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COLLEGE OF OBSTETRICIANS AND GYNAECOLOGISTS, SINGAPORE

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RECOMMENDED 'BEST PRACTICE' GUIDELINES ON ANTENATAL SCREENING FOR TRISOMY 21 (DOWN'S SYNDROME) AND OTHER FETAL ANEUPLOIDY

Statement

These guidelines are based on the data currently available on the performance of the screening tests. It is acknowledged that new data on screening tests are being produced as programs continually audit their performance, and these may alter the recommendations below. It is recognised that not all women will want to use a pre-natal screening or diagnostic test. It is also recognised that in Singapore some of the other tests mentioned in other guidelines may not be available.

1 All women, regardless of age, should be considered to be at risk for aneuploidy and should be offered screening for Down Syndrome (DS). All women should be made aware of the availability of screening tests for Down syndrome and other chromosomal abnormalities. This should include Nuchal Translucency Screening (NTS) combined with first trimester maternal serum screening (also known as the first trimester combined screening) or NTS combined with second trimester maternal serum testing (also known as step-wise sequential screening). Combinations of ultrasound and biochemical screening tests given as a single result have higher detection rates than either test used alone. The combined first trimester screening and the step-wise sequential screening test are available in Singapore.

2 Maternal age alone is a poor screening test for aneuploidy and should be removed as the sole indication for invasive testing.

3 The advantages and disadvantages of first trimester versus second trimester screening should be discussed with the woman or couple.

4 Nuchal translucency scanning should only be performed by trained operators and should be done when the fetus has a CRL of 45 to 84 mm, which corresponds to 11 weeks and 0 days to 13 weeks and 6 days.

5 All women who are referred for second trimester serum screening should have had their gestational age determined by an early ultrasound dating scan (or a known conception date). In the event of any uncertainty about gestational age, a pre-test ultrasound examination should be done.

6 The second trimester serum screen should usually be done between 15 and 18 weeks gestation and a risk-assessment performed incorporating maternal age using a software package developed for that purpose. Some programs offer a wider gestational age range or incorporate different parameters, such as maternal weight, in their risk calculation and referring practitioners should be aware of the laboratory's guidelines.

7 Soft markers (with a high likelihood ratio like absent nasal bone or thickened nuchal skin fold) discovered by a trained ultrasonographer during the 18- to 22-week ultrasound scan can be used to modify the risk of aneuploidy established by prior robust screening (see statement No.1). Likewise, in the absence of ultrasound soft markers or anomalies, performed by a trained ultrasonographer, the Down Syndrome risk is halved.

8 Sequential independent testing (i.e. NTS followed by second trimester serum screening, with independent results given after each test) is not currently recommended as a population screening method. It should be recognised that if a woman chooses to have a number of independent screening tests, it will decrease the sensitivity of the tests and increase her chance of getting a false-positive result. A sequential step-wise approach where the final risk estimate incorporates first and second trimester results is a better option.

9 All women who choose to have an antenatal screening test for DS and other fetal aneuploidy should have appropriate pre-test counselling, including the provision of written information to ensure they have a good understanding of the test and its accuracy. All women receiving a high-risk test result should be provided with adequate post-test counselling. An abnormal screening test result should be dealt with as an urgent clinical problem requiring early counseling and definitive diagnosis.

10 All providers of biochemical and ultrasound-based screening tests for DS and other fetal aneuploidy should maintain comprehensive records. Regular audit of their screening practice is encouraged to provide the users with accurate information on the detection rate and the screen positive rate.

11 Appropriate information and education should be offered based on an assessment of the woman's understanding of the screening methods and associated procedures.

12. Chorionic Villous Sampling (CVS) or amniocentesis should be available to women who are not sufficiently confident to proceed with combined first trimester screening or second trimester maternal screening despite receiving full information about such screening and procedure related fetal loss.

13 The data for first Trimester Combined screening or second trimester screening in multiple pregnancies is limited and the counselling for such pregnancies is complex. However, it is perfectly reasonable to offer it to monochorionic twin pregnancies. NT screening alone is suitable for dichorionic twin pregnancies or higher-order pregnancies.

Acknowledgements

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The panel reviewed recommendations from the American College of Obstetricians & Gynaecologists, The Royal College of Obstetricians & Gynaecologists, UK, the Royal Australian and New Zealand College of Obstetricians and Gynaecologists and the Society of the Obstetricians and Gynaecologists of Canada.

These were noted to be fairly comprehensive and the panel felt that there was no advantage in attempting to start the process again. The panel therefore has adopted large components of the recommendations from these bodies and we acknowledge their contribution to our recommendations.

1. Joint HGSA/RANZCOG Prenatal Diagnosis and Screening Committee recommended 'Best Practice' Guidelines on antenatal screening for Down Syndrome and other fetal aneuploidy
2. NHS, National Screening Committee policy - Down's Syndrome screening, Compiled by the National Screening Committee, July 2006
3. Antenatal Screening for Down Syndrome, RCOG, July 2003
4. Bulletins ACOG. ACOG Practice Bulletin clinical management guidelines for obstetrician–gynecologists No. 77: screening for fetal chromosomal abnormalities. Obstetrics & Gynecology 2007;109(1):217-27.
5. Summers AM, Langlois S, Wyatt P, Wilson RD, Society of O, Gynaecologists of C Clinical Practice Guideline. Prenatal screening for fetal aneuploidy. Journal of Obstetrics & Gynaecology Canada: JOGC 2007;29(2):146-79.

This consensus statement is produced on behalf of the College of Obstetricians and Gynaecologists, Singapore by:

Dr Lai Fon-Min, Dr George Yeo Seow Heong, Dr Arijit Biswas, Dr Mary Rauff, Dr Yong Tze Tein, Dr Chang Tou Choong, Dr Ann Tan Sian Ann, Dr Tan Hak Koon.

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Valid until March 2011
unless otherwise indicated

FREQUENTLY ASKED QUESTIONS (FAQS)

S/N	Question	Proposed Response
1	I am currently offering NT + serum screening to my patients less than 35 years old and amniocentesis to those over 35 years old. If I offer universal screening to all my patients regardless of age, will I be medico-legally liable if I offer screening to a 38 year old lady, whose screen result is low risk, but eventually delivers a baby with Down's syndrome?	No, you are not liable so long as your patient has been counselled as to the accuracy of the tests offered.
2	I am worried about the type of screening test I should offer to my patient. Is maternal serum screening alone sufficient?	Maternal serum screening alone (triple test) is an appropriate form of screening if the patient presents later than 13 weeks and 6 days. However, the patient should be counseled regarding the detection rate of the second trimester serum screening (triple test) with is around 60%.
3	If I offer maternal serum screening alone and the patient is screen negative but subsequently delivers a baby with Down's syndrome, am I medico-legally liable as I have not offered a test with a higher detection rate?	If the patient presents later than 13 weeks 6 days gestation, second trimester maternal serum screening (preferably using triple test) is appropriate. If a patient presents in the first trimester, there should be a discussion of the choice of screening available and the detection rates given a fixed test positive rate. The approximate detection rates for first trimester combined screening, NT ultrasound alone and second trimester serum screening are 90%, 80% and 60% respectively, for a test positive rate of 5%. The patient may choose to opt for one of the above screening methods.
4	Can a NT scan followed by second trimester serum screening be as acceptable as first trimester combined screening?	Yes, if the NT is measured appropriately and recognized software is used to calculate the likelihood ratio. The second trimester serum screening result can then be integrated with the likelihood ratio derived earlier to obtain the final risk (step-wise sequential screening)
5	Can I offer NT ultrasound scan as the only screening test?	Yes, it is possible but the patient needs to be informed that detection rate of the test would only be 80%. It would be better accompanied by a blood test as this would increase the detection rate to 90%.
6	If I offer amniocentesis to all women above 35 years of age, will I be medico-legally liable (in view of the COGS guidelines) if the patient suffers a procedure-related miscarriage and claims that I did not offer screening in the first place?	Yes, you could be potentially liable if you have not provided your patient with information on all the available screening methods for her to make an informed choice.
7	Do I have to be accredited to do NT scans and if so by whom?	The guideline states that "Nuchal translucency scanning should only be performed by trained operators and should be done when the fetus has a CRL of 45 to 84 mm, which corresponds to 11 weeks and 0 days to 13 weeks and 6 days." There is ample evidence that good quality control of the scan which impacts greatly on the results can only be achieved by training. The Fetal Medicine Foundation, which provides the software to calculate the risk, runs the accreditation programme to ensure a high degree of quality control. The screening programme results are optimal when a risk-based cut-off is used rather than absolute NT measurements.