Second Trimester Genetic Ultrasound for Down Syndrome Screening at Srinagarind Hospital

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Objective: *To assess the value of second trimester genetic ultrasound for screening of Down syndrome conducted at Srinagarind Hospital, Khon Kaen, Thailand.*

Material and Method: *The present study sample comprised of 4,033 pregnant women at high risk for fetal chromosomal abnormality, from 17th to 23th week, who had performed second trimester genetic ultrasound before genetic amniocentesis between September 1996 and December 2011. Archived medical records relating to results of genetic ultrasound and genetic amniocentesis were extracted and studied.*

Main outcome measure: *Sensitivity of genetic ultrasound in the detection of fetal Down syndrome.*

Results: *There were 3,966 chromosomally normal pregnancies (98.3%), 43 fetuses with Down syndrome (1.1%), and 24 fetuses with other chromosomal abnormality (0.6%). 30 of 43 (69.8%) fetuses with Down syndrome had abnormal genetic ultrasound. The overall sensitivity of second trimester genetic ultrasound for detecting Down syndrome was 69.8% with a false-positive rate of 50.4% and likelihood ratio of 1.38. Of all the sonographic markers, short femur, and short humerus indicated the highest sensitivity at 65.1% and 44.2%. According to likelihood ratio (LR+), chest abnormality, 2 vessel umbilical cord, and facial abnormality, including cleft lip and palate, have highest likelihood ratio (LR+) of 61.49, 46.12, and 46.10, and had sensitivity at 4.7%, 2.3%, and 2.3% respectively.*

Conclusion: *The sensitivity of second trimester genetic ultrasound for detection of fetal Down syndrome at Srinagarind Hospital was rather high, and probably is an alternative method of prenatal prediction for high risk pregnant women who refused genetic amniocentesis.*

Keywords: *Genetic ultrasound, Down syndrome screening, Second trimester*

J Med Assoc Thai 2014; 97 (Suppl. 10): S89-S96
Full text. e-Journal: *http://www.jmatonline.com*

Down syndrome is one of the most common chromosomal abnormalities caused by extra genetic material in chromosome 21 (also called trisomy 21) which influences and alters the course of physical, mental and cognitive development in a child born with the condition[1,2]. It is the leading cause of prenatal chromosome aberrations in the world, accounting for 53.0% of all reported chromosome conditions[3]. According to the World Health Organization (WHO), the estimated incidence of Down syndrome is between 1 in 1,000 to 1 in 1,100 live births worldwide[1]. The incidence of Down syndrome at Srinagarind Hospital, Khon Kaen, Thailand is somewhat similar to the figures, 8 in 5,420 total births, according to one year data collected from April 1997 to March 1998[4].

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Amniocentesis has been used worldwide for over the last two decades for prenatal testing to detect Down syndrome in fetuses[5]. Srinagarind Hospital also has been utilizing amniocentesis, in which 0.95% of all procedures from 1993 to 2003, fetal Down syndrome were detected[6]. But amniocentesis is not without risks; it is highly invasive and is associated with procedure-related miscarriage[5]. According to one comprehensive study on amniocentesis, when maternal age is used as screening criteria, one normal fetus may be lost for every 2 fetuses detected with trisomy 21 as a complication of amniocentesis, and combining maternal age and triple screen criteria, one fetus may be lost for every 3-4 fetuses identified with trisomy 21[7,8]. A study conducted at Srinagarind Hospital in 2011 found that the incidence rate of miscarriage after amniocentesis to be 0.6%[9]. Similarly, the perception of pregnant women who were at high risk of having babies with Down syndrome also indicate the low popularity of amniocentesis, with 46.2% expressing fear of

miscarriage and 15.5% declining the procedure[6].

Over the years, genetic ultrasound has come up as a screening option for Down syndrome, which offers prospects of non-invasiveness and good detection rates (60.0 to 90.0%)[5]. Genetic ultrasound is performed in the second trimester to detect fetal aneuploidy, particularly trisomy 21, in which presence of fetal structural abnormalities, aneuploidy markers and anomalous fetal biometry are studied[8]. The main markers include nuchal fold, pyelectasis, short femur and short humerus, hyperechogenic bowel, echogenic intracardiac focus, and any other significant abnormality. The presence of any of the sonographic markers may raise the risk of Down syndrome[10,11].

Many studies have been conducted all over the world on the use of genetic ultrasound for screening of Down syndrome. This descriptive retrospective study aimed to assess the value of genetic ultrasound for screening of Down syndrome conducted at Srinagarind Hospital.

Material and Method

This descriptive retrospective study involved a comprehensive study of the results of genetic ultrasound for screening of Down syndrome conducted at Srinagarind Hospital, Faculty of Medicine, Khon Kaen University over a 15-year period between September 1996 and December 2011. The present study population comprised of pregnant women who had performed second trimester genetic ultrasound by Maternal and Fetal Medicine specialists before genetic amniocentesis. A sample of 4,033 of such cases was evaluated. Archived medical records related to results of genetic ultrasound and genetic amniocentesis were extracted and studied. The data extraction was carried out during June to August 2013. Data extraction procedure included collection of basic information such as age, parity, and previous pregnancy history, current pregnancy information about age of gestation, and indication for amniocentesis. The data collected from genetic ultrasound findings included fetal biometry, structural abnormality, and ultrasound markers.

Statistical analysis were performed by SPSS 11.5 and Epi Info 6. Simple descriptive statistics, such as frequency, percentage, mean and standard deviation was used to summarize the data, and calculate sensitivity, specificity, false-positive rate (FPR) and likelihood ratios (LR+) with 95% confidence interval (CI). The sensitivity was calculated as the proportion of cases with positive test results. The false positive rate was calculated as the proportion of normal fetuses with positive test results; which is equivalent to 1-specificity. The LR+ was calculated as the sensitivity/false-positive rate. Expected humeral length (EHL) = -7.9404 + 0.8492 x BPD If EHL <0.9 is considered to be abnormal. Expected femur (EFL) = -9.645 + 0.9338 x BPD; If EFL <0.91 is considered to be abnormal. This research project was approved by the Human Research Ethical Committee of Khon Kaen University based on the principle of Declaration of Helsinki and ICHGCP standards (Reference No. HE561067).

Results

Of the 4,033 women sample in this study, the mean age was 37.19 years (SD: 3.264) with 88.8% of advanced maternal age (≥35 years). The mean gestational age for the fetuses was 18.54 weeks by ultrasonography (SD: 1.170). The mean gravidity was 2.40 (SD: 1.123), while the mean parity was 1.09 (SD: 0.814). Out of the total sample, 13.8% had previous remarkable obstetric history, while 86.2% had either no or normal obstetric history. The major indication for genetic amniocentesis was elderly gravida (91.5%) (Table 1).

According to results of genetic amniocentesis, 43 fetuses (1.1%) were found to have Down syndrome and 24 (0.6%) fetuses had chromosome abnormality, while 3,966 (98.3%) were found to have normal chromosome results (Table 2). According to the ultrasound findings, out of 4,033 women, 30 from 43 (69.8%) were suspected to have pregnancies with Down syndrome, and 14 from 24 (58.3%) were suspected to have pregnancies with other chromosomal abnormality, while 1,991 (49.4%) had normal fetus (Table 2). A normal genetic sonogram reduced a woman's risk of having a fetus with Down syndrome, likelihood ratio (LR+) 1.38 [95% CI (1.135, 1.690)] (Table 3).

The overall sensitivity of genetic ultrasound to detect Down syndrome was found to be 69.8%, 95% CI (56.040, 83.495) with a false-positive rate of 50.4% and likelihood ratio (LR+) 1.38 [95% CI (1.135, 1.690)] (Table 3). This infers that the likelihood of finding a fetus with Down syndrome increases by approximately one-fold given the positive test result. The data in Table 3 showed the sensitivity, specificity, false positive rate, and likelihood ratio (LR+) for the detection of Down syndrome based on the presence of various markers. The presence of any isolated marker resulted in sensitivity for the detection of Down syndrome of 23.3% and likelihood ratio (LR+) 0.23, 95% CI (0.135, 0.400) while for any combination of markers, the sensitivity was high at 76.7% and likelihood ratio

(LR+) 0.77, 95% CI (0.651, 0.905).

Isolated markers detected by ultrasound showed much less sensitivity on their own, as compared to the combination of them. Two markers; short femur and short humerus showed sensitivity of 65.1% and 44.2% with likelihood ratios (LR+) 1.39, 95% CI (1.110, 1.729) and 2.22, 95% CI (1.578, 3.126) respectively. Similarly, isolated markers like echogenic intracardiac foci, nuchal fold (≥6 mm), showed 20.9% and 18.6% sensitivity with likelihood ratios (LR+) 3.64, 95% CI (2.009, 6.597) and 0.88, 95% CI (0.469, 1.648). Of all the structural abnormality, of chest, abnormality 2 vessel umbilical cord, facial abnormality including cleft lip and palate, spine and extremities, and abdomen carried the highest likelihood (LR+) of 61.49, 46.12, 46.10, 36.89, and 23.06 respectively. Positive likelihood ratios of other sonographic markers like hyperechogenic bowel, pyelectasis (>3 mm), and hypoplasia of middle phalanx of 5th digit could not be calculated because of absence of fetuses with Down syndrome in these cases.

When the data was analyzed in terms of combination or number of sonographic markers, it was found that the presence of two markers showed the highest sensitivity of 32.6% for detection of Down syndrome in fetuses. Of all other combinations and numbers of markers, markers of three or more carried the highest likelihood ratio (LR+) of 3.82, 95% CI (1.786, 8.155). Absence of any marker or presence of at least one marker showed positive likelihood ratios of 0.46 and 0.89 respectively, which infers less likelihood of Down syndrome in fetuses with either one or absent marker (Table 4).

Table 1. Demographic data of pregnant women received genetic ultrasound performed

	Total (n = 4,033) n (%)
Maternal age, mean ± SD = 37.19±3.264	
<35 years	450 (11.2)
≥35 years	3,583 (88.8)
Gestational age, mean ± SD = 18.54±1.170	
Gravidity, mean ± SD = 2.40±1.123	
1	863 (21.4)
2	1,515 (37.5)
3	1,088 (27.0)
4	567 (14.1)
Parity, mean ± SD = 1.09±0.814	
0	896 (22.2)
1	2,056 (51.0)
2	927 (23.0)
>2	154 (3.8)
Previous obstetrics history	
Remarkable	555 (13.8)
Unremarkable	3,478 (86.2)
Indication for genetic amniocentesis	
Elderly gravida	3,689 (91.5)
Previous down syndrome	141 (3.5)
Other abnormalities	203 (5.0)

Discussion

Many studies to date have advocated the use of prenatal second trimester ultrasound testing as a decisive tool to consider the need for further amniocentesis[12,13]. The results of the present study show a medium accuracy of second trimester genetic sonography for the detection of Down syndrome and chromosomal abnormalities. In the present study a sensitivity of 69.8% and a false-positive rate of 50.4% were obtained for the diagnostic accuracy of second trimester prenatal ultrasound. This is consistent with other studies conducted on the second trimester genetic sonography in which Nyberg et al[14] reported 50.0% sensitivity and 7.0% false-positive rate, Sohl et al[15] reported 67.0% sensitivity and 17.0% false-positive rate, and Nyberg et al[16] reported 68.0% sensitivity and 12.0% false-positive rate. The result of the present study also showed a likelihood of 1.38 for positive test of Down syndrome, which infers that the likelihood of a fetus with Down syndrome increases by approximately one-fold given the positive test result, and finding a positive test for Down syndrome with genetic sonogram increases the probability of actually finding a fetus

Table 2. Results of genetic ultrasound and genetic amniocentesis

	Total cases	Normal n (%)	Abnormal n (%)	Down syndrome n (%)	Other chromosomal abnormalities n (%)
Genetic ultrasound	4,033	1,991(49.4)	2,042 (50.6)	30/43 (69.8)	14/24 (58.3)
Genetic amniocentesis	4,033	3,966 (98.3)	67 (1.7)	43/43 (100.0)	24/24 (100.0)

Table 3. Sensitivity, false positive rate, and likelihood ratio in the prediction of fetal Down syndrome

	Sensitivity (%)	95% CI	Specificity (%)	95% CI	False positive rate	Likelihood ratio (LR+)	95% CI
Genetic ultrasound	0.698 (69.8)	0.560, 0.835	0.496 (49.6)	0.481, 0.512	1-0.496 (50.4)	1.384 (1.38)	1.135, 1.690
Nuchal fold (≥6)	0.186 (18.6)	0.069, 0.302	0.788 (78.8)	0.776, 0.801	1-0.788 (21.2)	0.879 (0.88)	0.469, 1.648
Short femur	0.651 (65.1)	0.509, 0.794	0.531 (53.1)	0.514, 0.546	1-0.531 (46.9)	1.385 (1.39)	1.110, 1.729
Short humerus	0.442 (44.2)	0.293, 0.590	0.801 (80.1)	0.789, 0.813	1-0.801 (16.9)	2.221 (2.22)	1.578, 3.126
Choroid plexus cyst	0.069 (6.9)	-0.006, 0.146	0.975 (97.5)	0.970, 0.980	1-0.975 (2.5)	2.823 (2.82)	0.932, 8.557
Hyperechogenic bowel	0.000 (0.0)	0.000, 0.000	0.995 (99.5)	0.994, 0.998	1-0.995 (0.5)	0.000 (0.00)	0.000, Na N
Pyelectasis (>3 mm)	0.000 (0.0)	0.000, 0.000	0.995 (99.5)	0.993, 0.997	1-0.995 (0.5)	0.000 (0.00)	0.000, Na N
Two vessel umbilical cord	0.023 (2.3)	-0.022, 0.068	0.999 (99.9)	0.999, 1.001	1-0.999 (0.1)	46.12 (46.12)	4.261, 499.071
Wide space between 1st and 2nd toe	0.023 (2.3)	-0.022, 0.068	0.993 (99.3)	0.991, 0.996	1-0.993 (0.7)	3.416 (3.42)	0.475, 24.573
Echogenic intracardiac foci	0.209 (20.9)	0.088, 0.331	0.943 (94.3)	0.935, 0.949	1-0.943 (5.7)	3.640 (3.64)	2.009, 6.597
Absence or hypoplasia of middle phalanx of 5th digit	0.000 (0.0)	0.000, 0.000	0.529 (52.9)	0.513, 0.544	1-0.528 (47.2)	0.000 (0.00)	0.000, Na N
Facial abn.	0.023 (2.3)	-0.022, 0.068	0.999 (99.9)	0.999, 1.001	1-0.999 (0.1)	46.104 (46.10)	4.261, 498.945
Chest abn.	0.047 (4.7)	-0.016, 0.109	0.999 (99.9)	0.998, 1.001	1-0.999 (0.1)	61.488 (61.49)	10.539, 358.753
Spine and extreamity abn.	0.047 (4.7)	-0.016, 0.109	0.999 (99.9)	0.998, 0.999	1-0.999 (0.1)	36.893 (36.89)	7.359, 184.949
Abdomen abn.	0.069 (6.9)	-0.006, 0.146	0.997 (99.7)	0.995, 0.999	1-0.997 (0.3)	23.058 (23.06)	6.747, 78.804
Any isolated markers	0.233 (23.3)	0.106, 0.359	0.000 (0.00)	-0.001, 0.001	1-0.001 (99.9)	0.232 (0.23)	0.135, 0.401
Any combined markers	0.767 (76.7)	0.641, 0.894	0,000 (0.00)	-0.001, 0.001	1-0.001 (99.9)	0.767 (0.77)	0.651, 0.904

Table 4. Sensitivity, false positive rate, and likelihood ratio in the prediction of fetal Down syndrome in isolate and combination markers

Number of markers	Sensitivity (%)	95% CI	Specificity (%)	95% CI	False positive rate	Likelihood ratio (LR+)	95% CI
Zero marker	0.233 (23.3)	0.106, 0.359	0.499 (49.9)	0.483, 0.515	1-0.499 (50.1)	0.464 (0.46)	0.269, 0.799
One marker	0.256 (25.6)	0.125, 0.386	0.714 (71.4)	0.699, 0.728	1-0.714 (28.6)	0.894 (0.89)	0.536, 1.493
Two markers	0.326 (32.6)	0.185, 0.466	0.843 (84.3)	0.832, 0.854	1-0.843 (15.7)	2.075 (2.08)	1.340, 3.211
≥Three markers	0.139 (13.9)	0.036, 0.243	0.963 (96.3)	0.958, 0.969	1-0.963 (3.7)	3.816 (3.82)	1.786, 8.155

with Down syndrome from 46.0% to 87.0%.

On evaluation of the isolated aneuploidy markers, it was found that except for nuchal fold (≥6), short femur, any isolated marker, and any combination of markers, all of the other markers had likelihood ratios of greater than 2. This result is in agreement with a meta-analysis by Smith-Bindman et al[17], which concluded that isolated markers (other than structural abnormalities) cannot significantly predict Down syndrome in fetus.

In the present study, isolated marker of increased nuchal fold thickness of 6 mm or more showed 18.6% sensitivity to detection of Down syndrome with a positive likelihood ratio of 0.88 and false-positive rate of 21.2% Similar results were obtained at Meir Hospital in Tel-aviv, Israel, in which the sensitivity of increased nuchal fold thickness was 23.0% with a false-positive rate of 9.6%[18]. Increased nuchal fold thickness is considered to be one of the most important isolated markers for predicting Down syndrome. Various study results show higher likelihood ratios of a positive test with Down syndrome with this marker; Aagaard-Tillery[19] found a likelihood ratio of 49.04, while studies by Sacco[20], Vintzileos[7], and DeVore[21] found likelihood ratios of 61.27, 43.50, and 19.23 respectively. In the present study a positive likelihood ratio of only 0.88 was obtained indicating a very small likelihood of Down syndrome with presence of nuchal fold marker.

Presence of structural anomalies of chest, 2 vessel umbilical cord, facial abnormality including cleft lip and palate, spine and extremities, and abdomen showed high positive likelihood ratios of 61.49, 46.12, 46.10, 36.89, and 23.06 respectively for the detection of Down syndrome with a positive test result, which is in congruence with results from Aagaard-Tillery[19], DeVore[21], Wax[22] and Weisz[23]. In the present study, isolated markers of short femur and short humerus, both showed high sensitivity of 65.1% and 44.2% with positive likelihood ratios of 1.39 and 2.22 respectively. Similar results were obtained by studies conducted by Weisz[23] and Bromley[24], in which the likelihood ratios for short femur were found to be 1.74 and 1.2 respectively. For short humerus, other studies showed slightly higher likelihood ratios ranging from 1.6 to 5.8[17,24,25]. Four isolated markers, such as chest abnormality, 2 vessel umbilical cord, facial abnormality including cleft lip and palate, spine and extremities, and abdomen showed very high positive likelihood ratios at 61.49, 46.12, 46.10, 36.89, and 23.06 respectively but indicated very low sensitivities at 4.7%, 2.3%, 2.3%, 4.7%, and 6.9% respectively which infers their poor ability to confirm the disorder and thus warrants further investigations when these isolated markers are evaluated for detection of Down syndrome in a fetus.

On evaluation of the number of markers detected and combination of markers analyzed, it was found in this study that the presence of no marker during genetic sonography gave out a positive likelihood ratio of 0.46, which increased to a positive likelihood ratio of 0.89 and 2.08 when one and two markers were identified. The likelihood ratio (LR+) further increased to 3.82 when three or more markers were considered. This shows that the probability of detecting Down syndrome increases with the increase in the number of markers identified during the test. These results were similar to the results obtained by Bromley et al, in which a likelihood ratio of 0.20 was calculated for no marker, which significantly increased to 1.9, 6.2 and 80.0 when the number of markers increased to one, two and three respectively[24].

The findings from this present study show that detailed sonography in the second trimester with normal findings (no markers) can significantly reduce the risk of fetal Down syndrome in high-risk patients, which is consistent with the findings from an 11 year period prospective study by Bromley et al, in which a scan with normal results showed an 80.0% reduction in chances of a fetus with Down syndrome (likelihood ratio: 0.20)[24]. Similarly, Agathokleous et al, concluded that the presence of sonographic markers increase, while absence of sonographic markers decrease the risk for Down syndrome[26]. However, a multicenter prospective study conducted by Smith-Bindman et al, reported in contrast that the negative likelihood ratios were not low enough to decrease the risk of Down syndrome in fetuses[13]. However, Bromley et al disagree and conclude that in experienced hands, normal genetic sonography test without any markers can reduce the probability of Down syndrome by 60.0% to 80.0% in fetuses[24].

Although prenatal second trimester screening with ultrasound may provide conclusive results on the probability of fetus with Down syndrome, the decision on further testing, or further selection of outcome-based options, and the feeling of reassurance, all come down to the pregnant women themselves. It is therefore important to use the findings of the ultrasound to help enable the women to make decisions on the course of action needed. In a similar study conducted at Hospital San Gerardo, Monza, Italy, it was found that second trimester ultrasound findings had an important

reassuring function for women, in which normal sonographic findings affected women's decisions more than abnormal findings[25].

Conclusion

The present study set out to assess the value of second trimester genetic ultrasound for the screening of pregnancies at increased risk of Down syndrome. A rather high accuracy of second trimester genetic sonography was obtained for the detection of Down syndrome. The probability of detecting Down syndrome increases with the increase in the number of markers identified during genetic ultrasound.

Finally, genetic ultrasound in this study probably is an alternative method of prenatal prediction for high risk pregnant women who refuse genetic amniocentesis.

Acknowledgement

The authors wish to thank the patients and the Department of Obstetrics and Gynaecology, Faculty of Medicine, Khon Kaen University for allowing and helping with the research study, and my research team for their guidance and support in the analysis of data. We would also like to thank the Center of Cleft Lip-Cleft Palate and Craniofacial Deformities, Khon Kaen University,in Association with "Tawanchai Project" for support, and Mr. Bryan Roderick Hamman and Mrs. Janice Loewen-Hamman for assistance with the English Language presentation of the manuscript.

Potential conflicts of interest

None.

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คลื่นเสียงความถี่สูงทางพันธุกรรมไตรมาสที่สอง สำหรับการตรวจกรองกลุ่มอาการดาวน์ที่โรงพยาบาลศรีนครินทร์

ธนวรรณ รัตนสิริ, ณวัลย์วงค์ รัตนสิริ, รัตนา คำวิลัยศักดิ์, ปิยะมาศ ศักดิ์ศิริวุฒโฒ

วัตถุประสงค์: เพื่อประเมินคุณค่าของคลื่นเสียงความถี่สูงทางพันธุกรรมไตรมาสที่สองสำหรับการตรวจกรอง กลุ่มอาการดาวน์ที่โรงพยาบาลศรีนครินทร์ จังหวัดขอนแก่น ประเทศไทย

วัสดุและวิธีการ: กลุ่มตัวอย่างได้แก่สตรีตั้งครรภ์ที่มีความเสี่ยงสูงต่อทารกในครรภ์มีโครโมโซมที่ผิดปกติและมีอายุครรภ์ระหว่าง 17-23 สัปดาห์ จำนวน 4,033 ราย ซึ่งได้รับการตรวจกรองกลุ่มอาการดาวน์ โดยคลื่นเสียง ความถี่สูงทางพันธุกรรมไตรมาสที่สอง ก่อนการเจาะน้ำคร่ำ ที่โรงพยาบาลศรีนครินทร์ ในระหว่างเดือนกันยายน พ.ศ. 2539 ถึง เดือนธันวาคม พ.ศ. 2554 ข้อมูลจากเวชระเบียนที่เกี่ยวข้องกับผลการตรวจคลื่นเสียงความถี่สูงทางพันธุกรรมไตรมาสที่สองและการเจาะน้ำคร่ำได้ถูกนำมาทำการศึกษา

ตัววัดที่สำคัญ: ความไวของการตรวจคลื่นเสียงตวามถี่สูงทางพันธุกรรมในการตรวจพบทารกในครรภ์เป็นกลุ่มอาการดาวน์

ผลการศึกษา: สตรีตั้งครรภ์จำนวน 3,966 รายมีผลโครโมโซมของทารกในครรภ์ปกติคิดเป็นร้อยละ 98.3 และมี 43 รายที่ทารกในครรภ์เป็นกลุ่มอาการดาวน์ (ร้อยละ 1.1) และ 24 รายที่ทารกในครรภ์มีโครโมโซมผิดปกติชนิดอื่นๆ (ร้อยละ 0.6) ทารกในครรภ์ที่เป็นกลุ่มอาการดาวน์ 43 รายนี้มี 30 ราย (ร้อยละ 69.8) ที่ผลการตรวจคลื่นเสียงความถี่สูงทางพันธุกรรมไตรมาสที่สองผิดปกติ และพบว่าความไวของการตรวจกรองกลุ่มอาการดาวน์โดยคลื่นเสียงความถี่สูงทางพันธุกรรมไตรมาสที่สองคือ ร้อยละ 69.8 โดยมีอัตราผลบวกลวงร้อยละ 50.4 และความน่าจะเป็นที่ผลการทดสอบเป็นบวก 1.38 กระดูกต้นขาสั้นและกระดูกต้นแขนสั้นมีความไวสูงสุดถึงร้อยละ 65.1 และ 44.2 ตามลำดับ และยังพบว่าความผิดปกติของทรวงอก สายสะดือมี 2 หลอดเลือดและความผิดปกติของใบหน้าซึ่งรวมปากแหว่งและเพดานโหว่ มีความน่าจะเป็นที่ผลการทดสอบเป็นบวกสูงถึงร้อยละ 61.4 ร้อยละ 46.12 และร้อยละ 46.10 ตยมีความไวร้อยละ 4.7 ร้อยละ 2.3 และร้อยละ 2.3 ตามลำดับ

สรุป: ความไวของการตรวจคลื่นเสียงความถี่สูงทางพันธุกรรมไตรมาสที่สอง ในการตรวจพบทารกในครรภ์เป็นกลุ่มอาการดาวน์ที่โรงพยาบาลศรีนครินทร์ มีค่อนข้างสูง และน่าจะเป็นทางเลือกหนึ่งในการทำนายก่อนคลอดกลุ่มอาการดาวน์ในสตรีตั้งครรภ์ที่มีความเสี่ยงสูงแต่ปฏิเสธการเจาะน้ำคร่ำ