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## OP-07 Prenatal diagnosis of Pfeiffer Syndrome case report

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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 healthy 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 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 turriccephaly 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 pregnant with gestational diabetes and normal glucose tolerance

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**Objective:** Gestational diabetes is correlated with metabolic disorders like, obesity, insulin resistance, hyperlipidemia, and hypertension.<sup>[1]</sup> The physiological changes providing the accumulation of maternal serum content towards the fetus to support its growth mimic the metabolic syndrome, and they are exaggerated in women with gestational diabetes.<sup>[2]</sup> 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

glucose tolerance were compared in terms of obesity, hypertension, serum lipid profile, and neonatal birth weight. Hypertriglyceridemic and normotriglyceridemic patients were compared regarding maternal metabolic syndrome criteria and neonatal birthweight.

**Results:** Diabetic pregnant had significantly higher body mass index and triglyceride levels and lower high-density lipoprotein levels than non-diabetics. The hypertension rate was also higher; however, it was not statistically significant (Table 1). Those with hypertriglyceridemia had higher body mass index, HbA1c level, and neonatal birth weight in the diabetic group (Table 2). Triglyceride level did not impact neonatal birthweight in non-diabetic patients. Obesity, high HbA1c and triglyceride levels, and low high-density lipoprotein levels were the parameters leading to fetal macrosomia in gestational diabetes.

**Table 1.** Lipid Profile HbA1c and rate of hypertensive diseases in the NGT and GDM groups.

	NGT (n=93)	GDM (n=83)	
<b>TG (mg/dl)</b>	181.1 ± 62.8 176 (80-375)	245.1 ± 90.1 226 (115-522)	< 0.001
<b>LDL (mg/dl)</b>	135.5 ± 41.8 130 (60-305)	124.3 ± 52.0 116 (33-420)	0.121
<b>Total cholesterol (mg/dl)</b>	250.4 ± 45.6 245 (155-392)	226.0 ± 47.8 226 (66-342)	0.001
<b>HDL (mg/dl)</b>	81.4 ± 21.2 78 (47-187)	59.6 ± 16.0 59 (33-107)	< 0.001
<b>HbA1c (%)</b>		5.7 ± 0.8 6 (5-10)	
<b>HDP</b>	7 (7.5%)	13 (15.7%)	0.090*

**Table 2.** Comparison of the hypertriglyceridemic and normotriglyceridemic groups among the GDM patients

	TG< 200 (n=26)	TG ≥ 200 (n=57)	p
<b>Age</b>	31.9 ± 5.6 32 (19-42)	33.4 ± 4.6 33 (23-42)	0.200
<b>BMI</b>	25.1 ± 3.2 25 (20-32)	28.4 ± 4.8 27 (20-41)	0.001
<b>Total Weight Gain</b>	12.6 ± 4 14 (6-22)	14.1 ± 5 14 (6-30)	0.194
<b>HbA1c (%)</b>	5.4 ± 0.5 5(5-6)	5.8 ± 0.9 6 (5-10)	0.013
<b>Birth weight</b>	3492 ± 368 3510 (2890-4020)	3725 ± 612 3700 (2340-5390)	0.077
<b>LGA newborn</b>	10 (38.5%)	26 (45.6%)	0.779
<b>HDP</b>	2 (7.7%)	11 (19.3%)	0.177

Independent sample test/Chi-square test

**Discussion:** The extension of the changes in maternal lipid and carbohydrate metabolism to support fetal growth differs depending on the gestational diabetes status. The studies in the literature report that despite adequate glycemic control proved by standardized measures like fasting, 1st-hour, and 2nd-hour blood glucose levels or HbA1c level, macrosomia rates are higher in pregnant women with gestational diabetes. This raises the question of whether there are other factors leading to macrosomia in insulin-resistant patients.<sup>[3]</sup>

**Conclusion:** GDM is a pathology related to several metabolic disorders, such as obesity, insulin resistance, hyperlipidemia, and hypertension. All of these disorders are components of metabolic syndrome interacting with each other, changing the intrauterine environment and leading to fetal macrosomia. The prevention of obesity in reproductive age, the prevention of excessive weight gain throughout pregnancy, and more liberal use of anti-diabetic agents to avoid the lipolytic effects of insulin resistance in gestational diabetes, instead of insisting on long-term dietary restrictions, may decrease the macrosomia risk.

**Keywords:** Gestational diabetes, insulin resistance, macrosomia, metabolic disorder

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## OP-09 Treatment modalities in twin reversed arterial perfusion

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**Objective:** This study's objective was to examine the follow-up and treatment modalities applied to four cases of twin reversed arterial perfusion (TRAP) sequences observed in monochorionic twin pregnancies.

**Methods:** Four cases diagnosed with TRAP who applied to the perinatology clinic of Necmettin Erbakan University Meram Faculty of Medicine between 2019 and 2022 were included in the study. Written informed consent was obtained from all patients to present the cases. A karyotype study was performed on all cases, and the other baby was found to be healthy. Four cases were acardiac twins. The diagnosis of TRAP was confirmed