A Prenatally Diagnosed Non-Syndromıc AnencephalıcConcordant Twin Pregnancy

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ÖZET

(Prenatal tanı konulan Non sendromik Anensefalik Korkodon İkiz Gebelik).

33 yaşında gravida 5 para 3 olan olgu Kahramanmaraş Sütçü İmam Üniversitesi Tıp Fakültesi, Obstetri Kliniği'ne 28. gebelik haftasında monokoryonik monoamniotik gebelik ile ve her iki fetüste anensefali ön tanısı ile refere edildi. Prenatal ultrasonografide her iki fetüste anensefali gözlendi. Vajinal doğumu takiben dismorfolojist tarafından yapılan detaylı değerlendirme sonucunda prenatal bulgu doğrulandı. Bu vakada; prenatal teşhis edilen non-sendromik anensefalik konkordant aynı cins, kız, ikiz gebelik sunulmuştur.

Anahtar kelimeler: *Nöral tüp defekti, anensefali, spina bifida, ikiz gebelik.*

SUMMARY

(A Prenatally Diagnosed Non-Syndromıc Anencephalıc Concordant Twin Pregnancy)

A 33-year-old pregnant woman gravida 5 para 3 was referred to the obstetric clinic of Kahramanmaras Sutcu Imam University presenting with monochorionic monoaniotic twin pregnancy and anencephaly in both fetuses at 28 weeks of gestation. Prenatal ultrasonographic examination revealed concordant anencephaly. Meticulous neonatal examination by a dysmorphologist was performed following vaginal delivery and confirmed the prenatal findings. In this case; prenatally diagnosed non-syndromic anencephalic concordant like-sexed, female, twin pregnancy is presented.in the light of literature.

Key-words: *Neural tube defects, anencephaly, spina bifida, twin pregnancy.*

INTRODUCTION

Neural tube defect (NTD) is a common disorder and its incidence has been estimated as nearly 1 per 1000 live-births. The distribution of neural tube defects show considerable geographical and ethnic variation. Insults occuring before the end of the 6th week result in anencephaly (failure of closure of the rostral neuropore) or neural tube defect (failure of closure of the caudal neuropore) (1). There are different types of neural tube defects which can be seperated into two main groups: open and closed. Most of the reported studies has concentrated on the open types as the clinical severity and high frequency. The most frequent types of open NTDs are anencephaly and spina bifida. Anencephaly, the most severe form of NTDs, implies an absence or deficiency of a major portion of the cranial vault with nearly normally formed facial bones and base of the skull. Ossification of the skull is normally present after 12 weeks' gestation. However, the frontal bone is always absent and the brain tissue is always abnormal (2). Anencephaly seems to be occured more often among females, whereas spina bifida rates have shown only a slight female predominance. Although many studies have clearly presented the high recurrence risk of NTDs in family members of affected persons, the majority of the cases (over 95 %) have been occured to women without a prior family history of this disorder. Recurrence risk is nearly 3 % in first degree relatives (3). Most of the cases do not present with any type of Mendelian inheritance patterns and NTD is a multifactorial disorder. This occurs when there is a genetic predisposition to the malformation which is triggered by an environmental risk factor.

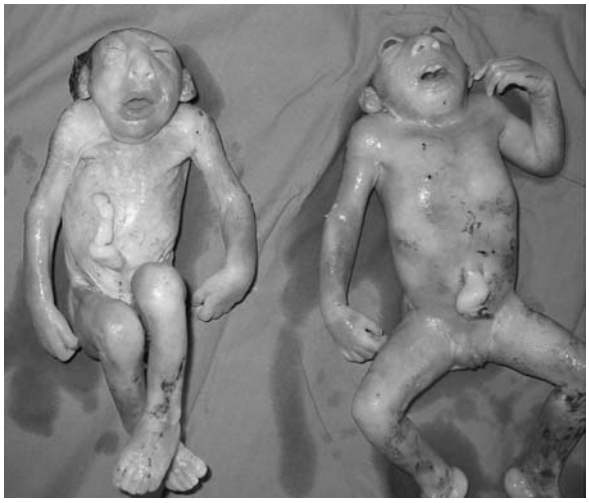
There are other risk factors for NTDs out of genetic factors including; socioeconomical status (4), the lead in drinking water (5), influenza (6), maternal heat exposure (7), parental occupation (8), maternal obesity (9,

10), maternal nutritional status (11), maternal use of some drugs especially valproic acid and carbamazepine and maternal hyperthermia (1). One nutritional factor; folic acid, has also been shown to play a very powerful role in the occurrence of neural tube defects (12, 13). Twining is described as another risk factor for neural tube defects and anencephaly (14, 15). Nearly two fold increase of the frequency of NTDs were detected in twin pregnancies. Both concordant and discordant cases were published, while the number of concordancy reported so rarely. We present a case of monoamniotic twins concordant for anencephaly.

CASE REPORT

A 33-year-old pregnant woman gravida 5 para 3 was referred to the obstetric clinic of Kahramanmaras Sutcu Imam University presenting with monochorionic monoaniotic twin pregnancy and anencephaly in both fetuses at 28 weeks of gestation. Chorionicity of the pregnancy was told the patient at 8 weeks of gestation. Her medical history was unremarkable. Prenatal ultrasonographic examination revealed concordant anencephaly . Detailed evaluation of fetal anatomy revealed no other abnormality. Because of the lethality of the anomaly, parents opted for pregnancy termination. Meticulous neonatal examination by a dysmorphologist was performed following vaginal delivery and confirmed the prenatal findings **(Figure 1)**.

Figure 1: Postmortem view of the both children presenting with anencephaly.



Anencephaly is the single most common prenatally detected neural tube defect (4). The ultrasound diagnosis of anencephaly is made on the basis of the absence of the upper portion of the cranial vault. The major consideration in the differantial diagnosis is to distinguish anencephaly from the presence of amniotic bands. It is important to note that the cranial defect associated with anencephaly is always symmetric. However, with amniotic bands, there should be evidence of other defects, such as limb or digital amputations, asymmetric ventral-wall defects, or spinal defect. Other conditions in the differential diagnosis include inenncephaly which does not involve the forebrain. It has been demonstrated that the incidence of congenital anomalies was found to be 2.5 time more common in monozygotic twins than in dizygotic twins or singletons (15). In monozygotic twins, both fetuses have the same genetic background and predisposing factors. There are some case reports in the literature presenting with NTDs in monozygotic twins (16-19) such as anencephaly.

In one series of 1424 twin pairs, 445 pairs were monozygotic, 26 of which (%6) had congenital malformations. Even among monozygotic twin pairs with malformations, however, the majority of fetuses will be discordant for the abnormality, with only 6 of the 26 twin pairs (%23) in Cameron et al.'s study showing concordance for the abnormality (20).

When both fetuses in a twin pregnancy are concordant for malformations, subsequent management of that pregnancy is straightforward such as termination of pregnancy. So, concordancy is a less severe clinical problem during prognostic decision of the pregnancy than that of discordancy in which pregnancy management becomes considerably more complex when one twin has a congenital malformation but the co-twin is normal.

Kallen et al., (21) reported a number of cases with NTDs in twin pregnancies. The author concluded that twins concordant for anencephaly or encephalocele were mainly found when the defect occured as a part of a syndrome, and only in like-sexed pairs, however our case are non-syndromic and like-sexed. Anencephaly is more frequent among females and so far, nearly all of the concordant monozygotic twins with anencephaly reported were females as in our

case. Although there are a number of risk factors for NTDs out of genetic predisposition, patients history did not reveale any obvious detrimental factor leading to concordant anencephaly in monozygotic twins. In our case the only risk factor for NTD was monozygotic twining.

In conclusion, we present this case to stress the rarity of this non-syndromic concordant anencephaly pregnancy in monozygotic twin. Further, determining chronicity is vital in twin pregnancy and an increased anomaly incidence in monozygotic pregnancies should be kept in mind.

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