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Ultrasound Characteristics and Management of Fetal and Neonatal Trisomy 18

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ABSTRACT

Introduction: Trisomy 18 is a severe chromosomal disorder associated with multiple congenital anomalies and high mortality. Prenatal detection relies on maternal risk assessment, biochemical screening, and fetal ultrasonography, while definitive diagnosis requires cytogenetic confirmation. Early diagnosis is essential for prognosis assessment, pregnancy management, and parental counseling.

Objectives: To analyze prenatal screening and diagnostic modalities for trisomy 18, identify the most frequent morphological abnormalities and assess the relationship between timing of diagnosis and severity of malformations and finally, to explore the implications for obstetric and neonatal management and parental counseling.

Methods: A retrospective descriptive study was conducted on five pregnancies affected by trisomy 18 taken from January 2015 to December 2025. Data collected included maternal and obstetric characteristics, prenatal screening and diagnostic methods, gestational age at diagnosis, identified fetal abnormalities, and management strategies. Obstetric and neonatal outcomes were analyzed descriptively without comparative statistical testing.

Results: Maternal age ranged from 31 to 37 years, with most patients 35 years. Prenatal screening using NIPT and ultrasonography identified recurrent abnormalities, including choroid plexus cysts, cardiac defects, single umbilical artery, limb anomalies, and growth restriction. Cytogenetic testing confirmed all cases. Early diagnosis allowed pregnancy termination, whereas later diagnosis resulted in continuation of pregnancy and poor neonatal outcomes.

Conclusion: Early prenatal diagnosis of trisomy 18 is crucial for assessing prognosis and optimizing pregnancy management. Delayed diagnosis is often associated with more severe fetal and neonatal outcomes. These findings emphasize the importance of prenatal screening and multidisciplinary care.

Keywords: Trisomy 18, prenatal diagnosis, morphology ultrasound, neonatal prognosis.

INTRODUCTION

Trisomy 18, also known as Edwards syndrome, is an autosomal aneuploidy characterized by the presence of an additional chromosome 18. It is the second most common viable autosomal trisomy after trisomy 21 and is associated with severe multisystem involvement and high morbidity and mortality rates [1,2]. The most common cytogenetic

form is complete free trisomy 18, which is usually caused by maternal meiotic nondisjunction. Less common forms include mosaic trisomy 18 and translocation trisomy [1,3].

Clinically, trisomy 18 is associated with numerous major congenital malformations, particularly involving the cardiovascular, neurological, and musculoskeletal systems, as well as nearly constant intrauterine growth restriction. The prognosis is poor, with a high rate of fetal loss and limited survival among affected neonates [1,4]. The incidence at birth is estimated to range from 1 in 5,000 to 1 in 8,000 live births; however, this likely underestimates the true incidence because of the high rate of spontaneous fetal loss. Advanced maternal age is the principal identified risk factor [4-8]. Phenotypic severity is correlated with the extent of chromosomal imbalance, with mosaic forms sometimes presenting a partially attenuated phenotype [9,10].

Trisomy 18 is associated with a broad spectrum of fetal morphological abnormalities detectable on prenatal ultrasonography, often as early as the second trimester, reflecting multisystem impairment of fetal development [1,11]. Congenital heart defects are the most common abnormalities, predominantly septal defects, particularly ventricular septal defects [1,12-14]. Gastrointestinal anomalies are also frequent and include omphalocele and esophageal atresia, which is often suspected based on indirect signs such as polyhydramnios and the absence of a gastric bubble [15,16]. Neurological abnormalities mainly include ventriculomegaly, choroid plexus cysts, posterior fossa abnormalities, and agenesis of the corpus callosum, reflecting early disruption of brain development [17,18]. Urogenital anomalies are common and are primarily represented by hydronephrosis, dysplastic kidneys, and, less frequently, renal agenesis; cryptorchidism may also be observed in male fetuses [19-21]. Musculoskeletal abnormalities are particularly suggestive of trisomy 18 and include overlapping fingers, persistent flexion of the hands, and rocker-bottom feet, all of which constitute characteristic ultrasonographic markers [1,11,21]. Craniofacial abnormalities mainly include micrognathia and low-set or dysplastic ears, while cleft lip and palate are less frequent findings [14,21]. Finally, symmetric intrauterine growth restriction is almost constant and may represent the earliest presenting sign, often associated with abnormalities of the amniotic fluid and fetal adnexa, such as a single umbilical artery or a dysmature placenta [4,11,14].

Prenatal screening primarily relies on first-trimester combined screening, which includes increased nuchal translucency and other ultrasonographic findings, together with decreased maternal serum markers, notably pregnancy-associated plasma protein A (PAPP-A) and free β-human chorionic gonadotropin (β-hCG). Non-invasive prenatal testing (NIPT), based on the analysis of cell-free fetal DNA in maternal blood, has further improved screening performance [22,23]. Prenatal diagnostic confirmation requires invasive testing. Chorionic villus sampling, performed between 11 and 13 weeks of gestation, and amniocentesis, usually performed after 15 weeks of gestation, allow early diagnosis through conventional karyotyping or rapid molecular techniques such as quantitative fluorescent polymerase chain reaction (QF-PCR) and fluorescence in situ hybridization (FISH), although confined placental mosaicism remains a recognized limitation [1]. In some cases, diagnosis is established postnatally based on a suggestive polymalformative clinical presentation and subsequently confirmed by cytogenetic analysis of peripheral blood samples [24].

The prognosis of trisomy 18 remains extremely poor because of its severe multisystem involvement. Fewer than 10% of affected children survive beyond the first year of life, with cardiorespiratory and infectious complications representing the leading causes of death [25].

Management is complex, as no curative treatment currently exists, and therefore requires an individualized, multidisciplinary approach. During the prenatal period, management includes specialized obstetric follow-up and anticipation of potential complications. When the diagnosis is established antenatally, medical termination of pregnancy may be considered following multidisciplinary evaluation and in accordance with parental informed consent [26,27]. After birth, care is primarily supportive and symptom-directed, with ethical consideration regarding the appropriate intensity of treatment, ranging from palliative care focused on comfort to selected intensive interventions in specific circumstances. Psychological support for families is essential throughout all stages of care.

MATERIALS AND METHODS

Study Design and Setting

This was a retrospective, descriptive, observational, single-center study conducted at Hôtel-Dieu de France Hospital (HDF), involving both the Department of Obstetrics and Gynecology and the Neonatology Department. The study covered a ten-year period, from January 2015 to December 2025.

The objective of the study was to describe cases of trisomy 18 managed at our institution. Given the rarity of this condition, a descriptive approach was adopted without comparative statistical analysis.

Eligible participants included pregnant women carrying fetuses diagnosed with trisomy 18, as well as fetuses and neonates affected by this condition.

Data were collected from hospital medical records, ultrasound reports, and available biochemical and genetic test results. Prenatal screening modalities were reviewed for each case, including maternal serum marker screening combined with ultrasonography and analysis of cell-free fetal DNA in maternal blood through non-invasive prenatal testing (NIPT). Diagnostic procedures were also evaluated for each case and included chorionic villus sampling, amniocentesis, and postmortem examination when a strong suspicion of trisomy 18 was present.

Neonatal prognosis and management were assessed throughout the study period, including both medical and surgical aspects of care.

The study was conducted in accordance with the principles and regulations of the institutional ethics committee and was approved under ethics committee reference number CEHDF 2857.

Study Variables

The variables extracted from medical records were categorized into five main groups:

I. Maternal Data

- Maternal age
- Parity
- Medical and surgical history
- Obstetric history
- Consanguinity

- Maternal medications during pregnancy
- Gestational age at delivery
- Mode of delivery
- Pregnancy-related complications

II. Fetal Data

- Fetal sex
- Estimated fetal weight (grams) at diagnosis
- Fetal and adnexal ultrasonographic markers and malformations
- First-trimester maternal serum markers (PAPP-A and β-hCG)
- Second-trimester maternal serum markers (AFP, unconjugated estriol [uE3], inhibin A, and β-hCG)
- Cell-free fetal DNA testing (NIPT)
- Diagnostic procedures (amniocentesis and chorionic villus sampling)
- Gestational age at detection of fetal abnormalities

III. Neonatal Data

- Birth weight (grams)
- Birth length (centimeters)
- Head circumference (centimeters)
- Apgar score at 1 and 5 minutes
- Congenital malformations identified at birth

IV. Fetal and Neonatal Outcomes

V. Maternal-Fetal and Neonatal Management

Statistical Analysis

Because of the rarity of trisomy 18 cases and the limited sample size, no inferential statistical analysis was performed. Data were analyzed exclusively using descriptive methods. Qualitative and quantitative variables were summarized through narrative descriptions and presented in summary tables when appropriate. No statistical comparisons or association analyses were conducted.

This descriptive approach was considered the most appropriate method for providing a detailed and accurate presentation of the clinical, ultrasonographic, diagnostic, management, and outcome characteristics of the cases included in the study.

RESULTS

Data from five cases were extracted from the medical records of Hôtel-Dieu de France Hospital (HDF). Each case presented distinct clinical features, screening findings, and diagnostic pathways.

Case 1

The patient was a 35-year-old multiparous woman (G3P2A0) with a history of one previous cesarean section. She had no significant medical or surgical history, no consanguinity, and was not receiving any chronic medication. Her partner was 37 years old and had no known medical or surgical history.

The pregnancy was regularly monitored. Intrauterine growth restriction (IUGR) was identified at 17 weeks of gestation. At 16 weeks, second-trimester maternal serum screening (triple test) revealed a high risk of trisomy 18, estimated at 1 in 50.

A detailed fetal morphological ultrasound performed at 19 weeks of gestation demonstrated a nuchal translucency measurement of 1.9 mm, fetal biometric parameters approximately one week below the expected gestational age, bilateral choroid plexus cysts, a single umbilical artery, a ventricular septal defect, and overlapping fingers. Amniocentesis was subsequently performed to confirm the diagnosis.

The pregnancy progressed until 26 weeks of gestation, with intrauterine fetal demise occurring at 24 weeks. Vaginal delivery was performed without complications. The fetus was female and stillborn, with a birth weight of 450 g. Placental histopathological examination suggested chorioamnionitis. No fetal autopsy was performed.

Case 2

The patient was a 32-year-old multiparous woman (G3P2A0) with a history of two previous cesarean deliveries. She had no known medical or surgical history and was not receiving any chronic treatment. There was no history of parental consanguinity. Her partner was 36 years old and had no significant medical or surgical history.

First-trimester screening showed a normal nuchal translucency measurement, while second-trimester maternal serum screening (triple test) indicated a low risk of trisomy 18, estimated at 1 in 1,743.

However, a detailed fetal morphological ultrasound performed at 18 weeks of gestation revealed several abnormalities, including a unilateral choroid plexus cyst, a single umbilical artery, and overlapping fingers. In addition, a suspected vascular anomaly involving the great vessels was noted, although the precise anatomical defect could not be determined.

Cytogenetic analysis of amniotic fluid obtained by amniocentesis confirmed the diagnosis of trisomy 18.

Following parental counseling and approval by the institutional ethics committee, medical termination of pregnancy was performed at 22 weeks of gestation by repeat cesarean section because of the patient's obstetric history. The procedure was completed without maternal complications.

The female fetus died immediately after delivery.

Case 3

The patient was a 31-year-old woman (G2P1A0) with a previous cesarean section. She had no known medical or surgical history and was not receiving any regular treatment. There was no parental consanguinity. Her partner also had no significant medical or surgical history.

The pregnancy was monitored from the first trimester. Ultrasound examination demonstrated increased nuchal translucency measuring 3.7 mm. Crown-rump length was measured at 45.7 mm. No additional morphological abnormalities were identified at that stage.

Non-invasive prenatal testing (NIPT) showed a high suspicion of trisomy 18. To confirm the diagnosis, chorionic villus sampling was performed at 11 weeks and 5 days of gestation. Cytogenetic analysis confirmed trisomy 18.

Following disclosure of the diagnosis and comprehensive parental counseling, medical termination of pregnancy was requested. Uterine curettage was performed at 13 weeks of gestation at the parents' request. No pathological examination of the products of conception was performed. The procedure was uncomplicated.

Case 4

The patient was a 37-year-old primigravida (G1P0A0) with no significant medical or surgical history and no chronic medication use. Her partner had no relevant medical or surgical history. There was no parental consanguinity.

Because of advanced maternal age, NIPT was performed early in pregnancy at 10 weeks and 4 days of gestation. The test was positive for trisomy 18, prompting further diagnostic investigations.

A morphological ultrasound examination performed at 13 weeks and 1 day of gestation showed normal nuchal translucency and crown-rump length measurements for gestational age but identified a single umbilical artery.

To confirm the diagnosis suggested by NIPT and ultrasound findings, amniocentesis was performed at 15 weeks and 3 days of gestation. Cytogenetic analysis of the amniotic fluid confirmed trisomy 18.

After confirmation of the diagnosis and detailed parental counseling regarding prognosis and management options, medical termination of pregnancy was requested. Termination was initiated at 15 weeks of gestation, followed by vaginal delivery at 16 weeks. No maternal complications occurred.

Case 5

The patient was a 33-year-old woman (G5P4A1) with a history of asthma and previous gestational hypertension. During pregnancy, she was treated with salbutamol as needed. No other medical or surgical history was reported. Her partner had no known medical history and was not receiving any treatment. The parents were consanguineous, although the degree of consanguinity was not specified.

The pregnancy was not monitored until the sixth month of gestation, when multiple fetal abnormalities were first identified on ultrasound examination.

Neurological abnormalities included a choroid plexus cyst. Cardiovascular abnormalities consisted of a large ventricular septal defect associated with a major vascular anomaly. Thoracic abnormalities included pneumomediastinum and type III esophageal atresia. Musculoskeletal abnormalities included overlapping fingers, rocker-bottom feet, a sacral malformation, and congenital hip dislocation. Polyhydramnios was also present.

No prenatal cytogenetic testing was performed, and the diagnosis was therefore not confirmed before birth.

Delivery occurred at 36 weeks and 3 days of gestation. The female neonate was born alive with a birth weight of 1,275 g, a length of 38 cm, and a head circumference of 30 cm, consistent with severe intrauterine growth restriction.

Postnatal clinical examination confirmed several morphological abnormalities, including persistent wrist flexion deformities and marked limb hypertonia. Postnatal echocardiography further characterized the cardiac defects, revealing a large ventricular septal defect, a dysplastic atrioventricular valve, type IV truncus arteriosus, and a large patent ductus arteriosus.

Neonatal karyotyping confirmed trisomy 18.

The infant was admitted to the neonatal intensive care unit and intubated immediately after birth because of respiratory distress. No corrective surgical intervention was undertaken. Clinical evolution was unfavorable, and death occurred on the seventeenth day of life.

DISCUSSION

The analysis of this series of five cases highlights several key aspects regarding the screening, diagnosis, and management of pregnancies complicated by a severe chromosomal abnormality. These observations illustrate the contribution of different prenatal screening tools, the limitations of early ultrasonography, and the impact of diagnostic timing on clinical presentation and postnatal outcomes.

Advanced maternal age emerged as a recurrent factor in this series. Several patients were aged 35 years or older, which led, in some cases, to the early use of specific screening methods such as non-invasive prenatal testing (NIPT). This approach enabled suspicion of trisomy 18 during the first trimester, before the development of major structural abnormalities. These findings emphasize the importance of increased vigilance in pregnancies involving advanced maternal age while also demonstrating that maternal age alone does not predict the severity or diversity of fetal abnormalities.

Morphological ultrasound represented a central component of the diagnostic pathway. Across all cases, ultrasonography identified suggestive abnormalities, although their nature varied according to gestational age. The most frequent findings included choroid plexus cysts, congenital cardiac defects, particularly ventricular septal defects and great vessel anomalies, as well as a single umbilical artery. Limb abnormalities, including overlapping fingers and rocker-bottom feet, were also recurrent findings. In addition, intrauterine growth restriction and polyhydramnios frequently served as indirect markers of fetal pathology.

It is important to note that some pregnancies initially presented with reassuring ultrasound findings. Normal nuchal translucency measurements and fetal biometric parameters were observed during the first trimester despite subsequent confirmation of trisomy 18. These observations highlight that the absence of early ultrasonographic abnormalities does not exclude the presence of a chromosomal disorder and support the use of complementary screening tools, particularly in pregnancies considered at increased risk.

This case series also suggests a close relationship between delayed diagnosis and the severity of fetal abnormalities. Early diagnoses were generally associated with isolated or limited abnormalities, whereas later diagnoses revealed the progressive accumulation of multisystem malformations. In cases diagnosed during the second or third trimester, abnormalities were more numerous, more severe, and involved multiple organ systems, including the cardiovascular system, characterized by ventricular septal defects and great vessel anomalies; the gastrointestinal system, illustrated by esophageal atresia; the respiratory system, affected by pneumomediastinum; as well as the musculoskeletal and nervous systems. These findings underscore the importance of structured and timely prenatal screening, which facilitates early identification of high-risk pregnancies and supports informed clinical decision-making.

Cytogenetic confirmation played a decisive role in patient management. In this series, confirmation was obtained through chorionic villus sampling, amniocentesis, or, in one case, postnatal karyotyping. Early diagnostic confirmation allowed timely discussions with parents and most often resulted in medical termination of pregnancy following

approval by the institutional ethics committee. Conversely, the absence of prenatal confirmation resulted in continuation of the pregnancy and the need for intensive neonatal management, illustrating the clinical and organizational consequences of delayed diagnosis.

The cases that progressed to birth highlight the severity of postnatal manifestations and the multisystem nature of trisomy 18. The neonatal period was characterized by substantial morbidity requiring admission to a neonatal intensive care unit, mechanical ventilation, and close hemodynamic monitoring. Complex congenital cardiac defects represented a major contributor to clinical deterioration. In these situations, no corrective surgical procedures were undertaken because of the multiplicity of anomalies and the poor overall prognosis. Postnatal survival was limited, with death occurring within the first weeks of life, further emphasizing the severe nature of the condition.

Finally, this series underscores the importance of parental counseling and multidisciplinary support. Decisions regarding continuation or termination of pregnancy rely on clear, individualized, and progressive communication with parents. The role of the multidisciplinary team is essential in integrating medical findings, prognostic information, and parental preferences within an often complex ethical framework.

CONCLUSION

In conclusion, the analysis of these cases highlights the importance of early prenatal screening, combining maternal risk factors, biochemical screening methods, and detailed fetal ultrasonography, in order to optimize clinical management and parental counseling. Delayed diagnosis was associated with increased severity of congenital malformations and the need for intensive postnatal management with poor outcomes, underscoring the critical role of timely prenatal diagnosis in pregnancies affected by trisomy 18.

Early identification and cytogenetic confirmation of the condition facilitate informed decision-making, multidisciplinary care planning, and appropriate parental support. These findings emphasize the value of a structured prenatal screening strategy for improving the detection and management of this severe chromosomal disorder.

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