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Prenatal screening for Down syndrome: a survey of health care professionals' opinions and practice

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Abstract

Background: **Down Syndrome (DS) is a common genetic disorder that is associated with high intrauterine lethality. Morbidity for the survivors includes congenital anomalies and Intellectual Disability (ID). Genetic screening for DS is an ever evolving field with remarkable progress made over the years. Health care workers remain the main source of information to patients on DS screening. In the absence of a national policy on screening, there is bound to be disparity in practice both at individual and institutional levels.**

Objective: **To assess the opinions and practice on DS screening amongst health care professionals attending the 2012 annual Kenya Obstetrical and Gynaecological Society (KOGS) Coast symposium held on 27-28th October 2012.**

Methods: **A semi structured questionnaire was administered to the study participants during a plenary session of the conference. Different questions assessing opinion and practice of antenatal screening for DS were then summarized by health worker designation through percentages and comparisons assessed. Qualitative analysis was done by coding and tabulating the emerging themes of concern**

Results: **There was a response rate of 47% amongst the symposium participants. Eighty eight percent indicated that DS screening should be part of routine antenatal care, though the service is offered only rarely by a majority. Knowledge of the various DS screening and diagnostic tests was generally low. Reasons given for not offering DS screening included lack of facilities, low awareness and training amongst health care workers and the high costs involved.**

Conclusion: **Whereas a majority of the study participants were of the view that DS screening should be routinely offered, this is only rarely done. Intended programmes and protocols for prenatal screening for DS should take into consideration the concerns highlighted by the study. There is also need for continued professional training on DS screening.**

Introduction

Down Syndrome (DS) is the most common viable aneuploidy and the leading genetic cause of Intellectual Disability (ID) (1). The condition is found in 1 in 800 - 1000 newborns. In approximately 95% of cases, the syndrome is caused by trisomy due to nondisjunction of chromosome 21. The rest results from mosaicism or translocation (2).

Of the affected fetuses, 30% die between the 12th and 40th week of pregnancy. For the live newborns congenital heart defects remain the major cause of early mortality. Cohort studies indicate improvement in survival rates over the past 50 years with 85% surviving into their 50s (3-5). This is due to improvements in cardiac surgery and general health management. Planning for the future care of adults with ID remains a challenge (6). The health and economic burden of DS is no doubt substantial for the family of a person with DS, as well as to society and the improved longevity will impact on social security (3,7). These factors must be clearly laid out during genetic counselling.

Until the 1980s, patients were selected for diagnostic tests for DS based on maternal age. There has since been tremendous advancement in screening. Screening tests can now be done in the first and second trimester and may include a combination of ultrasound and biochemical tests. There is currently a lot of research ongoing on cell-free fetal DNA (cffDNA) in maternal circulation. CffDNA is likely to be adopted in future either as a second tier screening test or as a non invasive prenatal diagnostic test for DS, different from the now well established and validated analyte/ultrasound approach (8).

Consent for screening is only valid once the patient is fully informed (9). A number of studies have consistently found patients to be deficient in their knowledge and understanding of genetic screening tests (10-12). The American College of Obstetrician and Gynaecologists (ACOG) thus recommends that patients should be provided with information about the detection and false-positive rates, advantages, disadvantages, and limitations, as well as the risks and benefits of diagnostic procedures so that they can make informed decisions on Down syndrome screening (13).

Health care providers are often the major source of information to patients on prenatal screening tests (14). There is currently no national guideline on DS screening in Kenya and thus it is likely that there is variation in practice between individual clinicians and institutions. There is also a disparity in availability of trained health

care personnel and equipment to actualize effective screening. For instance, competence in anomaly or nuchal translucency ultrasound is not widespread.

Many studies have also demonstrated gaps in the knowledge of health care professionals on the details and meaning of various screening and diagnostic tests (15-18). None of these are local studies. There is also the sensitive issue of the ethical, social and/or cultural views and beliefs of the health care provider at times creating dissonance (18). This study documents the experiences, knowledge and attitudes of a sample of health care workers on DS screening that may lay the basis for further research or policy formulation.

The objective of this study was to assess the opinions and practice on DS screening amongst health care professionals attending the 2012 annual Kenya Obstetrical and Gynaecological Society (KOGS) Coast symposium held on 27-28th October 2012.

Materials and methods

Study design: This was a cross sectional study conducted on 27th October 2012 at the third annual KOGS Coast branch symposium held in Mombasa, Kenya. Authorization to conduct the study was given by the symposium's organizing committee. Approval was also granted by the Aga Khan University's Research and Ethics Committees.

Participants: The participants were mainly health care professionals drawn from the locality. Those eligible to participate in the study were health care workers involved in the provision of antenatal care of such cadres as obstetricians, obstetricians in training (residents), medical officers and nurses/midwives.

Data collection: For this study, a self administered, semi structured questionnaire was distributed to the study participants during a plenary session of the symposium by the principal investigator and an assistant. The questionnaire was to be filled anonymously. It captured the cadre of the health care professionals as well as their opinions and practice, knowledge and views on DS screening.

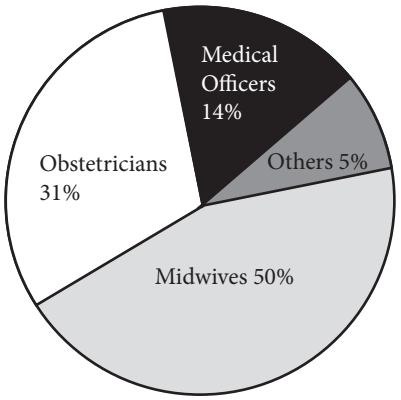
Data analysis: Quantitative data were managed and analyzed via Microsoft® Office Excel® 2007. Different questions assessing opinion and practice of antenatal screening for DS were then summarized by health workers designation through percentages and comparisons assessed. Qualitative analysis was done by coding and tabulating the emerging themes of concern.

Results

Of the seventy nine questionnaires distributed to the study participants during the symposium, thirty seven were returned (47% response rate). One was excluded from analysis as the designation of the health care worker was not indicated.

The final study sample of thirty six respondents consisted of eleven obstetricians, eighteen midwives five medical officers, a trainee obstetrician and one paediatrician. Their distribution was as shown in Figure 1.

Figure 1: Cadres of health care workers



In this survey, 88% of the respondents agreed that DS screening should be offered to pregnant women as part of routine antenatal care. However, none of the respondents indicated the presence of a universal screening policy in their hospital or practice. A majority of respondents indicated that they rarely offer DS screening (58%). Fourteen respondents (39%) do not offer DS screening at all. Where DS screening is offered, 80% indicated advanced maternal age as the commonest criteria for selecting women for screening. Nuchal Translucency (NT) ultrasound was the commonest screening modality for these respondents.

Of the practitioners offering DS screening, only 21% proceed to diagnostic testing for screen positive cases, the majority proceeding to counsel such patients on either preparation for a special needs child or termination of pregnancy. Amniocentesis is the diagnostic test of choice for all, with none picking chorionic villous sampling. Seventy percent of the respondents were in agreement that the option for pregnancy termination should be included during the counselling process in women undergoing DS screening.

The most recurrent themes on why DS screening is rarely offered included lack of facilities, low awareness and training amongst health care workers and the high cost of screening. Other reasons stated included the lack of policy on DS screening as well as existence of competing priorities in maternal health care.

Table1: Knowledge of DS screening

Question	% Right response	% Not sure
Risk of DS decreases with advancing gestational age	11	6
Low PAPP-A and elevated β HCG is diagnostic of DS	19	72
CVS is best done before 11 weeks gestation	6	47
Cystic hygroma at 12 weeks may be found in DS	39	53
NT ultrasound is best offered in the first trimester	44%	44%

The questionnaire also contained five questions designed to test the knowledge of the health care practitioners on the various DS screening and diagnostic tests. The results of this are summarized in the Table 1.

Discussion

The study shows support amongst the health care providers interviewed on the principle of DS screening. However, screening is only rarely offered by the majority. Different reasons were given for the lack of screening, as highlighted above. However, the study shows that there is a general lack of knowledge on DS screening tests, including differentiation between screening and diagnostic tests. This is consistent with other studies which have highlighted gaps in the knowledge of health care providers on DS screening (15). Any strategy, therefore, aimed at increasing the uptake of DS screening must incorporate training of staff, both in the theoretical and practical aspects of screening.

This survey indicates that setting up an effective DS screening service is likely to be expensive from the reasons indicated for not screening. Institutions should be encouraged to formulate innovative protocols for screening and consider pooling of resources within localities e.g. ultrasound machines, biochemistry labs and personnel such as ultrasonographers or radiologists.

A majority of respondents indicated advanced maternal age as the trigger to initiate DS screening. Whereas the risk of aneuploidy increases with advanced maternal age, those getting pregnant beyond the age of 35 years represent only a minority of the pregnant population. The sensitivity of such an approach for screening is therefore likely to be low with little impact on the prevalence of DS. A universal screening policy might be ideal, after due consideration of financial, staffing and infrastructure limitations.

Training and capacity building will also offer more and better choices to women undergoing DS screening. In this study, only amniocentesis was mentioned as a diagnostic test available to practitioners, whereas chorionic villous sampling, if available would offer an earlier first trimester diagnostic test to those who may wish for this. Those offering DS screening indicated NT ultrasound as the modality of choice. This might indicate that women presenting for their booking visit beyond the first trimester might have a request for 'routine' obstetric ultrasound, without the realization that this is a screening anomaly scan. It is therefore, only proper that women are appropriately counselled and offered choices before such investigations.

Limitations of this survey include the small number of respondents and the fact that many could be working in the same hospital or practice. However, this survey should form a basis for further exploration of this subject in the country with a view of coming up with a uniform policy and strategy for DS screening.

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