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Effectiveness of Prenatal Screening Tests on Predicting Cardiac Anomalies

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INTRODUCTION
Congenital heart disease (CHD) is the leading cause of birth defect-associated infant death. With 1 in 111 newborns born with CHD and an elevated infant mortality rate of 30-50%, early detection is imperative. Current prenatal screening mainly focuses on detecting chromosomal anomalies. However, the accuracy of these exams on detecting fetal cardiac abnormalities is not well researched. The primary goal of this study is to compare three prenatal screening tests' potential, i.e anatomy ultrasound, nuchal translucency (NT), and cell-free DNA (cfDNA), in identifying a risk for fetal cardiac anomalies.
METHODS
A retrospective chart review utilized a convenience sample of obstetric patients who were treated at Beaumont Royal Oak Hospital from January 2017 to January 2018 and had completed at least an anatomy ultrasound, NT, or cfDNA. The results of the tests were compared with the newborn's postnatal diagnosis.
RESULTS
A total of 2917 patients completed at least one of the tests – 1793 (61.47%) patients had anatomy ultrasound, 478 (16.39%) had NT, and 646 (22.15%) had cfDNA. 132 (4.53%) of the newborns had cardiac anomalies, 47.73% of which had major defects that require follow-ups or surgeries while 52.27% had minor defects that do not affect quality of life. Sensitivities for ultrasound, NT, and cfDNA were 20.00%, 18.75%, and 5.56%, respectively. Specificities for ultrasound, NT, and cfDNA were 99.7%, 97.97%, and 97.41%, respectively.
CONCLUSION
The study suggests that anatomy ultrasound is still the most accurate in detecting cardiac anomalies. A study limitation is the small sample population because not every patient had an ultrasound, leading to a low ultrasound sensitivity. However, the study demonstrates that NT should be encouraged among physicians because the similar sensitivities of the anatomy ultrasound and NT illustrate that NT can be as important as ultrasound, the current gold standard, when screening for cardiac defects.