Screening for fetal anomalies: old habits, new challenges

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Editorial

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Résumé

LE RÉSULTAT D'UN NOUVEAU PROGRAMME DE SANTÉ PUBLIQUE est fonction dans une grande mesure des connaissances et des attitudes des praticiens qui les offrent. Dans le présent numéro (page 775), le Dr June C. Carroll et ses collaborateurs présentent un rapport sur les pratiques, les connaissances et les opinions des médecins et des sages-femmes de l'Ontario en ce qui a trait à l'examen du sang maternel pour le dépistage du syndrome de Down, de défauts du tube médullaire et de la trisomie 18. Ils démontrent que l'anxiété de la mère, des taux élevés de résultats faussement positifs et l'éthique des tests prénataux préoccupent toujours les praticiens. L'auteur de cet éditorial discute de ces préoccupations en regard des pratiques antérieures de dépistage et soutient que l'éducation améliorée des fournisseurs de soins de santé et des patients, et l'expérience, sont les seuls moyens de surmonter les difficultés posées par le dépistage prénatal.

In this issue (page 775) Dr. June C. Carroll and associates report on the practices, knowledge and opinions of Ontario family physicians, obstetricians and midwives with respect to the recently introduced maternal serum screening (MSS) program for Down syndrome, neural tube defects and trisomy 18. They reveal that practitioners continue to be concerned about maternal anxiety, high rates of false-positive results and the ethics of prenatal screening. They also document a significant level of misunderstanding about MSS.

Prenatal screening for genetic disorders is not new, although many practitioners will not think of some of the strategies they use as screening. Asking a pregnant woman about her age or her family history of neural tube defects, for example, are forms of screening for fetal anomalies. The inadequacy of such strategies is clear. Given that 90%–95% of infants with neural tube defects are born to families *not* known to be at risk, the sensitivity of a positive family history as a screening tool is less than 10%, while a maternal age of 35 years or more would identify only 20%–25% of women carrying a fetus with Down syndrome or trisomy 18. Similarly, with respect to positive predictive value, less than 5% of pregnant women with a positive family history of neural tube defects will have an affected child, while a much smaller proportion of older pregnant women will have an infant with Down syndrome or trisomy 18. Nevertheless, current practice guidelines advise that diagnostic testing should be offered to all women identified as "at risk" by these criteria.

Many practitioners surveyed by Carroll and associates did not respond to survey questions testing their knowledge about the proportion of women who initially receive positive test results or about false-positive rates. Of those who did answer, many underestimated the false-positive rates. There seems little doubt that failure to appreciate the high probability of having an unaffected child despite a positive screening result could lead to significant anxiety on the part of both patient and physician.

Before diagnostic testing became available, older women and those with a family history of genetic disorders or birth defects were concerned about their probability of having an affected child. Amniocentesis provided a way to address this anxiety but also brought the additional anguish of the risk it poses to the fetus and the possibility of facing a decision about terminating a pregnancy. Despite these concerns, amniocentesis is now generally perceived as routine for older women. By contrast, women referred for amniocentesis after MSS may feel singled out.[1]

What can be done about the anxiety provoked by MSS? For the program as a whole, time will help. In the meantime, the concerns of the individual patient are real and immediate and must be dealt with. Counselling women who receive a positive result is a key component of any successful screening program.[1–3] Counselling women *before* testing is also crucial. Pregnant women surveyed in the UK were well informed about the logistic aspects of MSS for Down syndrome but not about the implications of positive and negative results.[4]

A relatively high proportion of the physicians surveyed by Carroll and associates found that counselling patients about MSS was time consuming or complex. Interestingly, a much smaller proportion of midwives found this aspect of screening troublesome. In a similar study conducted in Manitoba, physicians expressed concern about maternal anxiety and high false-positive rates but seldom raised the issue of counselling, perhaps because the program had been in existence for more than 7 years.[5]

Given the complexities of MSS and the effort required to make it "work," it might be argued that it makes poor use of scarce resources. The midwives in Carroll and associates' study were those who commented most favourably on MSS as an alternative to invasive testing in older women; most genetic counsellors and medical geneticists would echo that sentiment. Despite the routineness of an amniocentesis referral for older women, it is a procedure that many women contemplate with trepidation. To them, more precise information on their *own* risk — rather than the statistical risk assigned to all women their age — can help them to decide about testing. At the same time, many younger women who would not have thought their fetuses to be at risk would consider amniocentesis if MSS indicated a substantial increase in that risk. Women offered amniocentesis on the basis of the age cutoff of 35 years have only half the chance of having an existing fetal chromosomal anomaly detected by such testing than women selected on the basis of MSS results. Targeting amniocentesis toward those at greatest risk regardless of age makes better use of our diminishing health care dollars and, more important, reduces the number of fetuses lost as a result of the procedure itself.

Some of the obstetricians and midwives and over a quarter of the family physicians surveyed recommended that MSS screening be offered only to women at high risk. However, as Carroll and associates point out, it is inappropriate to recommend MSS for women who really should be referred for genetic counselling. It is the brave practitioner who would recommend MSS alone for patients with family histories of neural tube defects or Down syndrome without first being assured that the neural tube defect was isolated and not part of a genetic disorder such as Meckel syndrome, or that the uncle with Down syndrome had trisomy 21 and not a familial translocation. Both risks and appropriate investigations would differ significantly in such cases.

Should MSS become routine for all pregnant women? The answer is clearly no: the decision to be screened should never be considered routine. But what are the factors that influence that decision? Carroll and associates show that views on abortion colour feelings about MSS; 47% of the physicians surveyed who would *not* refer a woman requesting an abortion after an anomaly was confirmed wanted the program scrapped, whereas only 18% of those who *would* provide such a referral thought the program should be dropped. These findings are of concern; it should be for each woman to decide whether or not to be tested. Nevertheless, it would be too easy to dismiss the concerns of health care providers about MSS as echoing the anti-abortion sentiments of a few. Various problems with the implementation of MSS programs have been articulated by many practitioners and deserve to be addressed.

Screening for genetic disorders is here to stay. The pros and cons of various forms of genetic screening and testing will be debated as tests move out of the laboratory and into clinical practice. Each type of screening will necessitate public and professional education. More research is needed to determine the best ways of providing information to front-line health care workers and the patients they care for. We also need a better understanding of the patient's perspective. Despite the problems of MSS, most women feel that it should be available to them and would choose to be tested.[6–9] Our challenge now is to better serve those who make that decision.

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