Practice Guidelines for Health Care Providers Involved in Prenatal Screening and Diagnosis

These guidelines were prepared in consultation with the Canadian College of Medical Geneticists, the Canadian Association of Genetic Counselors, the Canadian Nurses Association, the College & Family Physicians of Canada and the Genetics Committee of the Society of Obstetricians and Gynaecologists of Canada.

SOGC GENETICS COMMITTEE MEMBERS

Christine Cadrin (Chair) Montreal, Que.
Jo-Ann Johnson Toronto, Ont.
Barbara McGillivray Vancouver, BC
Chris Harman Winnipeg, Man.
R. Douglas Wilson Vancouver, BC
David C. Young Halifax, NS

PRINCIPAL AUTHORS

E.M. Hutton, PhD, Department of Genetics, The Hospital for Sick Children, Toronto, Ont.
B.N. Chodirker, MD, Department of Paediatrics, University of Manitoba
B. McGillivray, MD, Department of Medical Genetics, University of British Columbia
D.R. McLeod, MD, Department of Medical Genetics, Alberta Children's Hospital, Calgary, Alta.
R.G. Wilson, MD, Department of Obstetrics and Gynaecology and Medical Genetics, University of British Columbia
E.J.T. Winsor, PhD, Department of Pathology, The Toronto Hospital, Toronto, Ont.

Prenatal Diagnosis Committee of the Canadian College of Medical Genetics

E.M. Hutton, PhD (Chair) Toronto, Ont.

We thank those who reviewed this document and provided a thoughtful approach to the interaction of health care professionals in the area of prenatal screening diagnosis.

N. Brochu, RN Ottawa, Ont.
L. Cartier, MSc Montreal, Que.
A. Dickes, MSc Vancouver, BC
C. Harman, MD Winnipeg, Man.
A. Lynch, PhD Toronto, Ont.
P. Niday, RN, EdD Ottawa, Ont.
J. L. Reynolds, MD London, Ont.
A. Sprague, RN, Med Ottawa, Ont.
M.-J. Trépanier RN Ottawa, Ont.

and members of the Canadian College of Medical Geneticists who provided careful review and comments. Ms. Sandra Goncalves MSc, (Toronto, Ont.) provided helpful editorial assistance.

Déclaration de principe : le document d'opinions fait état des percées récentes et des progrès cliniques et scientifiques à la date de publication de celle-ci et peut faire l'objet de modifications. Il ne faut pas interpréter l'information qui y figure comme l'imposition d'une procédure ou d'un mode de traitement exclusifs à suivre. Un établissement hospitalier est libre de dixter des modifications à apporter à ces opinions. En l'occurrence, il faut qu'il y ait documentation à l'appui de cet établissement. Aucune partie ne peut être reproduite sans une permission écrite de la SOGC.
ABSTRACT

Objective: to provide health care providers and genetic/ perinatal centres with an effective and ethical approach to prenatal genetic testing. The Royal Commission on New Reproductive Technologies has shown a need for consistent standards of clinical practice for prenatal diagnostic services.

Options: these guidelines apply to the offer of invasive (chorionic villus sampling, amniocentesis) and non-invasive techniques (maternal serum screening, ultrasound/diagnosis and screening).

Outcomes: outcomes include acceptance or refusal of prenatal genetic testing, and continuation or termination of pregnancy.

Evidence: expert opinions were obtained from health care providers and professionals in genetic/perinatal centres.

Values: opinions were provided by an ethicist, family physicians, obstetricians, midwives, medical geneticists, nurses, genetic counsellors, and genetic counsellors.

Benefits or harms, and costs: use of the guidelines will improve service and communication between health care providers and women eligible for testing.

Recommendations: health care providers should know current indications for testing and types of prenatal diagnostic and screening procedures. Prenatal genetic testing, where appropriate, should be offered after non-directive counselling about the advantages and disadvantages and implications of the procedures. The choice to have or not have testing is entirely the woman’s choice. If an abnormality is detected, a woman’s decision about continuation or termination of pregnancy should be fully supported.

Validation: the guidelines were reviewed by all selected representatives and by the Board of the Canadian College of Medical Genetics and the Council of the Society of Obstetricians and Gynaecologists of Canada.

Sponsors: the Canadian College of Medical Genetics, the Canadian Association of Genetic Counsellors, the Canadian Nurses Association, the College of Family Physicians of Canada, and the Society of Obstetricians and Gynaecologists of Canada.

KEY WORDS

Prenatal screening and diagnosis, guidelines, ethics, non-directive counseling, genetic/perinatal.

INTRODUCTION

Prenatal genetic testing has been available in Canada through genetic centres for more than 20 years. In recent years the scope of testing has expanded greatly with the development of non-invasive screening procedures. Many health care providers outside genetic centres now provide basic counselling prior to offering prenatal testing. These Practice Guidelines have been developed for those professionals offering prenatal tests, and for genetic centres providing counselling and specialized prenatal screening and diagnostic testing.

Part I is directed to the health care providers who offer prenatal genetic screening or diagnosis. These professionals are family physicians, obstetricians, nurses, midwives, medical geneticists, genetic counsellors, and any other professionals interacting with pregnant women.

Part II is directed to health care providers involved in the use of prenatal screening and diagnostic procedures in prenatal diagnosis centres.

Part III outlines the responsibilities of those professionals conveying information to patients in whom an abnormality or unusual finding has been detected in the course of prenatal diagnosis.

I. OFFER OF PRENATAL SCREENING AND DIAGNOSIS

DETERMINING THE INDICATIONS FOR PRENATAL SCREENING AND DIAGNOSIS

Health care providers should be aware of the current indications for prenatal diagnosis. The publication “Canadian Guidelines for Prenatal Diagnosis. An Update” published in 1993 is the general guide at present. Guideline revisions and current information regarding prenatal diagnosis may be obtained from local genetic centres. As major changes in policies with respect to the offer of prenatal testing may occur, there should be a link between health care providers and the genetic/perinatal centres so that updated information can be communicated.

To determine whether there is an indication for prenatal testing, an assessment should include the medical history and obstetric history of the woman, as well as a specific inquiry about ethnic background and any family history of possible genetic disorders in the woman or her partner.

Assessment of family history should include enquiry about siblings, parents, parents’ siblings, grandparents, and of other extended family members where appropriate. In some cases, a special genetic assessment is necessary, and the woman should be offered a referral to a genetic centre so that a review of medical records of the
woman and other family members can be undertaken to clarify relevant history.

Genetic screening should be offered to those women/couples with ethnic backgrounds known to be associated with an increased risk of certain single gene disorders, e.g. thalassaemia.

**OFFER OF TESTING**

Women are strongly encouraged to seek referral through their health care providers; however, they can also contact centres directly for genetic counselling and testing services. Regardless of the views of the health care provider towards abortion for fetal abnormalities, testing should be discussed with all women who have appropriate indications.

When the health care provider’s personal beliefs preclude such a discussion, a direct referral should be made to an alternative provider. Failure to discuss all options with women at risk or, alternatively, to refer them further, is unethical.

Women should understand that they are free to decline any aspect of the offer of testing. Although genetic/perinatal centres usually encourage the participation of a male partner in discussions, the woman may choose to exclude her partner from any or all discussions. Each woman should be allowed to decide whether prenatal testing is appropriate for her, based on informed choice.

The Report of the Royal Commission on New Reproductive Technologies states that “informed choice means providing relevant and understandable information about the options and the possible implications of various decisions. It means supporting individual decision-making by helping people identify what is important to them and how various decisions would coincide with their priorities, given their values and circumstances. Informed choice also entails allowing as much time as possible for discussion and reflection.”

The health care provider should be aware of new prenatal screening and diagnostic techniques, so that all appropriate prenatal diagnosis testing options can be discussed. Non-invasive tests include maternal serum marker screening and ultrasound examination for fetal anomalies. Invasive tests include chorionic villus sampling, early or mid-trimester amniocentesis and fetal blood sampling by cordocentesis.

It is important for the health care provider to be aware of local and regional variations in access to non-invasive and invasive prenatal diagnosis techniques. At present, all provinces have the availability of amniocentesis (invasive), while only two provinces (Manitoba and Ontario) have established provincial maternal serum screening programmes (non-invasive). Availability of non-invasive screening (maternal serum screening, ultrasound aneuploidy markers) is increasing but cannot be considered a standard of care in Canada at present.

Counselling of the woman and her partner should be non-directive and comprehensive, in a language they understand. Women considering prenatal testing should be made aware that agreement to terminate a pregnancy if an abnormality is found is not a precondition or requirement prior to counselling or testing.

Counselling should include a discussion of:
- available methods of prenatal diagnosis;
- risks and benefits of various techniques;
- timing of prenatal diagnostic procedures;
- the condition(s) for which testing is to be considered and current management of relevant genetic disorders;
- accuracy of prenatal test results (including the implications of false-positive and false-negative results where appropriate).

In addition, the option of abortion should be discussed and appropriate details given where desired. Counselling may be accompanied by written information from the local centre.

Referral to a genetic/perinatal centre is appropriate when invasive testing is indicated, when there is a significant family history or if the woman and/or partner request additional information. Care should be taken by health care providers to supply complete and accurate information about the patient to genetic centres or laboratories performing genetic testing as patient-specific factors can influence the interpretation of some screening tests, e.g. maternal serum screening risk estimation is affected by gestational age.

**REPORTING GENETIC TEST RESULTS**

(See also Part III Concerning discussion of abnormal results of prenatal genetic screening or diagnosis.)

It is the responsibility of the health care provider who initiates a referral for prenatal screening or diagnosis to ensure that, when test results are received, they are reviewed and reported to the woman as quickly as possible. Appropriate follow-up tests and counselling should be arranged through the local screening programme or genetic/perinatal centre. Referral to an outreach programme is often available for those families who do not have easy access to a centre.

All patient information should be treated with confidentiality. Safeguards should be in place to protect records from unauthorized access. Results from any genetic tests should be reported directly to that individual only, or their physician, unless prior arrangements have been made to report a result to the partner or other family member. The wishes of the woman concerning
the communication of prenatal genetic test results on the fetus must be respected. Her permission must be obtained before allowing access to her test results.

11. Prenatal Genetic Testing

Criteria for Testing

The Prenatal Diagnosis Centre should have a well-defined policy for offering invasive testing. The Canadian Guidelines for Prenatal Diagnosis provide basic criteria for determining whether a woman has sufficient risk to justify an invasive procedure. If a centre cannot accommodate an individual woman's request for prenatal diagnosis that falls within the Canadian Guidelines, the centre has a responsibility to refer the patient to another genetic centre that can perform the indicated procedure(s).

Centres may differ in their ability to provide testing for women with indications not listed in the Guidelines as availability of skilled operators and laboratory resources may vary. A policy to handle such referrals in a fair and consistent manner should be developed.

Prenatal Diagnosis Counselling

General information regarding genetic counselling and the offer of prenatal testing is reviewed in Part I. The genetic centre should indicate which tests are routinely offered. Women offered specialized testing should be given more extensive counselling, indicating how their situation may differ from the general application of prenatal diagnostic procedures. The policy of the genetic centre regarding the availability of pregnancy termination procedures should be clearly communicated. The referring health care provider should be advised about the nature of proposed tests and the expected timing of procedures.

Appropriate written information and other educational aids, including videos, can be made available for couples attending genetic clinics. Information brochures should be reviewed by the centre on a regular basis and updated where necessary. Written information can be made available to health care providers outside the health care centres who wish to counsel women/couples themselves.

If testing is offered to a woman before adequate research has been conducted to make certain that reasonable accuracy and/or safety can be achieved, great care must be taken to ensure that she understands the uncertainty and the experimental nature of the proposed investigations. A detailed explanation outlining the research nature of the procedure should be given.

New testing procedures should be reviewed by the local hospital practice ethics committee or other relevant bodies, e.g. provincial genetics advisory committee. Where possible, introduction of new procedures should be consistent with any provincial or national guidelines. In the absence of such guidelines, these procedures may be offered in specific situations only after detailed discussion with the woman about the possible advantages and disadvantages of using a testing procedure that lacks sufficient evaluation.

Prior to invasive testing, the woman and her partner should have sufficient time to consider the advantages and disadvantages of alternate screening or diagnostic procedures. Where practical, counselling appointments should be scheduled at least one day prior to invasive procedures so that couples have time to think about their options and not feel pressured into having genetic testing. Telephone discussions are an important adjunct, especially for those women who are geographically distant from the centre.

Testing and Reporting of Results

The person responsible for processing and reporting the results of a particular sample must be clearly defined. The referring health care provider should easily be able to contact the appropriate member of the prenatal diagnosis team to track the progress of investigations. The woman and her health care provider should be notified when a diagnosis is delayed beyond a reasonable pre-agreed time interval. The testing centre also has a responsibility to review the progress of the test (e.g. culture) to ensure prompt notification of problems and to allow repeat testing, alternative procedures and counselling.

The prenatal diagnosis centre should assess the reliability of referral laboratories if samples are being sent outside the centre. Laboratory specialists associated with the genetic programme should provide general direction about the referral process and reporting of the prenatal diagnosis result, whether the testing is performed locally or sent to other centres. Laboratory directors should develop guidelines about information to be included in a report. Laboratories should participate in proficiency testing programmes as a mechanism for maintaining standards.

After a verbal report has been given, a written statement of results of the prenatal screening and diagnosis should be received by the referring health care provider as soon as possible (usually within one week) after the completion of all testing.
III. ABNORMAL RESULTS OF PRENATAL DIAGNOSIS (CYTOGENETIC, DNA, BIOCHEMICAL, FETAL STRUCTURAL ABNORMALITIES)

Discussion of Abnormal Results

When a fetal abnormality is diagnosed as part of prenatal diagnosis for a specific indication, the woman will have undergone a consent process prior to the procedure and should have been counselled about possible outcomes. However, if fetal abnormalities are detected during an ultrasound assessment when there are no special risk factors, the woman may be unprepared. A specific protocol should be developed to offer support and guidance for the woman and her partner in this situation.

Both the laboratory and genetic counselling services should develop clear procedures for reporting abnormal findings. The results should be communicated as swiftly as possible by telephone, with rapid follow-up of written results by mail, fax or appropriate delivery system. If the results are received by the genetics service, they should first be reported to the referring health care provider. At this time, a decision can be made as to who will contact the patient, explain the abnormality and assume responsibility for any further testing and follow-up. If the results are not conveyed directly to a genetic service, the managing health care provider should arrange for genetic counselling, especially if a result is complicated or unclear.

The woman should be contacted promptly, and the diagnosis discussed in person where possible. Women should be encouraged to involve their partners in all discussions. The information should include an explanation of the results in lay terms, current information regarding the natural history of the condition, a description of other investigations which might provide further information and options for the pregnancy. Pregnancy options should be presented in an unbiased fashion, and may include continuation or termination of the pregnancy, with or without further investigations or treatments. Information should be available about carrying the fetus to term, care of the child and community support systems, and also opportunities should be provided for discussion with families who carried to term or are raising an affected child. For the couple not wishing to continue the pregnancy, information should be given by the appropriate specialists about the types and availability of termination procedures.

Consideration should be given to the involvement of other diagnostic or support services to provide more information to the couple. These resources could include speciality clinics, subspecialists and support services in the hospital or the community. In some difficult cases, a team of professionals from several different areas of expertise may be necessary to help the woman or couple to plan management of the pregnancy.

Support of the Woman’s Decision

If an abnormal pregnancy is to be continued, all appropriate information, results and recommendations should be available to the managing health care professional as soon as possible. In some cases, further studies at the time of birth may be necessary to clarify a diagnosis and determine appropriate ongoing care of the baby.

If the woman’s decision is to terminate the pregnancy or if there is intra-uterine death, discussion with the parents should include methods to preserve memories of the baby (where appropriate), burial or cremation arrangements, as well as discussion of the need for complete embryopathology studies. The termination procedure should be made available without undue delay. If parental consent is given, the genetic service has a responsibility to ensure adequate information about the fetal anomalies so that accurate recurrence risk counselling can be provided for the family.

If the prenatal centre provides genetic screening and diagnosis but not termination of pregnancy, when an abnormality is detected, there is an obligation to confer with the referring health care provider about alternative facilities.

Follow-up

As part of a quality assurance programme, the laboratory providing the testing should try to confirm any abnormal prenatal diagnosis results. The extent of confirmatory studies may vary depending on the type of abnormality detected.

The health care provider who refers a woman for termination of pregnancy when a fetal disorder is detected should be sure that arrangements are made for the support to the woman and her family before, during and after the termination procedure if it is not available through a prenatal diagnosis programme. Referral to self-help groups or associations may be appropriate.

Fetal pathology results and recurrence risks should be formally addressed, with a copy of the results and a summary of the discussion sent to the referring health care professional.

Conclusion

Prenatal screening and diagnosis is a constantly changing field with increasing emphasis on non-invasive screening methods, carrier detection and risk assessment, as well as invasive techniques performed in early pregnancy.
There should also be ongoing public discussion. A close communication between the genetic centres and the referring health care providers is important, to ensure that women or couples receive accurate and unbiased information about the range of testing that is currently available.

J SOC OBSTET GYNAECOL CAN 1998;20(9):865-70

REFERENCES

Reprints and copies can be ordered by contacting the Information and Documentation Department, Ribosome Communications Inc.
50 St. Clair Avenue East, 4th Floor
Toronto, Ontario M4T 1M9
Tel: (416) 925-1543; Fax: (416) 925-7682
E-mail: jsogc@ribosome.com