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Intrauterine Fetal Death at 26 Weeks of Amenorrhea Revealing Turner Syndrome (45,X0): A Case Report

Dr. Dina Houjjaj[1]; Dr. Sara Mouhmouh[2]; Dr. Sara Yasyn[3]; Dr. Fatima Zahra Azraq[4]; Amal Benbella[5]; Bouchaib Allae Eddine[6]; Abdelhai Adibe Filali[7]; Mohammed Hassan Alami[8]; Rachid Bezad[9]

[5,8]Professor

[1]Department of Obstetrics and Gynecology, Les Orangers Maternity Hospital, Mohammed V University, Rabat
[2]Department of Obstetrics and Gynecology, Mohammed VI University, Agadir
[3]Department of Obstetrics and Gynecology, Les Orangers Maternity Hospital, Mohammed V University, Rabat
[4]Department of Obstetrics and Gynecology, Les Orangers Maternity Hospital, Mohammed V University, Rabat
[5,6,7,8,9]Department of Obstetrics and Gynecology, Les Orangers Maternity Hospital, Mohammed V University, Rabat

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Abstract: Turner syndrome (45,X0) is a rare chromosomal aberration characterized by monosomy of the X chromosome, affecting 1 in 2,500 live female births. It is one of the major causes of intrauterine fetal death (IUFD) and second-trimester spontaneous miscarriage. We report the case of a 23-year-old primiparous patient, born from a first-degree consanguineous union, admitted urgently at 26 weeks of amenorrhea for the expulsion of an IUFD. Morphological examination of the fetus revealed signs suggestive of Turner syndrome: cystic nuchal hygroma, generalized anasarca, and a morphological appearance consistent with X monosomy. This case illustrates the importance of antenatal ultrasound follow-up, multidisciplinary management, and genetic counseling in high-risk pregnancies.

Keywords: *Turner Syndrome, 45,X0, Intrauterine Fetal Death, Cystic Hygroma, Consanguinity, IUFD, Anasarca, Antenatal Ultrasound.*

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I. INTRODUCTION

Turner syndrome (TS), first described by Henry Turner in 1938, is the only viable monosomy in humans. It is defined by the partial or total loss of one X chromosome, most often resulting in a 45,X0 karyotype. Its prevalence is estimated at 1 per 2,000 to 2,500 live female births, but this figure considerably underestimates its true frequency: approximately 99% of 45,X0 conceptions end in spontaneous abortion, making Turner syndrome the leading chromosomal cause of first-trimester miscarriage.

Intrauterine fetal death (IUFD) is defined as the death of a fetus occurring after 14 weeks of amenorrhea (WA) and before delivery. Its incidence ranges from 3 to 5 per 1,000 births in developed countries and is significantly higher in settings with consanguinity or limited medical resources. Among the chromosomal etiologies of second-trimester IUFD, Turner syndrome plays a prominent role, particularly because of the cardiovascular malformations and fetal anasarca it produces.

Consanguinity, defined as a union between individuals sharing at least one common ancestor, is a known aggravating factor for congenital anomalies. Although Turner syndrome

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is a de novo, non-hereditary anomaly, first-degree consanguinity may promote the expression of superimposed recessive disorders and constitutes an independent obstetric risk factor.

We report here a case of IUFD at 26 WA in a 23-year-old primiparous patient with first-degree consanguinity, whose morphological examination revealed Turner syndrome (45,X0).

II. CLINICAL OBSERVATION

➢ *Patient Presentation*

Table 1 Patient Presentation

Demographic data	Values
Age	23 years
Parity	Primiparous — G1P0
Consanguinity	First-degree (first-cousin union)
Medical history	No relevant personal history
Obstetric history	None
Gestational age (LMP)	26 weeks of amenorrhea (26 WA)
Reason for admission	Expulsion of an intrauterine fetal death (IUFD)
Antenatal follow-up	Irregular — 1 first-trimester consultation

➢ *History of the Disease*

Mrs. X., a 23-year-old primiparous patient from a spontaneous singleton pregnancy who had received only incomplete antenatal follow-up (a single non-contributory first-trimester ultrasound), presented to the obstetric emergency department with painful contractions and a perceived decrease in fetal movements for the past 48 hours. She also reported progressive abdominal distension and lower-limb edema evolving over the previous ten days or so.

On clinical examination, the patient was afebrile, normotensive, and in preserved general condition. Symphysis-fundal height was 24 cm, lower than the estimated gestational age of 26 WA based on the last menstrual period (LMP). Doppler auscultation of fetal cardiac activity was negative. Emergency obstetric ultrasound confirmed the absence of fetal cardiac activity, establishing the diagnosis of intrauterine fetal death.

The maternal laboratory work-up (complete blood count, coagulation panel, electrolytes, renal and hepatic function, infectious serologies) was within normal limits, with no findings suggestive of an underlying maternal pathology.

➢ *Obstetric Management*

After full information and informed consent of the patient and her family, labor induction with cervical ripening was undertaken. Delivery occurred vaginally after 8 hours of labor. A macerated male fetus, with a birth weight of 1,975 grams, was expelled without immediate maternal complications. Head circumference and complete placental data could not be fully collected in the absence of an exhaustive pathological work-up.

The placenta was macroscopically normal. The umbilical cord contained three vessels on visual inspection.

➢ *Fetal Morphological Examination*

External examination of the fetus after expulsion revealed several major morphological abnormalities:

- Bilateral, voluminous cervical cystic hygroma with a thickened, bulging nape
- Generalized anasarca: diffuse subcutaneous edema predominating on the extremities and face
- Distinctive facial morphology: infiltrated facies, edematous eyelids, half-open mouth
- Moderate cutaneous maceration consistent with old fetal death (> 48 h)
- Birth weight: 1,975 grams at 26 WA (weight increased by generalized edema)
- Apparent male sex on external examination (however to be interpreted with caution given the genital edema)

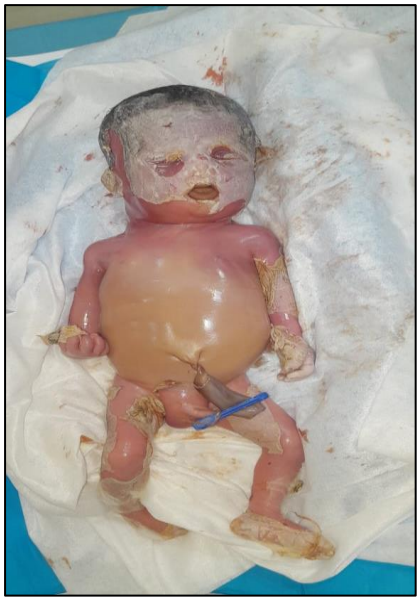

Fig 1 Fetus Expelled at 26 WA. Note the Generalized Anasarca, Thickened Nape with Cystic Hygroma, and Diffuse Edema of the Limbs and Face, Suggestive of Turner Syndrome 45,X0.

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➢ *Post-Mortem Investigations*

No further complementary analyses (complete fetal autopsy, placental biopsy, specific serologies) were performed in this context.

III. DISCUSSION

➢ *Turner Syndrome: Epidemiological and Genetic Background*

Turner syndrome (TS), or X monosomy, is defined by the partial or total loss of the second sex chromosome in a female individual (karyotype 45,X0 in its complete form). With a prevalence of approximately 1/2,500 live female births, it is the most frequent gonosomal chromosomal anomaly in women. In its complete 45,X0 form, it is almost invariably lethal in utero: fewer than 1% of 45,X0 conceptuses reach term.

From a genetic standpoint, Turner syndrome is in the vast majority of cases de novo, without Mendelian hereditary transmission. The origin of the non-disjunction error is most often paternal (loss of the paternal Y or X chromosome in 70–80% of cases). First-degree maternal consanguinity is therefore not directly implicated in the genesis of this numerical chromosomal anomaly; however, it can potentiate other superimposed recessive genetic anomalies and represents a context of heightened obstetric vulnerability.

- *From a Cytogenetic Perspective, Several Forms are Described:*

✓ Complete 45,X0 monosomy: the most severe form, the most frequent in IUFDs (80% of prenatal cases)
✓ 45,X/46,XX or 45,X/46,XY mosaicism: variable phenotype, sometimes milder
✓ Isochromosome i(Xq) or partial deletions of the X chromosome: partial forms

➢ *Intrauterine Fetal Death and Turner Syndrome*

IUFD secondary to Turner syndrome occurs mainly in the first and early second trimester of pregnancy. Survival until 26 WA, as in our observation, is rare and often reflects a milder form or mosaicism. Nevertheless, cases of late second-trimester IUFD have been reported, mainly related to fetal anasarca due to heart failure secondary to aortic coarctation or other cardiovascular malformations.

In our case, the fetal weight of 1,975 g at 26 WA exceeds the expected weight (approximately 850–950 g) because of the generalized edema, which is consistent with the literature on hydrops fetalis associated with TS.

The pathophysiological mechanism of hydrops in TS involves lymphatic obstruction related to agenesis or hypoplasia of the jugular lymphatic ducts, leading to cystic hygroma, fluid accumulation in the serous cavities, and generalized edema.

➢ *Antenatal Ultrasound Warning Signs*

Prenatal diagnosis of Turner syndrome rests on the combination of ultrasound features and fetal karyotyping (amniocentesis or chorionic villus sampling). The main ultrasound warning signs, as derived from the literature and summarized in the table below, are:

Table 2 Antenatal Ultrasound Warning Signs Suggestive of Turner Syndrome (According to the Literature).

Antenatal ultrasound signs suggestive of Turner syndrome
Thickened nuchal translucency, cystic hygroma, hydrops fetalis (anasarca)
Aortic coarctation, left-sided cardiac anomalies
Renal anomalies: horseshoe kidney, renal agenesis or ectopia
Brachycephaly
Polyhydramnios, oligohydramnios
Mild intrauterine growth restriction

Thickened nuchal translucency or cystic hygroma is the earliest and most specific sign, detectable as early as the nuchal translucency measurement in the first trimester (between 11 and 14 WA). A nuchal translucency > 3.5 mm should systematically raise suspicion of TS and prompt fetal karyotyping. Hydrops fetalis (anasarca), aortic coarctation, and renal anomalies (horseshoe kidney, renal agenesis or ectopia) are the other major markers.

➢ *Prognostic Impact and Post-Mortem Management*

The prognosis of pregnancies with Turner syndrome depends essentially on the course of cardiovascular anomalies. Forms with severe hydrops, aortic coarctation, or hypoplastic left heart are associated with near-total intrauterine lethality.

- *Management After IUFD Should Include:*

✓ A complete maternal etiological work-up (coagulation, serologies, autoimmune panel, glycemia)
✓ A systematic fetal morphological examination with precise description of the anomalies
✓ Post-mortem fetal karyotyping (fetal blood, skin, amniotic fluid)
✓ Fetal autopsy where resources allow
✓ Psychological support for the patient and her family
✓ Early genetic counseling for subsequent pregnancies

➢ *Role of Consanguinity*

First-degree consanguinity (first-cousin unions or first-degree-related unions) multiplies the risk of expression of autosomal recessive diseases by a factor of 2 to 4. In this case, IUFD is related to a numerical chromosomal anomaly (de

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novo) and not to a recessive mechanism. Nevertheless, consanguinity remains an obstetric risk factor to consider in the overall etiological work-up, as it may mask or amplify superimposed genetic anomalies. Genetic counseling must take this into account and propose screening for recessive diseases known in the family context.

IV. CONCLUSION

This clinical case illustrates a rare but severe presentation of Turner syndrome: an intrauterine fetal death occurring at 26 weeks of amenorrhea in a 23-year-old primiparous patient with first-degree consanguinity. The fetal morphological signs (cystic hygroma, generalized anasarca) were suggestive from the post-partum clinical examination, and the diagnosis was confirmed by karyotyping (45,X0).

- *This Case Highlights Several Essential Points:*

✓ The crucial importance of regular antenatal ultrasound follow-up, particularly nuchal translucency measurement between 11 and 14 WA, which could have guided the diagnosis from the first trimester.
✓ The need for systematic karyotyping in any second-trimester IUFD, especially in a consanguineous context.
✓ The value of multidisciplinary management involving obstetrician, geneticist, pediatrician, and psychologist.
✓ The role of genetic counseling before any subsequent pregnancy, in order to inform the couple about the risk of recurrence (low for de novo TS) and to consider prenatal diagnosis.

Raising clinician awareness of the early warning signs of Turner syndrome is essential to improve antenatal detection and optimize the management of affected couples.

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