

# A Retrospective Review of 25 cases of Lethal Fetal Anomalies

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## ABSTRACT

**Introduction:** To review the gestational age at diagnosis, method of diagnosis, pregnancy outcome and maternal complications of prenatally diagnosed lethal foetal anomalies. **Methods:** Retrospective review of 25 women who had aborted or delivered fetuses with lethal anomalies in a tertiary hospital in 2011 based on patient medical records. **Results:** There were a total of 10,088 deliveries, in which 25 (0.24%) women were found to have conceived fetuses with lethal anomalies. All of them were diagnosed by prenatal ultrasound and only 7 (28.0%) had both prenatal ultrasound and genetic study done. The women's mean age was 29.9 years old. The mean gestational age at diagnosis of lethal foetal anomalies was 25.5 weeks (SD=12.5) and mean gestational age at termination of pregnancy (TOP) or delivery was 28.5 weeks (SD=12.5). Seven (28%) women had early counseling and TOP at the gestation of < 22 weeks. Beyond 22 weeks of gestation, eight (32%) women had TOP and ten (40%) women had spontaneous delivery. Twenty (80%) women delivered or aborted vaginally, three (12%) women with assisted breech delivery and two (8%) women with abdominal delivery which were performed due to transverse foetal lie in labour and a failed induction, leading to emergency hysterotomy complicated by hysterectomy due to intraoperative finding of ruptured uterus. Overall, the associated post-partum adverse events included post-partum haemorrhage (12%), retained placenta (12%), blood transfusion (8%), uterine rupture (4%) and endometritis (4%). Mean duration of hospital stay was 6.6 days (SD 3.7 days). **Conclusion:** Late diagnosis of lethal foetal anomalies leads to various maternal morbidities, in this case series, which could have been prevented if they were diagnosed and terminated at early trimester. A new direction is needed in our local practice.

**KEYWORDS:** Lethal foetal anomalies, termination of pregnancy, anomaly scan, prenatal screening/ diagnosis

## INTRODUCTION

The advances of prenatal screening and diagnosis have resulted in an increasing number of lethal congenital foetal anomalies diagnosed during pregnancy.<sup>1</sup> Parents and obstetricians face a dilemma of either terminating or continuing with the pregnancy and delivering the child. That baby will either die soon after birth, or be severely handicapped.<sup>2</sup> Still, in many instances, the diagnoses were made at advanced gestational age, where therapeutic options are very limited or none.

Many developed countries practice routine prenatal screening and diagnosis for foetal anomalies, which can give reassurance for normal pregnancy, as well as time for the couple who have been diagnosed carrying fetus with anomaly to have more time for information, and to plan for possible antenatal intervention, time and mode of delivery and option of termination.<sup>1</sup>

Prenatal screening for structural and chromosomal abnormalities in most of the hospitals in Malaysia including ours is only on women with risk factors. This includes advanced maternal age, previous personal or family history of chromosomal or structural abnormalities, medical illness, abnormal amniotic fluid index (AFI) or intrauterine growth restriction, and they are offered prenatal anomaly ultrasound.

The majority of cases were diagnosed incidentally, causing delay in referral and appropriate management. The purpose of this study was to review the gestational age of diagnosis, the

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pregnancy outcome as well as the maternal complications in a local tertiary centre.

## MATERIALS & METHODS

This is a retrospective review done in a tertiary referral hospital in the year 2011. This hospital has foeto-maternal unit and consultants but does not practice universal routine prenatal screening tests. Foeto-maternal consultants will only perform 18<sup>th</sup> to 24<sup>th</sup> weeks prenatal foetal anomaly scan for women with risk of foetal anomalies or referred cases with suspicious antenatal ultrasound findings. Prenatal diagnostic procedures are offered only following suspicious ultrasound findings.

We define lethal foetal anomaly as fetal anomaly that causes a foetus to die in the womb or will die shortly after birth.<sup>3</sup>

Our data were collected from the clinic ultrasound and labour room books, and records of patients diagnosed prenatally carrying foetuses with lethal anomalies were collected from the hospital's record office and reviewed.

The cases in our series were either picked up during our foetal anomaly scan sessions or as referral from other health centres or clinics. The diagnosis of lethal foetal anomalies was made via ultrasound assessment and only few had genetic study confirmation. The maternal demographic data, gestation age at diagnoses, gestation age at termination or delivery, morbidities during antenatal, intrapartum, and postpartum period, mode of induction and the type of delivery were analysed.

## RESULTS

During the study period, the total deliveries were 10,088. There were 25 (0.25%) pregnancies diagnosed with lethal foetal anomalies in which 10 (40%) were foetuses with multiple structural abnormalities. Others include anencephaly, encephalocele, hydrops fetalis and syndromic fetuses as shown in table 1. The mean maternal age was 29.9 years (range 22-43 years old) and interestingly, 19 (76 %) of them aged less than 35 years. The majority were Malays [19

(76.0%)], six (24%) were diabetic, one (4.0%) woman had previous history of unexplained intrauterine death (IUD) and none of them had previous pregnancy with congenital anomalies.

The average gestational age at diagnosis was 25.5 weeks (SD=12.5) and mean gestational age at termination of pregnancy (TOP) or delivery was 28.5 weeks (SD=12.5). Only 7 (28 %) women were diagnosed early in the first trimester with the rest of them [18 (72 %)] were invariably diagnosed after 22 weeks.

All 25 foetuses with lethal anomalies were diagnosed via ultrasound. Only 7 (28.0%) women proceeded for prenatal invasive diagnosis (three amniocentesis, three cordocentesis and one chorionic villi sampling) for standard karyotyping studies. The remaining 18 (72.0%) pregnancies did not undergo prenatal invasive diagnostic procedures.

Eight (32%) cases agreed for TOP and 10 (40%) cases opted to wait for spontaneous delivery. Among the antenatal complications observed were IUD (24.0%), preterm labour (20.0%), symptomatic polyhydramnios requiring amnioreduction (16.0%) and two mothers (8.0%) developed gestational hypertension with one of them went on to develop impending eclampsia requiring intravenous MgSO<sub>4</sub> infusion at the gestational age of 35 weeks.

Out of 15 women who planned for termination, 9 women were induced with gemeprost (gestational age ranged from 13 to 24 weeks) and 6 women were induced with dinoprostone (gestational age ranged from 28 to 36 weeks).

Twenty three (92.0%) pregnancies delivered or aborted vaginally, and two (8.0%) pregnancies with abdominal delivery. The two abdominal deliveries were an emergency lower segment caesarean section for transverse lie in labour at 37 weeks of gestation and an emergency hysterotomy for failed induction at 24 weeks of gestation. The mother who went for emergency hysterotomy was a grandmultipara and the surgery ended with hysterectomy due to an intraoperative finding of ruptured uterus.

Post partum complications observed were postpartum haemorrhage (12.0%), retained placenta (12.0%), blood transfusion (8.0%), endometritis (4.0%) and uterine rupture (4.0%). There was no case of maternal mortality in this series.

The mean duration of hospital stay was 6.6 days (SD 3.7 days). Seven patients (28.0%) required re-hospitalisation: four for antenatal morbidity and three for repeated course of medical termination (Figure 1).

**Table 1.** Type of lethal fetal anomalies

Type of lethal foetal anomalies	n=25 (%)
Fetuses with multiple structural abnormalities	10 (40.0)
Anencephaly or severe encephalocele	8 (32.0)
Non-immune hydrops fetalis	4 (16.0)
Syndromic fetuses (two cases of Pentalogy of Cantrell and a case of Edward's Syndrome)	3 (12.0)

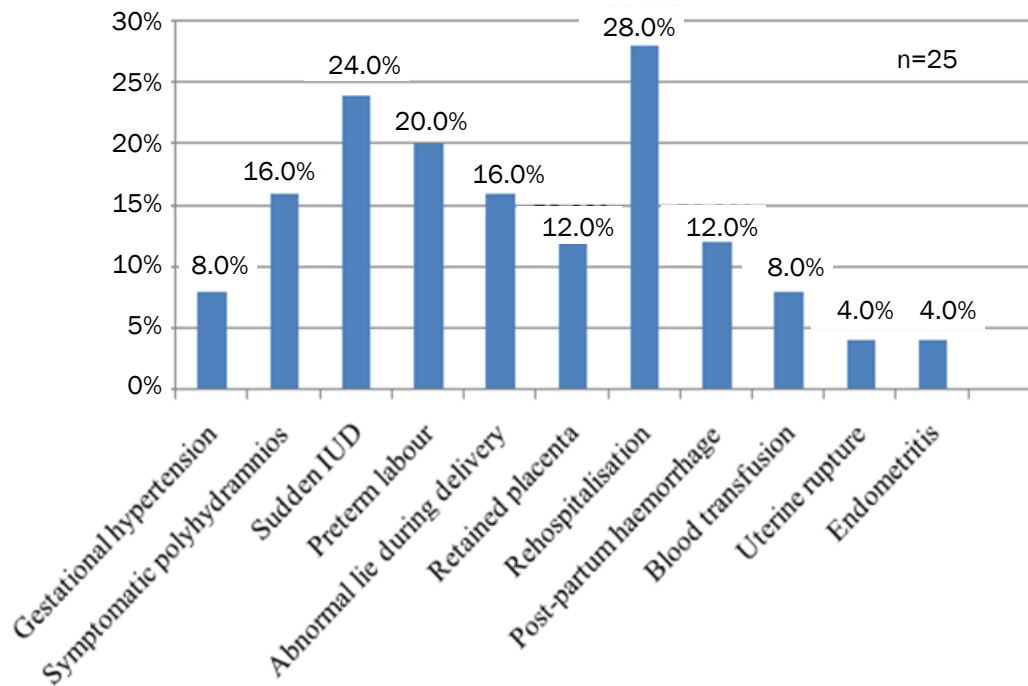


Figure 1. Pregnancy outcomes of prenatally diagnosed lethal foetal anomalies

## DISCUSSION

Most of the cases were diagnosed late. The mean gestational age at diagnosis was 25.5 (range 13-38) weeks, where only 7 cases were diagnosed before 22 weeks of gestation and 18 cases were diagnosed after 22 weeks. Most lethal foetal anomalies found in this series could be diagnosed by ultrasound much earlier, for example; anencephaly which can be detected as early as 11 weeks with an accuracy of almost 100% at 14 weeks, Pentalogy of Cantrell,<sup>4,5</sup> encephalocele which usually can be diagnosed by 13-14 weeks<sup>6</sup> and gross anomalies of Edward's syndrome at gestation age of 17.5-24 weeks. The sensitivity of screening can be increased up to 90% by combining ultrasound findings with the result of maternal serum marker.<sup>7</sup>

Due to the absence of first trimester prenatal screening services, cases were seen either as a result of referral from other clinics or health centers, incidental findings or findings of 18-24 weeks anomaly scan clinics for women with risk factors. Only 13 (52%) cases had risk factors; 6 mothers were 35 years old or more, 6 with preexisting diabetes mellitus and one with unexplained IUD. It has been reported that about 95% of abnormalities occur in fetuses born to mother without risk.<sup>8</sup> Our findings also emphasized the need for routine prenatal screening for all women not only those with risk factors. However, to prenatally diagnose one lethal foetal anomaly, we need to do prenatal anomalies scan on 400 pregnant ladies in a

referral tertiary hospital. The denominator used was the total deliveries in our centre per year, which does not include the total number of aborted cases or miscarriage.

With a combination of first and second trimester serum biochemical marker, nuchal translucency and ultrasound soft marker, anomaly scan at 18-20 weeks and improvement of ultrasound technology, diagnosis of foetal anomaly could be made at much earlier stage. Morbidities could be avoided when earlier diagnosis is made at the time when prenatal invasive diagnostic tests could still be feasible, and termination is still an option in a consented couple.<sup>9,10</sup> In our series just 7(28%) women had prenatal invasive procedure while 8(32%) women went into IUD or spontaneous labour because of late diagnosis.

Early diagnosis has potential benefits as termination is safer if it is performed earlier. The risk of termination increases with gestational age. Complication can occur from haemorrhage, uterine perforation and/or sepsis where the likelihood can increase from 5 per1000 medical procedures at 10-12 weeks to 16 per1000 at or after 20 weeks of gestation.<sup>9</sup> In our retrospective review 14 per 25(56%) cases have various morbidity that could be explained as almost all of these women (12 out of 14, or 85%) had termination after 22 weeks.

The adverse events of morbidities reported in our group (sudden IUD, preterm labour, abnormal lie during delivery and symptomatic polyhydramnios requiring amnioreduction, preterm labour, preterm prelabour rupture of membrane (PPROM), placenta abruptio, severe pre-eclampsia and coagulopathy and malpresentation requiring elective CS is similar to those found by Byrne and Morrison.<sup>11</sup> Diagnosis of lethal congenital anomalies in early pregnancy reduced the number of subsequent stillbirths.<sup>12</sup>

A study by Tariq et al shows polyhydramnios found in 31.7% cases diagnosed with congenital anomaly. Most of them were severe polyhydramnios.<sup>13</sup> In our review the 4 (16%) women with polyhydramnios were diagnosed at 25, 34, 31, and 37 weeks of gestation associated with foetal congenital anomalies, two fetuses with hydrops fetalis, one having hydrocephalus cyclop and one Edward syndrome.

Both fetuses with hydrops fetalis did not undergo karyotyping because the patients refused and also due to financial constraints. Cordocentesis revealed normal karyotyping for the foetus with hydrocephalus cyclop. All the three cases required symptomatic amnioreduction. The one with Edward syndrome had its karyotype confirmed after having done amniocentesis at 37 weeks. The woman refused termination or induction of labour and wished for conservative management. She required symptomatic amnioreduction at the tenth day post-date, and delivered on the same day.

Majority of the complications i.e. gestational hypertension with impending eclampsia, polyhydramnios, preterm delivery and abnormal lie during delivery requiring operative delivery arise when TOP is performed at later gestational age.<sup>14</sup>

#### Limitation

Being a retrospective study, we were not able to analyse the maternal psychological impact following prenatal diagnosis of lethal foetal anomalies and the effects of the delayed termination.

#### Recommendation

A universal ultrasound screening should be offered to all pregnant ladies regardless of their risk factors in view of:

- i. Better services in local health centers:
  - a. number of ultrasound machines available
  - b. availability of family medicine specialists who are trained to do obstetrics ultrasound imaging
- ii. Better services in tertiary hospitals
  - a. availability of high-ranged ultrasound machine
  - b. increasing number of feto-maternal medicine specialist

In our review, we found out that to diagnose one lethal foetal anomaly prenatally, we need to have 400 cases scanned. As such, we recommend the routine prenatal anomaly scan to be done by local health center in early gestation and then followed by early referral to the tertiary hospital.

#### CONCLUSION

Late diagnosis of lethal foetal anomalies leads to various maternal morbidities. In this case series, the morbidities could have been prevented if they were diagnosed and terminated earlier in the pregnancy. A new direction is needed in our local practice.

#### Competing interests

The authors declare that they have no competing interests.

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