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Olgu Sunumu / Case Report

Rare Birth Defects in Pregnancies of Women with Pregestational Diabetes: Absent Radius

Diyabetik Anne Bebeklerinde Nadir Görülen Bir Defekt: Radius Yokluğu

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ABSTRACT

Diabetes mellitus (DM) is an important disease that negatively affects fetal development and women with pregestational diabetes have an increased risk for adverse pregnancy outcomes, including a markedly increased risk for birth defects. In this report, a newborn with absent radius associated with pregestational diabetes was presented. A male newborn was born at 40th gestational week from a 20-year-old mother by cesarean section. The patient had shortness in the limbs, hypoplastic right thumb and flexion contracture at his right hand besides respiratory distress.To our knowledge, with this case report, we would like to share the second case of diabetic mother's infant with absent radius in the literature.

Keywords: Absent radius, diabetes mellitus, newborn

ÖZ

Diabetes mellitus (DM), fetal gelişimi olumsuz etkileyen önemli bir hastalıktır. Pregestasyonel diyabeti olan kadınlar doğumsal malformasyonlar için önemli ölçüde artmış bir riske sahiptir. Bu makalede, pregestasyonel diyabet ile ilişkili radius yokluğu olan bir yenidoğan sunulmuştur. 20 yaşındaki bir anneden sezaryen ile 40. gebelik haftasında doğan erkek bebek; ekstremitelerinde kısalık, sağ baş parmağında hipoplazi, solunum sıkıntısı yanında sağ elinde fleksiyon kontraktürü vardı. Bu olgu sunumu ile literatürde radius yokluğu ile ilişkili ikinci diyabetik anne bebeğini paylaşmak istiyoruz.

Anahtar kelimeler: Radius yokluğu, diyabet, yenidoğan

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INTRODUCTION

Diabetes mellitus (DM) is an important disease that negatively affects fetal development during pregnancy and causes important metabolic disorders in newborns.[1] Gestational diabetes occurs in 4-7% of pregnancies.[2] Among gestational diabetes, the incidence of infants born to diabetic mothers with gestational DM is 93% and mothers with insulin dependent pregestational DM is 7%.[3] Babies of pregestational diabetic mothers have an increased risk of congenital malformations. Birth defects are less common in babies of gestational diabetic mothers.[4]

Pregestational diabetes was found to be a statistically significant etiologic factor for birth defects (range, 2.5-80.2) and the highest probability was observed in sacral agenesis (adjusted odds ratio; 80.2) according to the latest National Birth Defects Prevention Study Group. More than 10-fold increased risk was also observed for longitudinal limb deficiency, single ventricle complex, truncus arteriosus, atrioventricular septal defect, holoprosencephaly and truncus arteriosus.[5]

Preventing hyperglycemia during pregnancy is important in preventing malformations and complications.[6] It is suggested that the frequency of congenital anomalies reflects that the mother had poorly controlled diabetes and increased HbA1c levels are effective in the first trimester of pregnancy.[7]

In this report, a newborn with absent radius associated with pregestational diabetes was presented with literature highlights. Informed consent was obtained for publication.

CASE REPORT

A male newborn was born at 40th gestational week from a 20-year-old mother by cesarean section. The newborn's 1st and 5th minute APGAR scores were 8 and 9, respectively. The patient was admitted to neonatal intensive care unit (NICU) with the diagnoses of transient tachypnea and infant of mother with Type 1 DM. On physical examination, birth weight was 3150 g (50-75 p), length was 52 cm (50-75 p) and head circumference was 35 cm (50-75 p). He had tachypnea, dyspnea and chest withdrawal. Ampicillin and gentamicin were started because neonatal sepsis could not be ruled out in the case that is followed with nasal Continous Positive Airway Pressure. Additionally, he had shortness in the limbs, hypoplastic right thumb and flexion contracture at his right hand. However, inspection of the face did not reveal any dysmorphic facial features. In his skeletal X-ray, right radius was found to be absent and hypoplastic right thumb was detected (**Figure 1**). Other systemic

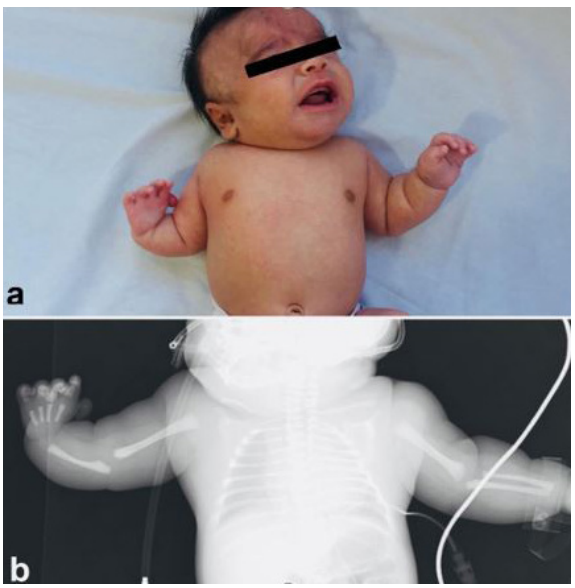

Figure 1. 1a. Infant's face with no dysmorphic features. 1b. Extremity anomalies of the patient.

findings were found to be normal. He had hypoglycemia and hypocalcemia. His laboratory investigations including complete blood count and blood biochemical tests except glucose and calcium were within normal range. The treatment was organized in accordance with the Turkish Neonatology Association protocols. Intraventricular septal hypertrophy was observed in the echocardiography of the patient. Abdominal ultrasonography (US) revealed horseshoe kidney. His respiratory distress regressed on the second day of his hospitalization. His chromosomal analysis was found to be normal and the infant was discharged on day 12.

DISCUSSION

Gestational diabetes is a problem that remains important at the present time due to increased

fetal, neonatal risks and long-term complications**.** Many factors have been accused for increasing congenital anomalies in infants of diabetic mothers including genetic causes, teratogenic agents, maternal vascular disease and metabolic effects of maternal diabetes. However, hyperinsulinemia and hyperglycemia developing on the basis of beta cell damage and fetal insulin resistance are the main effective pathogenic factors.[8] Although there is not enough information about the early pregnancy process, it has been observed in studies that excessive proinsulin occurring in the embryos of diabetic mothers causes teratogenic effect by reducing apoptosis.[3,8,9] Congenital malformations, maternal age, parity are also associated with a history of gestational diabetes and glycemic parameters.[10] The frequency of minor and major malformations was reported as 6% and 3.8%, respectively, in infants born to mothers with gestational diabetes.[6,8] No difference was detected in terms of congenital anomaly risk between diabetic pregnants, with HbA1c value below 7%, and non-diabetic pregnants. However, it has been observed that the risk of congenital malformations increases in infants born to mothers with a HbA1c value above 7%.[11] Until 2012, there is not a case report explaining absent radius as a manifestation of infants of diabetic mothers. Firstly, Gupta et al. reported a rare case of newborn, who presented with an additional feature of absent radius, which is not yet reported again in the literature[12]. According to the National Birth Defects Prevention Study Group;. pregestational diabetes was found to be strong and significant etiologic factor for birth defects. There was also an increased risk for longitudinal limb deficiency (adjusted odds ratio, 10.1; 95% confidence interval, 6.2-16.5). However, the rate for absent radius is not specified.[5]

Isolated radial ray defects affect 2:10,000 of the newborns, but the incidence of more complex upper limb deficiency is approximately 5.25:10,000.[13] Radial ray defects and associated anomalies include a large group. Majority of these defects are unilateral and bilateral defects are part of multiple anomaly syndromes.[14] In the study by Goldfarb; 67% of the patients with radial ray deficiency had medical or musculoskeletal abnormalities; only 33% had an isolated radial ray deficiency. The most common associated medical conditions with radial ray deficiency were cardiac anomalies (20%), TAR syndrome (15%), VACTERL (Vertebral, anorectal, cardiac, tracheoesophageal fistula, renal and limb) association (13%), Holt-Oram syndrome (4%), and Fanconi anemia (1%).[15] Congenital limb deficiencies are common birth anomalies associated with other anomalies or may occur isolated. A multidisciplinary assessment and management strategy tailored to a particular patient is required for best results and accurate genetic counseling.[16] It is also known that there may be speculations in the related literature about the relationship between VACTERL and maternal diabetes.[17] Our case may be evaluated as incomplete VACTERL due to cardiac, renal and limb deformities. In addition to metabolic disorders such as hypoglycemia, hypocalcemia, structural anomalies such as absent radius, hypoplastic right thumb, intraventricular septal hypertrophy, and horseshoe kidney were detected in our diabetic mother's infant.

To our knowledge, with this case report, we would like to report the first case of diabetic mother's infant associated with absent radius and horseshoe kidney in the literature.

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