

## Current Practice and Knowledge in Aneuploidy Screening: a nation-wide survey

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### ABSTRACT

**Objective:** To determine the current approach and knowledge of aneuploidy screening among obstetricians and gynaecologists in Jamaica.

**Methods:** A questionnaire of primarily multiple-choice questions was sent by mail or hand-delivered to obstetricians and gynecologists across the country. The questions concerned demographics, knowledge of screening and diagnostic tests available and test selection.

**Results:** Sixty-nine out of 100 (69%) doctors responded to the questionnaire; 63/67 (91.3%) reported that less than 25% of their patients had aneuploidy screening (95% confidence interval [CI], 0.85, 0.93) and 13/68 (19.1% [95% CI 0.11, 0.30]) of respondents offered screening to all their patients. The frequency of screening tests was 52.9%, 22.1%, 18.8%, 14.7%, 13.0% and 8.8% for the quadruple screen (QUAD), triple screen, second trimestre ultrasound, first trimestre screen, free fetal DNA and nuchal translucency, respectively. Most physicians answered most of the questions correctly (83.3% correct response rate) and the majority of persons answered at least 5/6 questions correctly.

**Conclusions:** Obstetricians and gynaecologists are aware of aneuploidy risks, screening tests and universal screening recommendations, however only a small fraction of their patients received these tests. This suggests the need for exploration of the barriers to screening and studies on patient's knowledge and views on screening.

**Keywords:** Aneuploidy, Down Syndrome, physician survey, screening

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## INTRODUCTION

Aneuploidy screening has rapidly evolved over the past five decades. It is widely accepted that the risk of Down Syndrome (DS) is 1:15000 in a 20 year-old compared to 1 in 100 in a 40 year old (1). With the observation that biochemical markers or proteins have specific patterns in aneuploidy, combined methods of screening such as the quadruple screen (QUAD) and triple screen were introduced with a view to increasing sensitivity of screening (2). With the increased use of prenatal ultrasound, certain features or markers on ultrasound can be used to modify the aneuploidy risk (3). The detection rate of aneuploidy is variable for the different tests. The use of free fetal DNA in maternal blood (Non-invasive prenatal screening/NIPT) offers the most sensitive and earliest method of screening (4). Universal aneuploidy screening (of all pregnant patients) is well established in many countries and is recommended by authoritative bodies such as the American Congress of Obstetrics and Gynaecology (ACOG) (5) as well as The National Institute for Health Care and Excellence (NICE) (6). Diagnosis with amniocentesis or chorionic villus sampling may be offered where available (7). It is imperative that physicians looking after obstetric patients be aware of aneuploidy risk for pregnancy and be *au fait* with the testing available in their region.

The incidence of Down Syndrome in our population is 1:868 (8). In our obstetric population, it has been observed anecdotally that screening is offered mainly to women age 35 years and older, however there is no data that demonstrates the practice pattern and knowledge. Previous studies examining doctor's knowledge and approach to screening have reported various rates of screening of their obstetric population. Driscoll *et al* found a greater than 95% rate of screening and that practice patterns regarding type of screening test offered varied among physicians (9) while Cleary-Goldman reported a 78% rate of screening in their study (10).

Many obstetricians may use ultrasound as the only screening tool for aneuploidy. Trisomy 18 and 13 have a 90% and 80% detection rate, respectively, on ultrasound as well as increased rate of pregnancy loss and are often detected in the absence of serum screening (11). Trisomy 21, however, has only a 59% detection rate (12) in the absence of major malformations and markers suggestive of Down syndrome are not always present on ultrasound, making this a less than ideal stand-alone screening tool. Down Syndrome has been associated with long-term disability such as developmental delay (13). Therefore, having the diagnosis during pregnancy is imperative for many couples to facilitate future planning for the child or to explore options regarding further pregnancy management.

In Jamaica, the current screening options include first trimestre screen (FTS), Nuchal translucency measurement, Triple screen, Quadruple Screen, second trimestre ultrasound and free fetal DNA (ffDNA). Although screens such as triple screen, nuchal translucency measurement and second trimestre ultrasound are not recommended (14, 15) as stand-alone tests due to lower detection rates, they may likely be more acceptable to patients and clinicians in developing countries due to cost.

## **SUBJECTS AND METHODS**

A cross-sectional study evaluating the practice trends and genetic knowledge base of practising obstetricians and gynecologists in Jamaica was done. A list of all Obstetricians and Gynecologists in Jamaica (approximately 107 at the time, 66 males, (61.2%) and 41 (38.3%) females) was obtained from the local medical fraternity, The Jamaica Association of Obstetricians and Gynaecologists (Grabham Society). The questionnaire was developed, pilot tested and validated by a sample of obstetricians and gynaecologists. The questionnaire contained multiple-choice and true/false questions which addressed

issues including physician aneuploidy screening knowledge and practices as well as demographics. Ethical approval was obtained from the University of the West Indies Ethics Committee.

This was a convenience sample of practising Obstetricians and Gynaecologists (ObGyns) in Jamaica who were willing to voluntarily participate in the study. A total of 100 questionnaires were administered from March to May 2015. The anonymous self-administered questionnaires were hand delivered to each participant. Phone calls for reminder and visits to the physician's practice were carried out for follow-up. Data that were collected by June 2015 were included in the analysis. A total of 69 questionnaires were collected. Obstetricians with sub-specialist training in prenatal diagnosis, such as maternal fetal medicine specialists were excluded. Obstetricians and Gynaecologists that were involved in the validation process were also excluded.

## RESULTS

One hundred questionnaires were distributed and 69 were returned, some of which were partially completed. There were 44/67 (65.7%) males and 23/67 (34.3%) females, which closely represented the group of practising of Obstetricians and Gynaecologists in Jamaica in which there is a male predominance. There were 37/66 (56%) and 29/66 (43.9%) practitioner who belonged to the 25–44 and 45–74-year age groups, respectively. 39/67 (58.2%) had been in practice for less than 10 years while 28/67 (62.3%) practiced for over 20 years. There were 44/68 (63.8%) were in both public hospital and private practice. Regarding the practice setting, 44/67 (65.7%) had a practice situated in the urban area while 8/67 (11.9%) were in the rural area and 15/67 (22.4%) in a town.

All physicians offered some form of aneuploidy screening as no one reported not offering screening at all; 13/68 (18.8%) screened all patients. Advanced maternal age was the most common indication for offering screening (69.6%). There were 63/67 (94.0%, [95% CI 0.85, 0.93]) persons stated that less than 25% of their patients had screening tests done. The QUAD screen was the most common method and the ffDNA was the least common, 10/67 (14.9%, [95% CI 0.07, 0.26]) indicated that they were unfamiliar with the free fetal DNA test while 3/67 (4.3%, [95% CI 0.01, 0.12]) persons reported offering it to all patients, 41/69 (59.4%, [95% CI 0.46, 0.71]) were not offering amniocentesis routinely while others offered it for advanced maternal age 5/67 (7.5%, [95% CI 0.02, 0.17]) and patients thought to be high-risk of genetic abnormalities 21/67 (31.3%, [95% CI 0.21, 0.44]). Most doctors did not personally perform diagnostic tests (58/68 (85.3%).

Category of patients that doctors offered screening to

	<b>Frequency</b>	<b>Per cent</b>	<b>95% Confidence Interval (CI)</b>
All obstetric patients	13	18.8	0.11-0.30
Patients of advanced maternal age	50	73.5	0.62-0.83
Patients that have a significant family or medical history	24	35.3	0.25-0.47
Patients that initiated the discussion	17	25.0	0.16-0.36
Patients with abnormal ultrasound findings	20	29.4	0.19-0.41

Age, gender, experience and practice setting were analysed as possible factors affecting screening, with varied significance noted. The group in practice for over 20 years had a larger proportion of patients screened (50% of physicians reported a 25–50%

screening) compared to those in practice for less than three years (25% doctors had less than 25% screening rate,  $p = 0.41$ ). However, of note, the more experienced group was a smaller number of doctors ( $n = 12$ ). About 1.5% of those in practice for over 20 years said that ffDNA was their first choice compared to 0% of those in practice for less than five years ( $p = 0.028$ ), indicating that older physicians were more likely to offer this test. In the female physician group, 33.9% reported that less than 25% of their patients had undergone screening. In the male group, 66% reported that  $< 25\%$  of their patients had received screening compared to 66% in the female group ( $p = 0.715$ ). Therefore, female physicians had more of their patients screened. Those in the rural setting reported that a smaller number of persons received screening; 87.5% of physicians in this setting reported that less than 25% of their patients had screening compared to 66.1% in the urban group ( $p = 0.690$ ). Only 1.6% of the private practice group reported less than 25% of their patients were screened while in the hospital-setting-only group, none of them reported that 25–50% received screening ( $p = 0.040$ ). Therefore, those in a group private practice had more patients screened, 37.5% of those in solo private practice perform diagnostic tests on singleton pregnancies as opposed to 62.5% in both hospital and private and 0% in hospital practice only ( $p = 0.00$ ). The younger physicians appeared to be offering more diagnostic testing, 34% of those in practice for less than three years compared to 14% of those in practice for over 20 years ( $p = 0.024$ ). Diagnostic testing approach also differed according to practice setting; 79.2% in the urban areas responded positively to offering diagnostic testing as opposed to 0% in the rural area ( $p = 0.049$ ).

Most of the physicians answered most of the questions correctly as the majority of persons, 83.3%, answered at least 5/6 correctly. Varied experience with aneuploidy pregnancies was noted. The majority of physicians (51/69; 73.9%) reported having managed less than 10 cases of aneuploidy. Most physicians reported feeling somewhat

qualified 46/69 (66%, 95% CI 0.54, 0.78) in counselling patients on genetics issues while the remainder felt well-qualified 11/69 (15.9%, 95% CI, 0.08, 0.27) or not qualified (7/69 [10.1%, 95% CI 0.04, 0.19]). There were 64/69 (92.7%) responded positively to using a screening and diagnostic counselling service if it were available. Most physicians used American College of Obstetricians and Gynecologists (ACOG) guidelines (46/69, 66.6%) and Royal College guidelines (45/69, 65.2%) to keep up-to-date on information in their field.

## DISCUSSION

The principal findings from this study is that most patients seen in obstetric practice in Jamaica do not receive screening for aneuploidy even though the doctors are aware of the available tests and have sufficient knowledge in aneuploidy screening and risk. Only 13/68 (18.8% 95% CI 0.11, 0.30) offered screening to all obstetric patients and most did not offer routine diagnostic tests. The most common test offered to patients is the QUAD screen while the least commonly offered is ffDNA.

In a similar study (10), only < 1 % of the participants reported not offering any kind of screening at all, similar to 0% in our study, 113/652 (17%) did not offer screening routinely but offered to 92% of those patients of advanced maternal age. Similarly, 69.6% of our respondents offered screening to patients of advanced maternal age. Cleary-Goldman *et al* (10) also found that 49% offered QUAD screen compared to 52.1% in our study. It was expected that a larger proportion of our physicians would offer the QUAD screen as it is more readily available and familiar to most doctors. There is also limited use of FTS possibly due to lack of availability of Chorionic Villus Sampling to confirm a diagnosis or reassure parents in the event of a positive FTS as well as late booking.

More than three-quarters of the physicians (78%) in a previous study (10) routinely offered aneuploidy screening to all obstetric patients compared to 18.8% in our study. Driscoll (9) *et al* found that 95 % of their population offered aneuploidy screening to all obstetric patients. This is not surprising as they have established guidelines and screening tests are widely available in their country as opposed to ours (7). The ffDNA test was offered by 11.6% of physicians and 10 people reported being unfamiliar with this test. This is not surprising as this was a new test at the time of the study.

There was a good response rate to questions concerning aneuploidy risk assessment of patients. Approximately 87% of respondents in a previous study (9) indicated that maternal age of 40 years or more placed women at a 1% or greater risk for trisomy 21 in the second trimestre, compared to 53.6% in our study. More of the physicians (48%) in a similar study performed their own invasive procedures (16) compared to 14.4% in our study. The physicians had indicated that they refer patients to a high-risk setting (most frequently the University Hospital of the West Indies) for invasive tests.

The strength of this study is that it is a nation-wide survey of Obstetricians and Gynaecologists caring for different categories of patients, both from rural and urban areas, public and private practice. Therefore, the socio-economic and educational background of the patients that these doctors looked after would have varied, illustrating that the rural physicians had less patients being screened. It also demonstrates that barriers to screening are likely not due to lack of knowledge on the part of the physicians as there was an acceptable correct response rate to the questions asked. It therefore, prompts us to explore what the barriers to offering universal screening of our obstetric population may be and to determine if patients and care-providers believe that this is of any significance to them. This provides some evidence to explore the establishment of local and/or institution



guidelines on the offering of aneuploidy screening to patients. Additionally, it demonstrates that there is a role for revising the level of counselling that is offered to patients.

This study is not able to address the reasons why women are not screened, which is another pertinent question to answer. Questions concerning the obstacles to screening could have been posed. One possible explanation may be the perception that women will not take-up screening due to cost factors as well as cultural or social factors. Also, patients may feel that if a chromosomal anomaly is found, it is not treatable therefore, screening and diagnosis is not helpful. Most women may not entertain the option of termination if chromosomal abnormality is found and as such opt not to screen. Another drawback is that practice may vary according to offering termination of pregnancy based on our laws against abortion except under certain circumstances. Time constraints on the part of physicians for providing the appropriate counselling for genetic conditions as well as lack of comfort in discussing these issues could also be contributing factors.

## **CONCLUSION**

This study suggests that there is little knowledge deficit, yet universal screening is not the norm. Another study to survey patient's views on aneuploidy screening would be helpful to determine the uptake of screening by patients. It also needs to be determined how to translate knowledge into a change in attitude and practice. Barriers to offering screening may be different from barriers to uptake.

## REFERENCES

1. Morris JK, Wald NJ, Mutton DE, Alberman E. Comparison of models of maternal age-specific risk for Down syndrome live births. *Prenat Diagn.* 2003;23(3):252-258. doi:10.1002/pd.568.
2. Merkatz IR, Nitowsky HM, Macri JN, Johnson WE. An association between low maternal serum alpha-fetoprotein and fetal chromosomal abnormalities. *Am J Obstet Gynecol.* 1984;148(7):886-894. doi:0002-9378(84)90530-1 [pii].
3. Aagaard-Tillery KM, Malone FD, Nyberg DA, et al. Role of Second-Trimester Genetic Sonography After Down Syndrome Screening. *Obstet Gynecol.* 2009;114(6):1189-1196. doi:10.1097/AOG.0b013e3181c15064.
4. Mercer BM. Practice Bulletin No. 163: Screening for Fetal Aneuploidy. *Obstet Gynecol.* 2016:1-15. doi:10.1097/AOG.0000000000001406.
5. Mercer BM. Practice Bulletin No. 163: Screening for Fetal Aneuploidy. *Obstet Gynecol.* 2016:1-15. <http://www.ncbi.nlm.nih.gov/pubmed/26938574>.
6. Consultation DFOR. Antenatal Care Routine Care for the healthy pregnant woman NICE guideline. *Heal (San Fr.* 2007;(June):1-46. <http://scholar.google.com/scholar?hl=en&btnG=Search&q=intitle:Antenatal+care:+routine+care+for+the+healthy+pregnant+woman#1>.
7. Bahado-Singh R, Driscoll D. ACOG Practice Bulletin No. 77: screening for fetal chromosomal abnormalities. *Obstet Gynecol.* 2007;109(1):217-227. <http://www.ncbi.nlm.nih.gov/pubmed/17197615>.
8. Scott C, Thame M. The incidence of cardiac lesions among children with Down's Syndrome in Jamaica A Prospective Study. *West Indian Med J.* 2014;63(7):693-697.
9. Driscoll DA, Morgan MA, Schulkin J. Screening for Down syndrome: changing practice of obstetricians. *Am J Obstet Gynecol.* 2009;200(4). doi:10.1016/j.ajog.2008.12.027.
10. Cleary-goldman J, Morgan MA, Robinson JN. Screening for Down Syndrome Practice Patterns and Knowledge of Obstetricians and Gynecologists. *Obstet Gynecol.* 2006;107(1):11-17. doi:10.1097/01.AOG.0000190215.67096.90.
11. Lai S, Lau WL, Leung WC, Lai FK, Chin R. Is ultrasound alone enough for prenatal screening of trisomy 18? A single centre experience in 69 cases over 10 years. *Prenat Diagn.* 2010. doi:10.1002/pd.2623.
12. Malone FD, Canick JA, Ball RH, et al. First-Trimester or Second-Trimester Screening, or Both, for Down Syndrome. *Obstet Gynecol Surv.* 2006;61(4):226-228. doi:10.1097/01.ogx.0000209667.65566.55.

13. Dykens E, Hodapp R, Evans D. Profiles and development of adaptive behavior in children with Down syndrome. *Down Syndr Res Pract.* 2006. doi:10.3104/reprints.293.
14. Chitayat D, Langlois S, Wilson RD. Prenatal screening for fetal aneuploidy in singleton pregnancies. *J Obstet Gynaecol Can.* 2011;33(7):736-750.
15. Benn P, Borell A, Chiu R, Cuckle H, Dugoff L, Faas B. Position Statement from the Aneuploidy Screening Committee on Behalf of the Board of the International Society for Prenatal Diagnosis , April 2013. *Prenat Diagn.* 2013;(April):1-17. doi:10.1002/pd.4139.
16. Wilkins-Haug L, Hill L, Schmidt. Holzman GB, Schulkin J. Genetics in Obstetricians' offices: A survey study. *Obstetrics and Gynaecology* 1999 May; 93(5 Pt. 1): 642-7.

**Synopsis:** Aneuploidy risk is present in all pregnancies, however many women in Jamaica are not screened. This may be due to concerns of the patient or physician. Increase in screening may allow for early management and preparation for abnormalities.

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