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The Journal of Maternal-Fetal & Neonatal Medicine

ISSN: 1476-7058 (Print) 1476-4954 (Online) Journal homepage: informahealthcare.com/journals/ijmf20

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To cite this article: Dong-Zhi Li & Han Liu (2012) Prenatal diagnosis of trisomy 18: Report of 76 cases in a mainland Chinese hospital, The Journal of Maternal-Fetal & Neonatal Medicine, 25:10, 2144-2144, DOI: 10.3109/14767058.2012.678428

To link to this article: https://doi.org/10.3109/14767058.2012.678428


Published online: 20 Apr 2012.


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The Journal of Maternal-Fetal and Neonatal Medicine, 2012; 25(10): 2144
© 2012 Informa UK, Ltd.
ISSN 1476-7058 print/ISSN 1476-4954 online
DOI: 10.3109/14767058.2012.678428

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LETTER TO THE EDITOR

Prenatal diagnosis of trisomy 18: Report of 76 cases in a mainland Chinese hospital

Dear Editor,

Trisomy 18 (Edwards syndrome) is the second most common autosomal trisomy. Approximately 95% of conceptuses with trisomy 18 die as embryos or fetuses. For those who survived, the estimated probability of survival up to the age of 1 month was 38.6% and up to the age of 1 year was 8.4%; median survival time was 14.5 days [1]. The high mortality rate is usually due to the presence of cardiac and renal malformations, feeding difficulties, sepsis, and apnea caused by CNS defects. Prenatal diagnosis allows for early informed choices and better preparation for the parents. The present study reports on prenatal diagnosis of trisomy 18 in a regional obstetric unit in mainland China.

From 1 January 2000 to 31 June 2010, there were 76 women with pregnancies confirmed prenatally to have trisomy 18 by karyotyping at the Prenatal Diagnosis Center of Guangzhou Maternal and Neonatal Hospital, a regional hospital with more than 5000 deliveries per year in mainland China. Patients were referred to invasive testing for indications of advanced maternal age, abnormal ultrasound findings, or trisomy 21 screening tests (Table I).

The mean maternal age was 31.0 (20–46) ± 6.0 years. The mean gestational age at which prenatal diagnosis of trisomy 18 was made was 23.4 (11–35) ± 7.2 weeks (Table II). Of the 76 women with prenatal invasive tests, most had diagnosis by cordocentesis (n = 34, 44.7%), 30 (39.5%) had amniocentesis, and 12 (15.8%) had chorionic villous sampling.

Early detection can be of benefit with all types of birth defects, particularly in giving the couple the option to either terminate the pregnancy or prepare for the delivery of an affected child. Trisomy 18 is incompatible with life beyond 1 year of age; thus, termination is quite reasonable. First- or second-trimester abortion is associated with lower rates of complications and mortality to the woman than induction of labour in the third trimester. However, at our center, more than one third of affected pregnancies were diagnosed beyond the late second trimester (24 weeks), and even in the third trimester. Only 11 cases were detected in the early gestation (≤14 weeks). Indeed, all of these 11 patients were identified in the past 3 years, when the first trimester combined screening program for trisomy 21 was established. Ultrasonographic features of trisomy 18 in the first trimester include increased NT, exomphalos, cystic hygroma, or hydrops fetalis.

In contrast with the well-established prenatal screening methods for trisomy 21, screening strategies for trisomy 18 are not fully developed because of its lower incidence and its lethality. Ultrasound scan for fetal anomalies is the most effective screening test [2–4]. As also shown in the current study, almost all of the affected pregnancies were referred for the indication of abnormal ultrasound findings. At our hospital nearly half of the pregnant women who come for their first prenatal care appointment are in the last half of gestation. For these women, ultrasound is the main assessment method for detecting fetal abnormalities, and chromosomal abnormailities are usually revealed in the late gestation. However, late prenatal diagnosis of trisomy 18 is still useful because unnecessary interventions, including caesarean section, can be avoided.

Table I. Indications for prenatal karyotyping.

Indications	No. of cases
Ultrasound abnormalities	59
Abnormal second-trimester serum screening and USG abnormalities	12
Abnormal first-trimester combined	4
Screening and USG abnormalities	
Advanced maternal age	1
Total	76

USG, ultrasonography.

Table II. Gestation at which prenatal diagnosis of trisomy 18 was made.

Gestation age	No. of cases (weeks)
11–14	11
15–20	21
21–24	13
25–28	10
≥29	21
Total	76

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***Declaration of Interest*:** The authors report no declarations of interest.

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