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The Performance of First Trimester Anatomy Scan: A Decision Analysis

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Abstract

Introduction—First trimester ultrasound (US) for anatomy assessment may improve anomaly detection but may also increase overall US utilization. We sought to assess the utility of first trimester US for evaluation of fetal anatomy.

Materials and Methods—A decision analytic model was created to compare first plus second trimester anatomy scans to second trimester anatomy scan alone in 4 populations: general, normal weight women, obese women, and diabetics. Probability estimates were obtained from the literature. Outcomes considered were number of: major structural anomalies detected, number of US performed, and false positive US. Multivariable sensitivity analyses were performed to evaluate the consistency of the model with varying assumptions.

Results—A strategy of first trimester US detected the highest number of anomalies but required more US examinations per anomaly detected. The addition of a first trimester anatomy US was associated with a small increase in false positive US (<10/10,000). In populations with higher anomaly prevalence and lower second trimester US sensitivity (i.e. diabetes, obese), the number of additional US performed per anomaly detected with the first trimester US was fewer than 60.

Discussion—In high-risk populations, a first trimester US in addition to a second trimester US may be a beneficial approach to detecting anomalies.

Keywords

Anatomy ultrasound; first trimester; second trimester; decision analysis; sensitivity

Introduction

Ultrasound to evaluate fetal anatomy at 18–22 weeks is now a routine obstetric practice.[1] The timing of this ultrasound is typically chosen in order to balance the ability to visualize structures and complete the scan in one appointment, but this may delay the diagnosis of some anomalies. With advances in technical skill and ultrasound technology, sonography in

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the first trimester to evaluate fetal anatomy has become a feasible option and even routine in some institutions.[2–5] Anatomical surveys can be completed during this time period with the use of transabdominal and transvaginal probes in up to 82% of subjects.[6] A recent systematic review and meta-analysis demonstrated an overall detection rate of fetal anomalies of 51% at 11–14 weeks gestation,[7] although costs and benefits of this practice have not been evaluated.

While it cannot supplant the second-trimester scan, a first trimester ultrasound for anatomy may supplement the mid-trimester scan by allowing identification of key anatomical features early in pregnancy, leaving the second trimester scan to target features that are not yet present or identifiable in the first trimester. This may be particularly beneficial in populations at high risk for fetal anomalies (e.g. pregestational diabetics) or in populations where second trimester ultrasound is technically difficult, such as in obese women.

On the other hand, initiating anatomy scans in the first trimester will necessitate an additional ultrasound visit, expertise, and expense. Unique features of first trimester anatomy compared to second trimester anatomy may be misdiagnosed as a fetal anomaly,[1,8] increasing the chances of a false positive diagnosis. Additionally, since some normal fetal structures (e.g. the cerebellar vermis) are not fully formed until the second trimester, a reassuring first trimester scan will be unable to exclude abnormalities in these structures.

Studies of first trimester anatomy scans have largely focused on the ability to visualize anatomical structures, optimal timing of the scan, optimal scanning technique (transabdominal versus transvaginal) and the sensitivity for anomaly detection.[6,9–22] With mounting evidence that first trimester anatomy scans are technically feasible and have a clinically useful sensitivity, it is important to understand for which populations a first trimester anatomy scan is an appropriate strategy for prenatal diagnosis. Therefore, we sought to assess the utility of first trimester ultrasound, in terms of additional anomalies detected per additional ultrasound.

Materials and Methods

We created a decision analytic model based on a systematic literature review to compare a strategy of first trimester anatomy scan followed by second trimester anatomy scan to a strategy of second trimester anatomy scan alone (Figure 1). Outcomes assessed included the number of major structural fetal anomalies detected (defined as a lethal anomaly or an anomaly requiring surgery; genetic syndromes without structural anomalies were not considered in this analysis), the number of anatomy scans performed, and the number of false positive results. The decision analytic model was used to analyze the utility of first trimester anatomy scan in 4 populations: a general unselected obstetric, normal weight women, obese women, and women with pregestational diabetes.

A structural fetal anomaly was considered detected if it was diagnosed on either the first or second trimester anatomy scan. If no structural fetal anomaly was detected on the first trimester ultrasound, it was assumed the subject would undergo a second trimester anatomy scan. In the model, the second trimester anatomy scan was repeated until a major structural

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fetal anomaly was detected or all components of an anatomy scan were visualized (i.e. a complete anatomy scan), with a maximum of 4 anatomic surveys permitted after the first trimester scan. Given the short time frame in which to perform a first trimester anatomy scan, we assumed that a first trimester anatomy scan would be performed only once.

We conducted a systematic literature review searching the PubMed database of English articles using the MeSH terms “Ultrasonography, Prenatal,” “Prenatal Diagnosis,” “Echocardiography,” “Congenital Abnormalities,” and “Obesity.” These terms were also searched as keywords in PubMed. Articles considered for review were randomized control trials, prospective cohorts, retrospective cohorts, and systematic reviews and meta-analyses that reported the sensitivity and specificity of first and second trimester anatomy scans for detecting major fetal structural anomalies. Point estimates for the incidence of fetal anomalies, test characteristics (sensitivity, specificity), and completion rates for anatomic surveys are shown in Table 1. Studies reporting the sensitivity and specificity of ultrasounds performed for anatomy at 12–15 weeks were considered first trimester and studies reporting the sensitivity of ultrasounds for anatomy at 16–20 weeks were considered for second trimester. Reports of the sensitivity of first trimester ultrasounds for nuchal thickness for the purposes of genetic screening were not considered in the sensitivity and specificity of first trimester anatomy scans.

To address uncertainty regarding several of the baseline assumptions and probability estimates, sensitivity analyses were performed varying estimates of probabilities across their plausible ranges, alone and in combination (Table 1).

All computations were performed using TreeAge Pro Software, 2014, Williamstown, MA. As no human subjects were involved, institutional review board approval was not obtained.

Results

In each of the 4 populations considered (general/unselected, normal weight, obese, and pregestational diabetics), a strategy of first trimester anatomy followed by second trimester scan detected the highest number of anomalies per 10,000 pregnancies (Table 2). However, this strategy was also associated with an increase in the number of anatomy scans performed per anomaly detected as well as small increase in the number of false positive ultrasounds (i.e. report of a major structural anomaly when none existed).

In one-, two-, and three-way sensitivity analyses, across the specified point estimate ranges, the approach of a first trimester US followed by a second trimester US detected the highest number of fetal anomalies. These results were not sensitive to the prevalence of anomalies, the sensitivity and specificity of first trimester anatomy scan, the sensitivity and specificity of second trimester anatomy scan, or completion rates of second trimester anatomy scans.

The number of additional ultrasounds performed for every additional anomaly detected varied based on the prevalence of anomalies (Figure 2). In the lowest risk population considered, normal weight women, an additional 233 anatomy scans were performed for every major structural anomaly detected. Conversely, in the highest risk population, pregestational diabetes, an additional 21 scans were performed for every major structural anomaly detected.

Discussion

Initiating anatomy scans in the first trimester may increase the detection of fetal anomalies; however, in the overall and normal weight populations (≤5% prevalence of fetal anomalies), excessive numbers of additional scans are needed to detect a single additional anomaly. A strategy of first trimester anatomy scans may therefore be most appropriate only in populations at high risk for fetal anomalies (>5% prevalence of fetal anomalies).

We did not assess costs in this study for several reasons. First, as we were considering any congenital anomaly, the lifetime costs of care and quality of life were impossible to accurately estimate in the model, as these vary significantly with the type of defect. Additionally, the rate of termination and the impact of prenatal diagnosis on outcomes also vary significantly with the type of defect. However, the average Medicare National Fee for a level 2 ultrasound is $138 (range $68–257). Thus in the diabetic population, where only an additional 21 ultrasounds are required to diagnose one anomaly, an additional $2,898 would be required to diagnose one additional anomaly. Given that the average hospital cost per child in the first year of life with a birth defect is estimated approximately $78,000 (or six times the cost of a newborn with no birth defect), first trimester ultrasound could lead to significant cost-savings even if only a small percentage of diagnoses led to pregnancy termination.[23] Additionally, prenatal diagnosis may lead to delivery in tertiary care centers, thus improving immediate neonatal outcomes and improving quality of life for affected infants and their parents.

The majority of studies examining the sensitivity of first trimester anatomy scans that we identified were observational cohorts where an attempted anatomy scan was performed in both the first and second trimester.[6,14,17,24–26] Although these studies provide a reasonable assessment of the sensitivity and specificity of first trimester ultrasound, a direct comparison of first and second trimester anatomy scans cannot be reasonably made due to the bias introduced by presumed provider knowledge of the results of the first trimester scan when performing the second trimester scan.

We identified only one randomized control trial of first versus second trimester anatomy scan; in this study, the detection of major fetal anomalies, including heart defects, was not significantly different at the two time points.[27,28] In this trial, subjects assigned to a first trimester anatomy scan did not routinely undergo a second trimester scan. However, most authors recommend performing the second trimester ultrasound even when the first trimester anatomy scan is completed due to the fact that some components of fetal anatomy do not form until the second trimester and this may partly explain the results.

Some limitations of our model must be noted. First of all, we assessed a very broad category of major structural fetal anomalies. Because of this broad designation, we did not consider outcomes such as stillbirth and termination. This would likely have minimal impact on the number of ultrasounds performed per anomaly since the model ended after the diagnosis of

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an anomaly (i.e. once an anomaly was diagnosed, no further anatomy scans would be performed in the model); nevertheless, this did prevent us from considering costs in our analysis, as early termination of pregnancy is less costly than a late termination or a term birth. Secondly, we considered the prevalence of anomalies at birth as this is the statistic most commonly reported in the literature. Due to miscarriages and fetal deaths, the incidence of anomalies at birth may be different from the incidence at the time of first and second trimester scan. Consequently, the first trimester scan may actually diagnose anomalies that would have been missed if only a second trimester scan was performed due to an intervening miscarriage. Also, we did not consider the possible increase in invasive testing that may be associated with an increase in prenatal diagnosis of major structural anomalies. This may increase chorionic villous sampling or amniocentesis uptake depending on the timing of ultrasound and the anomaly diagnosed; increased invasive testing may be associated with increased costs of prenatal diagnosis and increased procedure-related pregnancy losses.

Finally, the sensitivity of first trimester ultrasound for anomalies has not been extensively studied in some populations. As such, we assumed that sensitivity was the same in each population, which may not be true. For example, we assumed that the sensitivity of first trimester ultrasound was the same in obese and in normal weight women. In reality, obesity may result in a decreased sensitivity in the first trimester similar to as in the second trimester. On the other hand, first trimester anatomy scan may still be beneficial in the obese population if a transvaginal probe is used before the fetus and uterus have risen out of the pelvis.

In sum, this decision analysis to compare a strategy of first and second trimester anatomy scan to only second trimester anatomy scan suggests that more anomalies will be diagnosed by initiating anatomy scans in the first trimester. However, in low risk populations, a significant number of additional anatomy scans will be required. As the incidence of anomalies increases, the number of additional anatomy scans per anomaly decreases, assuming that the sensitivity of first trimester anatomy scan is not significantly reduced. Randomized control trials of first trimester anatomy scan in high risk populations, such as obese women and pregestational diabetics, are needed to determine the most effective strategy.

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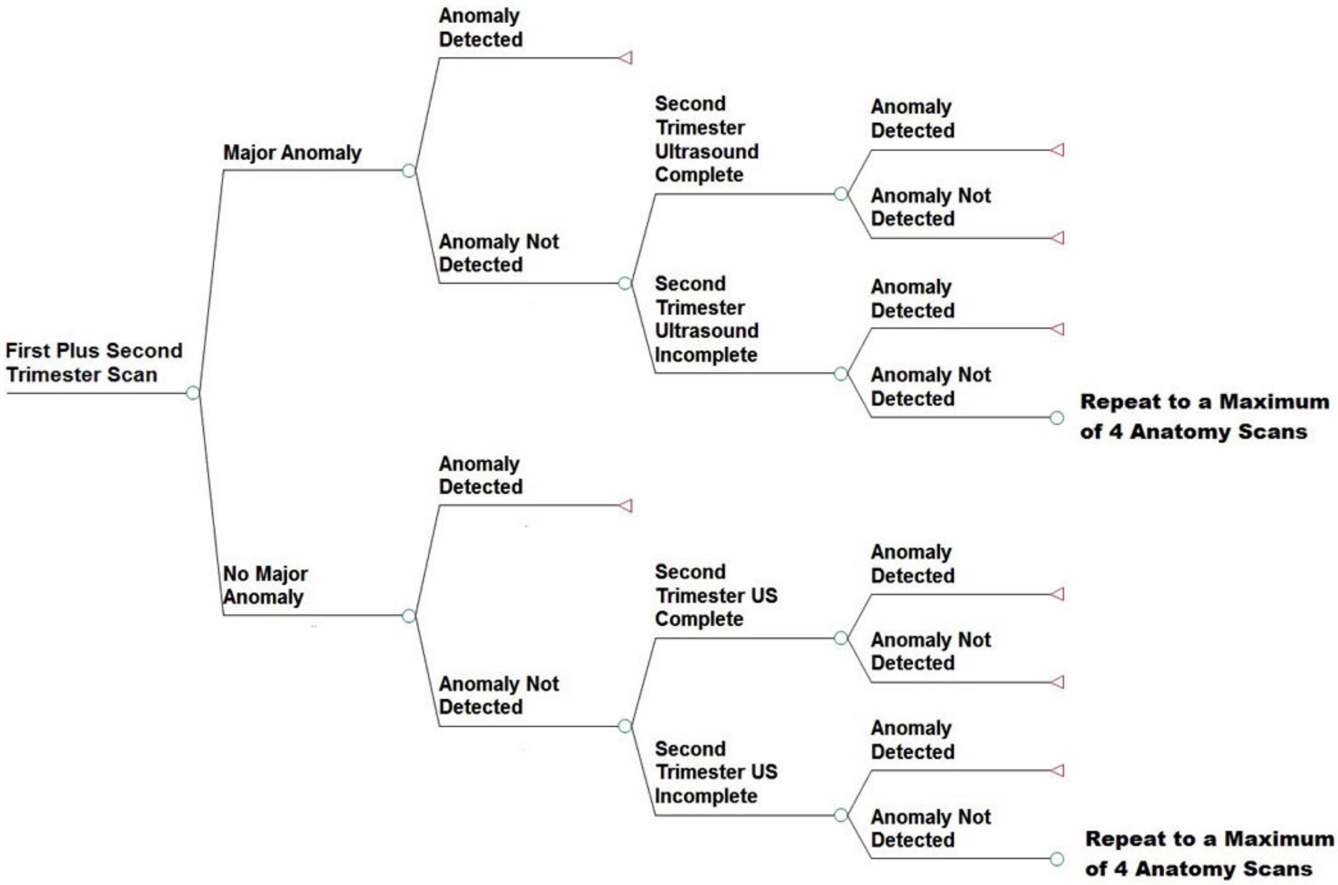

Figure 1.
Decision Analytic Model

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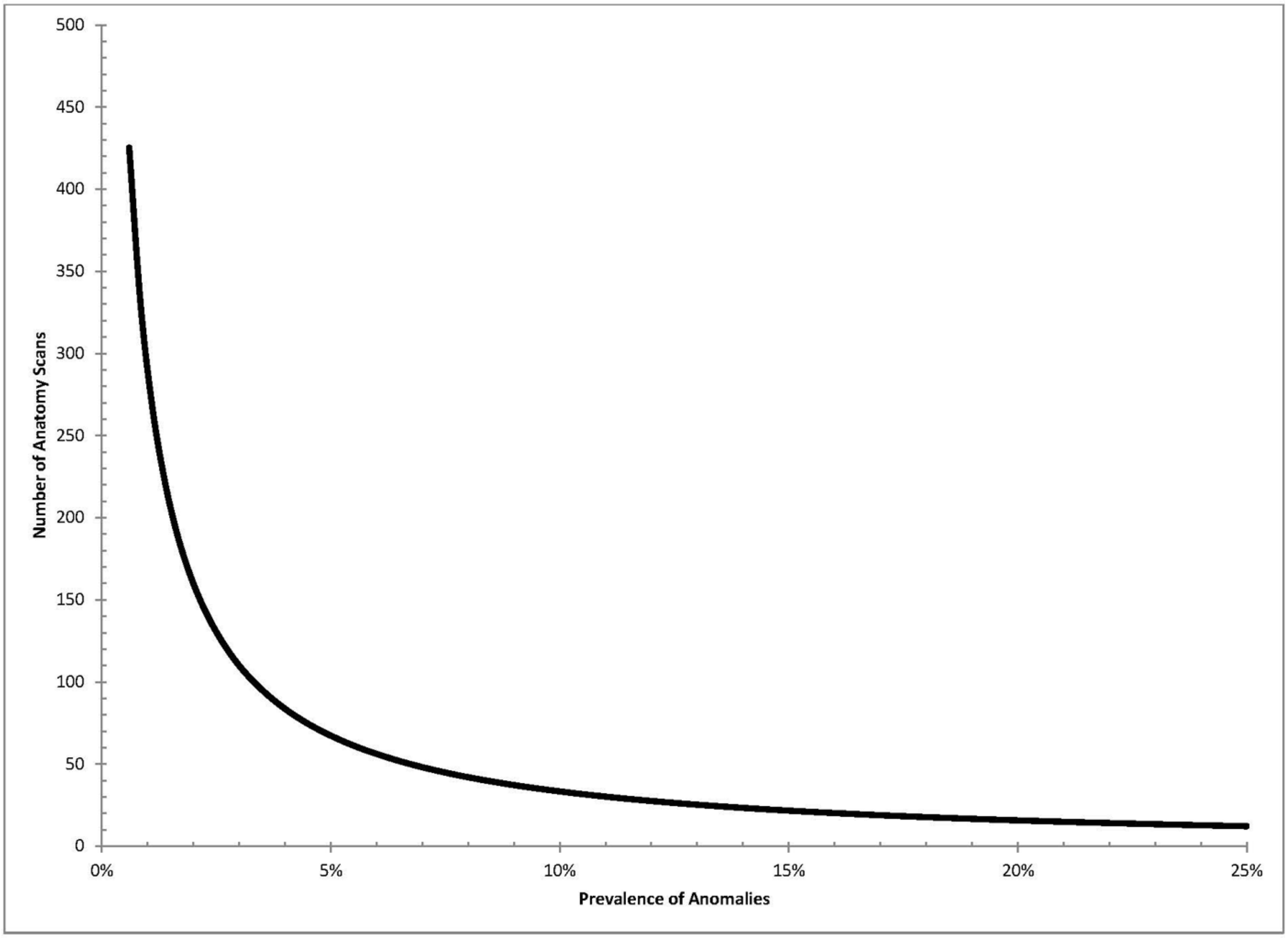

Figure 2.
Number of Anatomy Scans Needed to Detect One Anomaly

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Table 1

	Base Case	Range	Reference
Incidence of Fetal Anomalies			
General	0.03	0.02–0.08	Abu-Rustum[9] Baronciani[29] Becker[20] Callen[1] Carvalho[12] Chen[14] Crane[30] Hildebrand[31] Kontopoulos[3] Levi[32] McAuliffe[33] Novotna[34] Oztekin[35] Papp[36] Saltvedt[27] Whitworth[37]
Normal Weight	0.0209	0.0076–0.0245	Biggio[38] Dashe[39] Whitworth[37]
Obese	0.055	0.055–0.111	Anderson[40] Biggio[38] Callaway[41] Dashe[39] Naeye[42] Moore[43] Quiesser-Luft[44] Stothard[45] Watkins[46] Watkins[47]
Diabetic	0.14	0.14–0.18	Anderson[40] Biggio[38] Chung[48] Dashe[39] Moore[43]
Sensitivity of First Trimester Anatomy			
For All Populations	0.51	0.2–0.837	Abu-Rustum[9] Becker[20] Carvalho[12] Cedegren[13] Chen[14] D'Ottavio[24] Ebrashy[6] Hernadi[49] Ileiscu[15] Lim[17,35] Novotna[34] Oztekin[35] Pilalis[26] Rossi[7] Saltvedt[27] Souka[50] Srisupundit[18] Syngelaki[22] Weiner[51] Whitlow[52]
Specificity of First Trimester Anatomy			
For All Populations	0.9993	0.99–0.999	Abu-Rustum[9] Hernadi[49] Iliescu[15] Souka[53] Weiner[51] Whitlow[52]

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	Base Case	Range	Reference
Sensitivity of Second Trimester Anatomy Scan – Complete			
General Population	0.447	0.15–0.853	Baronciani[29] Best[54] Carvalho[12] Crane[30] Dashe[39] Hildebrand[31] Levi[32] McAuliffe[33] Oztekin[35] Saari-Kamppainen[55] Saltvedt[27] Tabor[56] Whitworth[37] Whitlow[52]
Normal Weight	0.66	0.3–0.97	Best[54] Dashe[39] Hildebrand[31] Tabor[56]
Obese	0.42	0.25–0.48	Best[54] Dashe[39] Hildebrand[31] Tabor[56]
Diabetic	0.38	0.35–0.38	Dashe[39] Miller[57]
Sensitivity of Second Trimester Anatomy Scan - Incomplete			
For All Populations	0.2*Sensitivity of Completed Scan		Local Data
Specificity of Second Trimester Anatomy Scan			
For All Populations	0.999	0.99–0.999	Abu-Rustum[9] Dashe[39] Levi[32] Saari-Kamppainen[55]
Completion of Second Trimester Anatomy Scan – First Attempt			
General Population	0.728	0.672–0.779	Dashe[58] Fuchs[59] Phatak[60] Thornburg[61]
Normal Weight	0.817	0.696–0.905	Dashe[58] Fuchs[59] Phatak[60] Thornburg[61] Tsai[62]
Obese	0.704	0.639–0.763	Chung[63] Dashe[58] Fuchs[59] Tsai[62] Thornburg[61]
Diabetic	0.728	0.672–0.779	Fuchs[59]
Completion of Second Trimester Anatomy Scan – Second Attempt			
General	0.844	0.744–0.917	Fuchs[59]
Normal Weight	0.999	0.715–0.99	Chung[63] Fuchs[59] Tsai[62]
Obese	0.818	0.704–0.902	Chung[63] Fuchs Tsai[62]
Diabetic	0.844	0.744–0.917	Fuchs[59]

	Base Case	**Range**	**Reference**
Completion of Second Trimester Anatomy Scan – Third & Fourth Attempt			
General	0.999	0.99–0.9999	Fuchs[59]
Normal Weight	0.999	0.99–0.9999	Fuchs[59]
Obese	0.999	0.99–0.9999	Fuchs[59]
Diabetic	0.999	0.99–0.9999	Fuchs[59]

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Table 2

Results of Decision Analytic Model, per 10,000 Pregnancies

	Anomalies Detected	**Number of Anatomy Scans**	**False Positive USs**	**Number of Additional Anatomy Scans per Anomaly Detected**
General Population				
First + Second Trimester	240	22,927	20	110
Second Trimester	151	13,132	13	
Normal Weight Population				
First + Second Trimester	194	21,693	18	233
Second Trimester	151	11,825	12	
Obese Population				
First + Second Trimester	532	23,065	134	58
Second Trimester	367	13,443	127	
Diabetic Population				
First + Second Trimester	1002	22,177	17	21
Second Trimester	564	13,104	11	