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Concordant fatal congenital anomaly in twin pregnancy: a case report and review of the literature

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Abstract

Background When a pregnant mother finds out she has a fetus with a congenital defect, the parents feel profound worry, anxiety, and melancholy. Anomalies can happen in singleton or twin pregnancies, though they are more common in twin pregnancies. In twins, several congenital defects are typically discordant.

Case summary We present a rare case of concordant fatal anomaly in twin pregnancy in a 22-year-old African patient primigravida mother from Western Ethiopia who presented for routine antenatal care. An obstetric ultrasound scan showed anencephaly, meningomyelocele, and severe ventriculomegaly. After receiving the counseling, the patient was admitted to the ward, and the pregnancy was terminated with the medical option. Following a successful in-patient stay, she was given folic acid supplements and instructed to get preconception counseling before getting pregnant again.

Conclusion The case demonstrates the importance of early obstetric ultrasound examination and detailed anatomic scanning, in twin pregnancies in particular. This case also calls for routine preconceptional care.

Keywords Twin pregnancy, Concordant, Anencephaly, Meningomyelocele, Ventriculomegaly, Gastroschisis

Introduction

A "congenital anomaly" is defined as any abnormal deviation from the expected structure, form, or function. "Malformations" are morphological abnormalities of organs or regions of the body resulting from an intrinsically abnormal developmental process, whereas "disruptions" are defects from interference with an initially normal developmental process [1, 2].

Congenital anomalies present in twins also include any anomaly that may occur in singletons, including primary structural malformations, chromosomal defects, and genetic syndromes [1, 2]. The anomalies may involve one or both twins [2, 3]. The former is called discordant, while the latter is termed concordant [1–3]. Due to the anomaly's multifactorial inheritance pattern, which is influenced by both genetic and environmental factors, twins are usually discordant for this anomaly, with only one co-twin affected [1, 3].

However, some research has shown that identical twins have concordant anomalies such as neural tube defects [3, 4]. Here we present a rare case of concordant congenital anomaly in twin pregnancy.

Case presentation

This 22-year-old African patient primigravida from western Ethiopia came to Wallaga University Referral Hospital for her second appointment as part of her

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routine prenatal care schedule. She said she had been amenorrheic for the past 4 months, but she could not recall the last time she had had a regular menstrual cycle. She received folic acid 5 mg (orally daily and iron sulphate 325 mg orally three times daily for 3 months, and two doses of tetanus–diphtheria vaccine during her prenatal care. However, she had no preconceptional care.

In the course of the index pregnancy, she had never experienced headaches, vaginal bleeding, blurred vision, or epigastric pain. In addition, she had no history of smoking tobacco, chewing khat, drinking alcohol, or using other forms of medication. She had never had bronchial asthma, hypertension, diabetes mellitus, or heart disease.

There was no history of twin pregnancies in her family. This patient was diagnosed with hyperemesis gravidarum and hospitalized to the gynecology ward a month prior to the current presentation. She was released from the hospital after 2 days, having improved. It proved that the pregnancy was twins at 11 weeks appropriate for gestational age (AGA). However, no fetal anomaly was detected.

On examination, she was healthy-looking. Her vital signs were blood pressure (BP) = 120/70 mmHg, pulse rate (PR) = 84 beats per minute, respiratory rate (RR) = 20 breaths per minute, and a temperature of 37.6 °C. She had slightly pale conjunctiva. An abdominal exam showed a 20-week-sized gravid uterus. The lymph glandular system, respiratory system, cardiovascular system, and genitalia were normal. On neurologic examination, reflexes were intact, and meningeal signs were negative.

Urinalysis; complete blood count; random blood sugar (RBS); serology for syphilis, hepatitis, and human immunodeficiency virus (HIV); obstetrics ultrasound; and blood group were done. Obstetrics ultrasound showed a twin intrauterine pregnancy with an anencephalic twin A and severe ventriculomegaly and lumbar myelo-meningocele in twin B (Table 1).

With the final diagnosis of a second-trimester twin pregnancy with a concordant fatal anomaly, the patient was admitted to the gynecology ward. In the ward, the patient was given mifepristone 200 mg orally. After 24 hours, 400 μg of misoprostol was inserted vaginally. A total of 8 hours later, she expelled twin A, weighing 120 g of anencephalic abortus with cervical, thoracic, and lumbar vertebral defect, and twin B, weighing 150 g of hydrocephalic abortus with a thoracic and lumbar vertebral defect, and protrusion of the intestine via the right side para umbilical area without the covering membrane (Fig. 1A, B). The placenta was monochorionic.

Table 1 Laboratory investigation results of a 22-year-old mother with concordant fatal anomaly in a twin pregnancy at Wallaga University Referral Hospital, Western Ethiopia, 2023

Lists of investigation	Result
WBC	6300/μl
Hemoglobin	9.8 g/dl
Platelet	370,000/μl
Urinalysis	Glucose +1
VDRL	Non-reactive
HBsAG	Non-reactive
Serology for HIV	Non-reactive
Random blood sugar	92 mg/dl
Blood group and Rh	O-positive
Obstetrics ultrasound	Twin A • AGA: 18 weeks • Non-visualized calivarium above the level of orbit
	Twin B • AGA: 18 weeks + 3 days • Dilated ventricles (each 18 mm) • Lemon-shaped calvarium • Defect of bonny spine at lumbar region with multisepated cystic swelling

HBsAG, Hepatitis B surface antigen; Rh, Rhesus factor

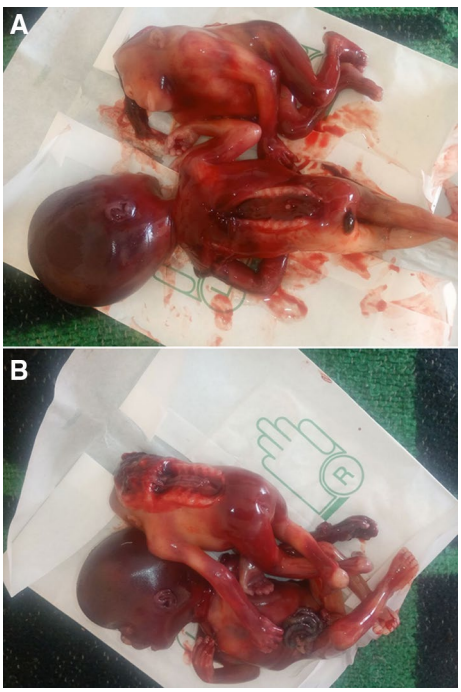

Fig. 1 **A** Twin abortuses with neural tube defects at Wallaga University Referral Hospital, Western Ethiopia, 2023. **B** Twin abortuses with neural tube defects and gastroschisis at Wallaga University Referral Hospital, Western Ethiopia, 2023

Discussion

When compared with singleton pregnancies, the rates of congenital anomalies are higher with multiple pregnancies [5, 6]. In fetuses in multiple gestations, these anatomic abnormalities are more commonly linked to monozygotic (MZ) twining than dizygotic (DZ) twining [2, 7].

Anomalies may affect all organ systems, but the commonest involve cardiovascular and central nervous systems, followed by ophthalmic and gastrointestinal abnormalities [1, 4, 7, 8]. The concordance rate of major congenital malformations is around 20% for monozygotic twins, with most dizygotic twin pairs being discordant [1]. Only in certain organ systems do monozygotic twins exhibit higher concordance rates than DZ twins [4, 7]. Our case is MZ twins. In both twins, the neural nervous system was affected. The twin pairs had neural tube defects. One twin had gastroschisis.

Anomalies in singleton and twin pregnancies are associated with maternal exposure to various factors such as diazepam use, cigarette smoking, maternal obesity, and nutritional deficiencies [9]. Our case was complicated by hyperemesis gravidarum. Because of repeated vomiting, it could result in nutritional deficiency, which could in turn result in neural tube defects and other anomalies.

There is limited published evidence about screening for structural abnormalities in twin or higher order pregnancies [10]. Careful sonographic surveys of fetal anatomy are indicated in multifetal pregnancies because the risk for congenital anomalies is increased [11]. A complete fetal anatomic survey is therefore recommended for all twin gestations at 18–22 weeks' gestational age [12]. The accuracy of ultrasonography for detecting congenital fetal anomalies in multiple gestations has not been adequately studied in large series [11].

Following diagnosis of an anomaly affecting only one fetus, practitioners may face the dilemma of expectant management versus selective termination. If the option of selective fetocide is considered, the main variable determining the technique to achieve this aim is chorionicity [13–15]. In a dichorionic pregnancy, passage of fetocidal agents from one twin into the circulation of the co-twin is unlikely due to the lack of placental anastomoses [13, 14]. When monochorionic (MC) twins are complicated with discordant fetal anomalies, the management scheme will be much more complex [13, 14]. In this case, selective termination needs to be performed by ensuring complete and permanent occlusion of both the arterial and venous flows in the umbilical cord of the affected twin. Bipolar cord coagulation under ultrasound guidance is associated with approximately 70–80% survival rates [14, 15]. However, management of concordant fatal anomaly in twin pregnancy is not controversial [3]. In our case, both twins had fatal congenital anomalies, which required immediate termination of the pregnancy using misoprostol.

Conclusion

The case demonstrates the importance of early obstetric ultrasound examination and detailed anatomic scanning, in twin pregnancies in particular. This case also calls for routine preconceptional care.

Abbreviations

DZ	Dizygotic
HBsAg	Hepatitis B surface antigen
HIV	Human immunodeficiency virus
MZ	Monozygotic
Rh	Rhesus factor
VDRL	Venereal disease research laboratory
WBC	White blood count

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Author contributions

All authors made a significant contribution to the work reported, whether that was in the conception, study design, execution, acquisition of data, analysis, and interpretation, or in all these areas; they took part in drafting, revising, or critically reviewing the article; gave final approval of the version to be published; have agreed on the journal to which the article has been submitted, and agree to be accountable for all aspects of the work.

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Availability of data and materials

The datasets used during the current study are available from the corresponding author on reasonable request.

Declarations

Ethics approval and consent to participate

Ethical clearance was obtained from the Research Ethics Review Committee of Wallaga University Referral Hospital. The study protocol is performed per the relevant guidelines.

Consent for publication

Written informed consent was obtained from the patient for publication of this case report and any accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal.

Competing interests

There are no competing interests.

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References

1. Weber MA, Sebire NJ. Genetics and developmental pathology of twinning. Seminars Fetal Neonatal Med. 2010; 15(6): 313–318). https://www.sciencedirect.com/science/article/pii/S1744165X10000466.
2. Cunningham F, Leveno KJ, Dashe JS, Hoffman BL, Spong CY, Casey BM. eds. Multifetal Pregnancy. *Williams Obstetrics, 26e*. McGraw Hill; 2022. Accessed February 01, 2024. https://accessmedicine-mhmedical-com-443.webvpn.sysu.edu.cn/content.aspx?bookid=2977&sectionid=263825445.
3. Momo RJ, Sama JD, Meka E, Temgoua MN, Foumane P. Challenge in the management of twin pregnancy with anencephaly of one fetus in a low-income country: a case presentation. J Gynecol Obstet. 2019;7(3):81–4.
4. Jung YM, Lee SM, Oh S, Lyoo SH, Park CW, Lee SD, Park JS, Jun JK. The concordance rate of non-chromosomal congenital malformations in twins based on zygosity: a retrospective cohort study. BJOG. 2021;128(5):857–64. https://doi.org/10.1111/1471-0528.16463.
5. Ogochukwu, Ugwu Rosemary and Eneh Augusta Unnoma. "Concordant congenital abnormalities in twins: report of five cases"; 2011. https://www.semanticscholar.org/paper/Concordant-congenital-abnormalities-in-twins%3A-of-Ogochukwu-Unnoma/0537e055cc465e842e15354b53e03b8909f272f2.
6. Piro E, Schierz IAM, Serra G, *et al*. Growth patterns and associated risk factors of congenital malformations in twins. Ital J Pediatr. 2020;46:73. https://doi.org/10.1186/s13052-020-00838-z.
7. Unal ER, Newman RB. Multiple Gestations. Gabbe's Obstetrics: Normal and Problem Pregnancies, 39, 751–783.e4. https://www.clinicalkey.com.
8. Tsoraides SS, Pacheco PE, Pearl RH. Concordant VACTERL anomalies in identical twins. Clin Surg. 2016;1:1145.
9. Singh S, Kayastha A, Thapa A, Thapa B, Dahal S. Omphalocele, exstrophy of cloaca, imperforate anus, and spinal defects complex: a case report. JNMA J Nepal Med Assoc. 2023;61(260):375–8. https://doi.org/10.31729/jnma.8048.
10. Bricker L. Multiple pregnancy: diagnosis and screening. In: Kumar B, Alfirevic Z, eds. Fetal Medicine. Royal College of Obstetricians and Gynaecologists Advanced Skills. Cambridge University Press; 2016:302–310. https://www.cambridge.org/core/books.
11. Malone FD, Dalton ME. Multiple Gestation: Clinical Characteristics and Management. creasy and Resnik's maternal fetal medicine, 8e: 654–678.
12. Frates MC. Multifetal pregnancy. Diagnostic ultrasound, 5e, 1115–1132. https://dokumen.pub/diagnostic-ultrasound-5nbsped.html.
13. Wataganara T, Nawapun K, Phithakwatchara N. Fetal anomalies in twin pregnancies. Donald School J Ultrasound Obstet Gynecol. 2016;10(3):308–12.
14. Rustico MA, Baietti MG, Coviello D, Orlandi E, Nicolini U. Managing twins discordant for fetal anomaly. Prenat Diagn. 2005;25(9):766–71. https://doi.org/10.1002/pd.1260.
15. Langedock A, Lewi L. Twin pregnancies discordant for fetal anomaly. In: Khalil A, Lewi L, Lopriore E, editors. Twin and higher-order pregnancies. Cham: Springer; 2021. https://doi.org/10.1007/978-3-030-47652-6_10.

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