นิพนธ์ต้นฉบับ • Original Article

ลักษณะทางคลินิกของมารดา ทารกในครรภ์และทารกแรกเกิดของกลุ่มอาการดาวน์ที่โรงพยาบาลศรีนครินทร์

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Maternal-Fetal and Neonatal Clinical Characteristics of Down Syndrome at Srinagarind Hospital

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วัตถุประสงค์: เพื่อศึกษาลักษณะทางคลินิกของมารดาที่คลอดบุตรกลุ่มอาการดาวน์ ลักษณะทางคลินิกของทารกในครรภ์กลุ่มอาการดาวน์และลักษณะทางคลินิกของทารกแรกเกิดกลุ่มอาการดาวน์ ที่โรงพยาบาลศรีนครินทร์

วิธีการศึกษา: เป็นการศึกษาเชิงพรรณนาย้อนหลัง โดยการค้นประวัติสตรีตั้งครรภ์ที่คลอดทารกกลุ่มอาการดาวน์ที่โรงพยาบาลศรีนครินทร์จากเวชระเบียน ระหว่างวันที่ 1 มกราคม พ.ศ. 2536-31 ธันวาคม 2555 จากนั้นจึงลงข้อมูลพื้นฐานต่างๆ ลงในแบบบันทึกข้อมูลและวิเคราะห์สถิติเป็นร้อยละ

ผลการศึกษา: อุบัติการณ์ของการคลอดทารกกลุ่มอาการดาวน์ที่โรงพยาบาลศรีนครินทร์คือ 0.61 ต่อ 1,000 ของการคลอดทั้งหมด ร้อยละ 57 ของสตรีตั้งครรภ์ที่คลอดทารกกลุ่มอาการดาวน์มีอายุมากกว่า 35 ปี ร้อยละ 52 คลอดก่อนกำหนดและไม่มีประวัติการตั้งครรภ์ผิดปกติในครรภ์ก่อน แต่มีเพียงร้อยละ 18 ที่ได้รับการวินิจฉัยก่อนคลอด และพบว่ามารดาที่คลอดทารกกลุ่มอาการดาวน์มักจะมีประวัติเป็นเบาหวาน ลักษณะจากการตรวจคลื่นเสียงความถี่สูงของทารกกลุ่มอาการดาวน์ที่พบมากที่สุดในการศึกษานี้คือการเจริญเติบโตช้าในครรภ์ ลักษณะของทารกแรกคลอดที่เป็นกลุ่มอาการดาวน์มักจะมีเส้นรอบวงศีรษะเล็กกว่าเกณฑ์ปกติ

Objective: To describe maternal-fetal and neonatal clinical characteristics of Down Syndrome (DS) at Srinagarind Hospital.

Materials and Methods: The present study was a retrospective descriptive study. The authors recruited all pregnant women who delivered babies with DS at Srinagarind hospital between January 1, 1993 and December 31, 2012. Histories of all participants were obtained from medical records. Clinical characteristics of them were recorded in case record form (CRF). The statistics used to describe the clinical characteristics were percentage.

Results: The incidence of DS at Srinagarind hospital was 0.61 in 1,000 of total births. 57% of pregnant women who delivered babies with DS are 35 years or older. 52% delivered before term gestation and had no abnormal obstetric history in previous pregnancies. Only 18% had definite prenatal diagnosis. DS babies occur more frequently in infants of diabetic mothers. The most common ultrasonographic finding of the fetuses with DS in our study was asymmetrical intrauterine growth restriction. For full term neonates with DS, the average value of head circumference was slightly smaller than normal.

สรุป: อุบัติการณ์ของการคลอดทารกกลุ่มอาการดาวน์ที่โรงพยาบาลศรีนครินทร์คือ 0.61 ต่อ 1,000 ของการคลอดทั้งหมด จากการศึกษานี้พบว่า มารดาที่มีอายุมากและมารดาที่เป็นเบาหวานอาจจะเป็นปัจจัยเสี่ยงหลักของการเกิดทารกกลุ่มอาการดาวน์ การตรวจวินิจฉัยก่อนคลอดยังมีอัตราที่ต่ำ และทารกกลุ่มอาการดาวน์มักจะมีเส้นรอบวงศีรษะเล็กกว่าเกณฑ์ปกติ

คำสำคัญ: กลุ่มอาการดาวน์, ลักษณะทางคลินิก, มารดา, ทารกในครรภ์, ทารกแรกเกิด

Conclusion: Incidence of DS at Srinagarind Hospital was 0.61 in 1000 of total births. In this study, advanced maternal age and maternal gestational DM, probably, may be the important risk factors of having a child with DS. Definite prenatal diagnosis was still lower rate. For neonatal clinical characteristics, head circumferences were smaller in DS babies.

Keywords: Down syndrome, Characteristic, Maternal, Fetal, Neonatal

ศรีนครินทร์เวชสาร 2560; 32(6): 506-10. • Srinagarind Med J 2017; 32(6): 506-10.

Introduction

Down syndrome (DS), also known as trisomy 21, is a genetic disorder caused by the presence of all or part of a third copy of chromosome 21[1]. Those with DS nearly always have both intellectual and physical disabilities such as low muscle tone, small stature, heart defect, digestive abnormalities, an upward slant to the eyes and a single deep crease across the center of the palm[2].

Though DS can't be prevented but it can be detected before a child is born. Two types of prenatal tests are used to detect DS in a fetus: screening tests and diagnostic tests.

Screening tests, prenatal ultrasonography and biochemical serum screenings, estimate the risk that a fetus has DS. Diagnostic tests include chorionic villous sampling, amniocentesis and percutaneous umbilical blood sampling (PUBS) can tell whether the fetus actually has the condition.

In Thailand, no previous reports mentioned about maternal-fetal and neonatal clinical characteristics of DS. The objective of this study was to describe maternal-fetal and neonatal clinical characteristics of DS at Srinagarind hospital.

Materials and methods

The present study was a retrospective descriptive study. The authors recruited all pregnant women who delivered babies with DS at Srinagarind Hospital between January 1, 1993 and December 31, 2012. Histories of all participants were obtained from medical records. Clinical characteristics of them consisted of: 1) gravidity, gestational age and maternal age at delivery, history of antenatal care, previous and present obstetric history 2) prenatal ultrasonography with details of anomalies, prenatal diagnosis and results 3) fetal gender, birth weight, growth, head circumference, length and abnormal clinical findings. The statistics used to describe the clinical characteristics were percentage.

This study conformed to the declaration of Helsinki and was approved by the Khon Kaen University Ethics Committee for Human Research, HE561041.

Results

During the period of study, there were 72,172 total births. Forty-four pregnant women who delivered babies with DS were enrolled in this study. The incidence of DS at Srinagarind hospital was 0.61 in 1000 of total births. The maternal clinical characteristics of DS are shown in Table 1. Most of their age are more than 35 years old with no abnormal obstetric history in previous pregnancies. For current pregnancy, 3 of them are gestational DM.

Table 1 Maternal clinical characteristics who delivered babies with Down Syndrome

Characteristics	Numbers (%)
1. Maternal age (years)	
< 35	19 (43)
≥ 35	25 (57)
2. Gravity	
1	15 (34)
≥ 2	29 (66)
3. Gestational age at delivery (wks)	
< 28	5 (11)
28-33	3 (7)
34-36	15 (34)
37-42	21 (48)
> 42	0 (0)
4. Antenatal care	
No	8 (18)
Yes	36 (82)
5. Abnormal obstetric history in previous pregnancies	
No	42 (96)
Yes	2 (4)*
Abnormal obstetric history in current pregnancies	
No	40 (91)
Yes	4 (9)**

*1 dead fetus in utero with fetal abdominal wall defect, 1 fetal ventriculomegaly with delayed development

**3 gestational DM, 1 premature rupture of membrane

The fetal clinical characteristics of DS are shown in Table 2. Asymmetrical intrauterine growth restriction is the most common prenatal ultrasound finding. Most of fetal clinical characteristics of Down syndrome have no history of prenatal diagnosis.

Table 2 Fetal clinical characteristics of Down syndrome (Total 44 fetuses)

Characteristics	Numbers (%)
1. Prenatal ultrasound	
Not performed	34 (77.3)
Performed	10 (22.7)
Normal	5 (11.4)
Abnormal	5 (11.4)
Duodenal atresia	1 (2.3)
Asymmetrical intrauterine growth restriction	2 (4.5)
Fetal cardiomegaly with ascites	1 (2.3)
Hydrops fetalis	1 (2.3)
2. Prenatal diagnosis	
No	36 (81.8)
Yes	8 (18.2)
Amniocentesis	7 (87.5)
Cordocentesis	1(12.5)

The neonatal clinical characteristics of DS are shown in Table 3. Most of them are term deliveries, normal birth weight with average head circumferences 32-33 cm.

Table 3 Neonatal clinical characteristics of Down syndrome (Total 44 neonates)

Characteristics	Numbers (%)
1. Gender	
Male	20 (45)
Female	19 (43)
Ambiguous	5 (12)
2. Birth weight (gm)	
<2,000	4 (10)
2,000-2,499	9 (20)
2,500-3,000	20 (45)
>3,000	11 (25)
3. APGAR score at 1 minute	
≤7	16 (36)
>7	28 (64)
4. Intrauterine growth	
AGA	30 (68)
SGA	3 (7)
LGA	3 (7)
No medical recorded	8 (18)
5. Head circumference (cm)	
<29	0 (0)
29-31	17 (39)
32-33	21 (48)
>33	6 (13)
6. Body Length (cm)	
<40	4 (10)
40-44	8 (18)
45-50	23 (52)
>50	9 (20)
7. Abnormal clinical findings	
Low set ears	1 (2.3)
Slant eyes	1 (2.3)
Micrognathia	1 (2.3)
Siamese crease	1 (2.3)
Clinodactyly	1 (2.3)
Heart defect	1 (2.3)

Discussion

The incidence of fetal trisomy is directly related to maternal age[1]. The risk of having a child with DS increases gradually until about age 30 and increases exponentially thereafter. In this study, 57% of pregnant

women who delivered babies with DS are 35 years or older which is similar to another study[3]. No previous report about the oldest and the youngest maternal age but, in this study, are 45 and 16, respectively. And also, no previous study reported about gestational age at delivery and abnormal obstetric history in previous pregnancies of DS babies but, in this study, 52% of pregnant women delivered before term gestation and had no abnormal obstetric history in previous pregnancies. For current pregnancies, 9% of pregnant women had abnormal obstetric histories. Three of them had gestational DM. When compared to study of Narchi[4], the result is similar, DS babies occur more frequently in infants of diabetic mothers.

The incidence of DS at Srinagarind Hospital was 0.61 in 1000 of total births. Comparing to Mandal A[5], the incidence is 1 in 1000 of total births. The incidence in our study is lower, may be due to the data missing from a retrospective study.

Second-trimester ultrasound assessment may be helpful for predicting the likelihood of trisomy 21 in pregnancies at increased risk[6,7]. Ten pregnant women underwent prenatal second trimester ultrasound, then ten fetal characteristics were evaluated. The ultrasound findings of five of them were normal. The rest of them were abnormal. Comparing to study of Chitty[8], 40% of DS fetuses had normal ultrasonographic findings, similarity to our study, 50% is normal. The most common ultrasonographic finding in our study is asymmetrical intrauterine growth restriction, which is different from others. As compared to other studies[6,7,9], the most common ultrasonographic finding associated with trisomy 21 is increased nuchal fold thickness. Advanced gestational age and getting the ultrasound from general obstetricians, not ultrasonographic specialists, may be the causes of difference.

Most of the full term neonates with DS in this study have average length and weight at birth between 45-50 cm and 2500-3000 gm, respectively, which are not different from normal full term babies[9]. But, for head circumference, the average value in this study is slightly smaller than normal, which is commonly seen in neonate with DS[5,10].

DS is usually identified soon after birth by a characteristic pattern of dysmorphic features. In this study, those dysmorphic features recorded seem to be less than normally, missing data or no recorded maybe the reason. There are no specific dysmorphic features. Comparing to other studies[11,12], the 2 most common dermographic signs are flat facial profile and poor moro reflex.

The strength of this study, this is the first study in Thailand that mentioned about maternal-fetal and neonatal clinical characteristics of DS. Our analysis has several limitations.

Some of data are missed due to a retrospective study. Further prospective study should be considered to complete all data.

Conclusion

Incidence of DS at Srinagarind Hospital was 0.61 in 1000 of total births. In this study, advanced maternal age and maternal gestational DM, probably, maybe the important risk factor of having a child with DS. Definite prenatal diagnosis was still lower rate. For neonatal clinical characteristics, head circumferences were smaller in DS babies.

Acknowledgement

This study was funded by Thai Health Promotion Foundation and Birth Defects Associations (Thailand).

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