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OP-07 Prenatal diagnosis of Pfeiffer Syndrome case report

Aydın Öcal[1], Sinem Tekin[1]
[1] University of Health Sciences, Haseki Training and Research Hospital, Sultangazi, Istanbul, Türkiye
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Objective: Our purpose was to describe and compare the cranial and extracranial abnormalities of Pfeiffer syndrome on prenatal imaging with postmortem findings.

Case: A healty 23-year-old expectant mother, nulliparous referred to our perinatology clinic for sonographic abnormalities in 19-week gestation. The cranial examination on fetal ultrasonography; The fetal sagittal suture was narrow. Its coronal and lambdoid sutures were nearly closed (Figure 1). We recorded severe ocular proptosis and hypertelorism. The lids were everted everted which is often seen in the more severe forms of Pfeiffer syndrome. We observed pes equinovarus on feet and the broad great toe. The patient was offered the option of termination but the patient and her husband refused. The patient was admitted in 27 weeks old to the emergency department. On ultrasound examination, the fetus was mort in the uterus and taken to delivery. Postnatal fetal examination revealed cloverleaf skull, ocular proptosis, flat midface and nose, clubfeet, broad great toe (Figure 2). FGFR2 gene sequence analysis, c.1019A>G (p.Tyr340Cys) missense variant (rs1554928884 ClinVar: 449398) was detected in heterozygous form. The genetic result supports our clinical findings of Pfeiffer syndrome.

Fig 1. The shape of the skull was turricephaly suggestive of craniosynostosis and is also called cloverleaf-shaped cranium.

Fig 2. Cloverleaf skull, ocular proptosis, Clubfeet, broad great toe

Conclusion: Pfeiffer syndrome is a rare genetic disorder with a very poor prognosis because of the many complications. Prenatal diagnosis of this syndrome remains difficult and is based on fetal ultrasonography exploring the head, face and extremities, with a molecular biology analysis.

Keywords: Cloverleaf skull, pfeiffer syndrome, prenatal diagnosis, ultrasound, proptosis

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OP-08 The effect of maternal metabolic factors and lipid profile on birth weight in pregnants with gestational diabetes and normal glucose tolerance

Lutfiye Uygur[1]
[1]Health Sciences University, Zeynep Kamil Women and Children Diseases Reseach Hospital, Department of Obstetrics and Gynecology, Division of Perinatology, Istanbul, Türkiye
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Objective: Gestational diabetes is correlated with metabolic disorders like, obesity, insulin resistance, hyperlipidemia, and hypertension.[1] The physiological changes providing the accumulation of maternal serum content towards the fetus to support its growth mimick the metabolic syndrome, and they are exaggerated in women with gestational diabetes.[2] This study aimed to investigate the impact of maternal metabolic syndrome parameters and lipid profiles on intrauterine fetal development in pregnancies with gestational diabetes and with normal glucose tolerance. The second aim was to compare the metabolic profiles of pregnant women with GDM and those with normal glucose tolerance.

Methods: Pregnant women who applied for an oral glucose tolerance test were examined for metabolic syndrome between 24th-28th weeks. The group diagnosed with gestational diabetes and those with normal