genetic, microcephaly

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OP-02 Serum prolactin levels in women with threatened miscarriage a prospective case-control study

Emine Zeynep Yılmaz¹, Gökhan Tulunay¹

¹ Ankara Etlik Zübeyde Hanım Hospital

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Objective: The aim of this study is to compare the serum prolactin levels of women with threatened miscarriage (TM) and women with uncomplicated pregnancies.

Results: There were statistically significant differences between groups in serum prolactin levels, significantly higher in group I (47.7 ± 42.6 vs 29.64 ± 13.01 , respectively; $p=0.003$). Mean serum prolactin levels of women were increasing directly with gestational age ($p=0.005$)

	Threatened miscarriage (n:226)	Control Group (n: 215)	p
Prolactin (n/ mL)	47.7 ± 42.6	29.64 ± 13.01	0.003
TSH	1.75 ± 0.99	1.84 ± 1.04	0.356

Conclusion: Our results demonstrate that serum prolactin levels differed in women with TM in early weeks of pregnancy. Prolactin may be a future biomarker in predicting the outcome of TM.

Keywords: Biomarker, miscarriage, prolactin

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OP-03 Osteogenesis imperfecta type 3 with COL1A1 mutation prenatal diagnosis and management

Mirac Ozalp¹, Cagdas Nurettin Emeklioglu², Guldem Durak³

¹ Prof. Dr. Cemil Tascioglu City Hospital, Department of Obstetrics and Gynecology, Perinatology Clinic, Istanbul, Türkiye

² Karabuk University Training and Research Hospital, Department of Obstetrics and Gynecology, Karabuk, Türkiye

³ Prof. Dr. Cemil Tascioglu City Hospital, Department of Obstetrics and Gynecology, Istanbul, Türkiye

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Objective: Osteogenesis Imperfecta (OI) is a rare genetic disease group consisting of defects in collagen synthesis. This disease is characterized by alteration of connective tissue structure of exposed patients, usually caused by mutation of type I collagen. Altered connective tissue structure leads to low bone mineral density, defective bone structure and strength, and multiple fractures, which are usually obtained after low-impact trauma.^[1] It is a rare disease that is seen in 0.5-1 per 10000 live births in general.^[2] There are several types of OI, type I being the mildest form of OI, which is usually asymptomatic, sometimes with scoliosis due to compression fractures of the vertebrae in adults. Type II is the most severe form and is generally incompatible with life, while type III is