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Current Practice and Attitudes of Australian Obstetricians Toward Population-Based Carrier Screening for Inherited Conditions

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An anonymous survey of Australian Fellows of the Royal Australian and New Zealand College of Obstetricians and Gynaecologists was conducted with the aim of understanding current practice and attitudes toward population-based carrier screening for inherited conditions in the setting of routine pregnancy care. Of 1,121 Fellows invited to complete the online questionnaire by e-mail, 237 (21%) responded, and of these 156 were practicing obstetricians and completed the whole survey. Of the respondents, 83% expressed support for population-based carrier screening for at least some conditions, with 97% supporting carrier screening for β-thalassaemia, and 83% supporting carrier screening for cystic fibrosis (CF). A small proportion of obstetricians reported offering carrier screening as part of routine pregnancy care (20% for β-thalassaemia, 8% for CF, 5% for fragile X syndrome, and 2% for spinal muscular atrophy). The main practical barriers identified for screening were cost, time constraints, and availability of supporting services. Addressing these issues is crucial for the successful implementation of population-based carrier screening programs in Australia and internationally.

■ **Keywords:** carrier screening, cystic fibrosis, thalassaemia, screening programs

The purpose of population-based carrier screening is to identify asymptomatic carriers of autosomal and X-linked recessive conditions and give prospective parents reproductive options to prevent the birth of an affected child. Screening programs began formally in the 1970s with screening for Tay–Sachs disease carrier status in the Ashkenazi Jewish community (Kaback, 1997). Subsequent programs have targeted cystic fibrosis (CF) in the United States and parts of Italy (Castellani et al., 2009; Hale et al., 2008), thalassaemia in Mediterranean at-risk populations (Cao et al., 1984, 1997; Modell & Mouzouras, 1982), fragile X syndrome in Israel (Berkenstadt et al., 2007), and most recently, spinal muscular atrophy (SMA) in the United States and Taiwan (Su et al., 2011; Sugarman et al., 2012). Several of these programs have reported reductions in the incidence of affected infants born with the conditions tested. Developments in genetic technology mean that it is now possible to simultaneously screen for an individual's carrier status for hundreds of inherited conditions using a single sample (Levenson, 2010), and such panel-based testing is likely to replace testing for individual conditions in the future.

In Australia, healthcare is available through the government-funded public health system, as well as through a user-pays private health system. The availability of carrier screening varies for different conditions and in different settings. Screening for β-thalassaemia carrier status is publically funded and generally triggered by abnormal results on full blood examination (FBE), which is performed as part of routine pregnancy care (Cousens

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et al., 2010). There are several well-established, non-government funded, community-based programs offering carrier screening to people of Ashkenazi (Eastern European) Jewish ancestry for conditions such as Tay–Sachs disease (Ioannou et al., 2010). A fee-for-service CF population carrier screening program has been in existence in the state of Victoria, Australia since 2006 (Massie et al., 2009). In its first three years of operation, the program screened 3,200 individuals, detecting 106 carriers, and 9 carrier couples. All the couples identified through the program altered their reproductive decisions, to avoid having a child with CF. Screening for other relatively common genetic conditions, such as SMA and fragile X syndrome carrier status, are less frequently offered in Australia (Metcalfe et al., 2008).

The uptake of carrier screening is generally higher when offered in pregnancy than when offered to the non-pregnant population (Harris et al., 1996; Mennie et al., 1992; Wald et al., 1993). The reasons may be that the first contact with a health professional does not occur until the woman is pregnant or that screening does not become a priority until this time. In addition, couples may not believe they need to consider screening until pregnant (Delatycki, 2008). There is little known about the attitudes of Australian obstetricians toward carrier screening for inherited conditions. Knowledge in relation to these attitudes is of great importance as the frequency with which obstetricians and other pregnancy healthcare providers offer tests to patients is a major determinant of the success of population-based carrier screening programs.

The aim of this study was to gather information about the current practice and attitudes of Australian obstetricians toward carrier screening for genetic conditions as part of routine pregnancy care.

Methods

Participants

Australian Fellows of the Royal Australian and New Zealand College of Obstetricians and Gynaecologists (RANZCOG) were invited by e-mail to complete an anonymous online survey between January 27 and March 3, 2011. Only Fellows actively practicing obstetrics were requested to complete the whole survey. A reminder was sent three weeks after the initial invitation to participate, inviting those who had not completed the survey to do so.

Measurement Tool

The survey was developed by a panel of clinicians and researchers with expertise in carrier screening programs and was informed by previous international studies (Morgan et al., 2004, 2005; Wilkins-Haug et al., 1999). The survey was reviewed by the RANZCOG Continuing Professional Development committee, and the content was modified in response to their feedback. The survey was divided into four parts: demographic information, current practice and attitudes toward screening for β-thalassaemia, current practice and attitudes toward screening for CF, and attitudes toward population-based genetic carrier screening in general. Respondents were asked to rate certain aspects of β-thalassaemia and CF screening tests on a 5-point scale, where 1 was *very poor*, 2 *poor*, 3 *satisfactory*, 4 *good*, and 5 *excellent*. They were also asked to rate their level of concern regarding general aspects of population-based screening for genetic conditions using a 4-point scale, where 1 was *no concern*, 2 *minor concern*, 3 *moderate concern*, and 4 *major concern*. Respondents were provided with two, free text boxes at the end of the survey and asked for any additional comments about specific barriers to screening or general comments. Responses to the questionnaire were anonymous.

Data Analysis

LimeSurvey software was used to generate the electronic version of the survey, and to store and analyze the responses. Using content analysis, open-ended responses were categorized independently by Zornitza Stark, Belinda McClaren, and Sylvia Metcalfe based on similarity and differences. Numbers of responses in categories are reported.

Ethics Committee Approval

The study was approved by the Royal Children's Hospital, Victoria, Australia Human Research Ethics Committee (HREC 30068).

Results

Demographic Details

A total of 1,206 e-mails were sent to practicing Australian Fellows of the RANZCOG who had supplied the College with an e-mail address. Eighty-five e-mails were returned as undeliverable, leaving 1,121 potential respondents. From those, 237 responses were received (response rate minimum of 21.1%) with representative proportion of responses received from each state/territory; 55 respondents identified themselves as not practicing obstetrics, and 26 did not complete the survey sufficiently for their responses to be included in the analysis. One hundred and fifty-six eligible Fellows completed the full survey. Demographic information and type of practice of the respondents is shown in Table 1.

Current Practice and Attitudes Toward Carrier Testing for β-Thalassaemia and Cystic Fibrosis

One hundred and fifty-two obstetricians (97%) supported carrier testing for β-thalassaemia in pregnancy, and 130 (83%) supported carrier screening for CF. Self-reported current practice patterns with respect to these two conditions are summarized in Table 2. The opinion of obstetricians regarding certain aspects of β-thalassaemia and CF carrier screening are presented in Table 3.

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TABLE 1

Demographic Information and Type of Practice of the 156 Survey Participants

Respondent characteristics	*N* (%)
Gender	
Male	82 (53%)
Female	74 (47%)
State/territory of main practice	
Australian Capital Territory	3 (2%)
New South Wales	39 (25%)
Northern Territory	2 (1%)
Queensland	37 (24%)
South Australia	17 (11%)
Tasmania	7 (5%)
Victoria	41 (26%)
Western Australia	10 (6%)
Years of obstetric experience	
<5	1 (0.6%)
6–10	26 (16.7%)
11–15	36 (23.0%)
>16	93 (59.7%)
Size of obstetric practice (deliveries/year)	
1–20	18 (11%)
20–100	32 (21%)
100–200	38 (25%)
>200	68 (43%)
Location of practice	
Metropolitan	115 (74%)
Rural/regional	41 (26%)
Type of practice	
Mostly private	80 (51%)
Mostly public, tertiary center	40 (26%)
Mostly public, other	36 (23%)
University appointment	
Yes	60 (39%)
No	96 (61%)

TABLE 2

Self-Reported Current Practice Regarding Offering Carrier Screening for β-Thalassaemia and CF in Routine Pregnancy Care (Total Number of Respondents: 156)

Current practice pattern	β-thalassaemia *N* (%)	Cystic fibrosis *N* (%)
Offer screening to all patients	32 (20%)	12* (8%)
Offer screening to some patients	113 (72%)	128 (82%)
Personal or family history	109 (70%)	123 (79%)
Higher risk ethnic group	85 (55%)	28 (18%)
Patient request	75 (48%)	88 (56%)
Private patients	0	9 (6%)
Screening not offered to any patients	11 (7%)	16 (10%)

Note: *All 12 practiced in states where there are established fee-for-service CF carrier testing programs (Victoria and New South Wales).

Current Practice and Attitudes Toward Carrier Testing for Other Inherited Conditions

One hundred and thirty obstetricians (83%) supported population-based carrier screening for at least some inherited conditions. However, only 9 (6%) felt this should take place during pregnancy, with 90 respondents (58%) stating it should ideally take place in adulthood before pregnancy, 33 (21%) at birth and 24 (15%) in high school. A very low number of obstetricians reported routinely offering carrier tests for any other conditions: 7 (5%) for Tay–Sachs disease, 8 (5%) for fragile X syndrome, and 3 (2%) for SMA.

TABLE 3

Respondents' Mean Rating on Scale of 1–5 of Practical Aspects of β-thalassaemia and Cystic Fibrosis Carrier Screening

	β-thalassaemia	Cystic fibrosis
Ease of access to test	4.0	3.7
Cost of test	3.4	2.9
Sensitivity and specificity of the test	3.7	3.6
Availability of laboratory and counseling support to help with the interpretation and follow-up of abnormal results	3.6	3.6
Availability of educational materials to help counseling patients	2.8	3.3
Community awareness of condition	2.3	2.5

The participants were asked to rate their level of concern regarding various aspects of population-based carrier screening and their responses are presented in Figure 1. Ninety-three (60%) of survey participants stated they would like more training in this area.

Additional Comments

Forty-six participants provided further comments in the open-ended questions. Five of these commented on survey design only and these are not reported. The responses of the remaining 41 participants were categorized based on similarity of content. Some participants' responses covered more than one topic and their comments were coded into more than one category. Forty-nine comments addressed topics raised in Tables 2 and 3 and Figure 1. There were eight new topics raised in 25 comments that were not already covered in the survey. These were: concern about equity of access and distributive justice, from the perspective of reaching disadvantaged or multicultural populations, or limiting testing to high genetic risk populations, or targeting populations such as preconception/pregnant couples ($n = 14$), potential for causing harm through creating a perception of eugenics in society ($n = 1$), potential for stigmatization ($n = 1$), or raising questions regarding paternity ($n = 1$), impacting on life insurance ($n = 2$), the lack of evidence of cost-benefit ($n = 3$), and the need for screening to be policy driven ($n = 1$). Two respondents commented on their personal view that screening has eugenic undertones.

Discussion

This is the first study to examine current practice and attitudes of Australian obstetricians toward population-based carrier screening for genetic conditions in routine pregnancy care. The majority of obstetricians expressed support for population-based carrier screening for at least some conditions, with 97% supporting carrier screening for β-thalassaemia, and 83% supporting carrier screening for CF.

The largely positive attitudes toward universal carrier screening among Australian obstetricians are not translated into practice, with only 20% reporting they routinely offer

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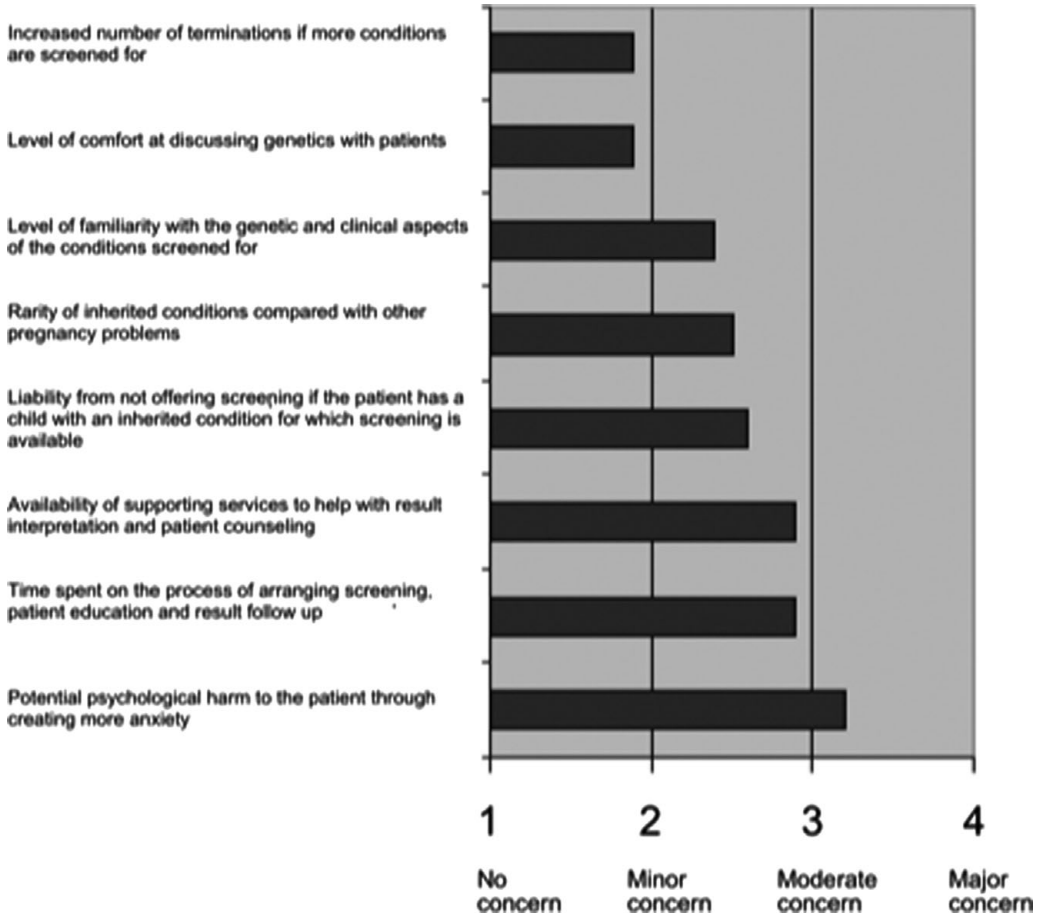

FIGURE 1

Responses to survey questions asking participants to rate their level of concern regarding general aspects of population-based carrier screening for genetic conditions on a scale of 1–4.

β-thalassaemia screening to all their patients, 8% offering carrier screening for CF, 5% for fragile X syndrome, and 2% for SMA. This contrasts with self-reported practice among obstetricians in the United States, where a similar survey found that 65.8% of respondents offered CF carrier screening to all prenatal patients (Morgan et al., 2004). The low number of Australian obstetricians reporting offering β-thalassaemia screening to all patients, when in practice the majority of pregnant women have a FBE performed, most likely reflects the indirect nature of β-thalassaemia screening, with FBE testing not being perceived by obstetricians as a screening test for β-thalassaemia. This finding is consistent with patients found to be carriers for β-thalassaemia typically reporting that they were unaware that screening had taken place (Locock & Kai, 2008).

Most Australian obstetricians report offering carrier testing in specific circumstances, most commonly in the presence of a personal or family history of a genetic condition. However, the majority of babies with CF are born to families with no family history of CF (McClaren et al., 2011), and even when a family history of CF is known, only a small proportion of relatives undertake carrier testing (McClaren et al., 2010). This is not unique to CF, but applies to all recessively inherited conditions. Therefore, the family history-based approach (so-called 'cascade testing') is likely to identify only a small proportion of couples who are at risk of having a child affected by an autosomal recessive condition.

In 2001, the American College of Medical Genetics (ACMG) and the American College of Obstetricians and Gynecologists (ACOG) issued joint guidelines recommending healthcare providers to offer CF carrier screening to all couples planning a pregnancy or seeking prenatal testing (ACOG/ACMG, 2001), and a similar position statement has been issued by the Human Genetics Society of Australasia (HGSA, 2009). The RANZCOG specifically recommends β-thalassaemia screening in routine pregnancy care (RANZCOG, 2009), and with regard to other conditions suggests that 'counselling should address availability of carrier status screening for genetic conditions of perceived high prevalence or consequence' (RANZCOG, 2010).

A number of practical issues were raised by the obstetricians in this survey as barriers to offering universal carrier

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screening. Chief among these was financial cost, both to individual patients and to the health system as a whole. As a guide, one Australian laboratory charges A$220 for CF carrier screening, A$250 for fragile X screening, and A$350 for SMA screening. With the exception of the FBE and hemoglobin electrophoresis that can diagnose carrier status for β-thalassaemia, the cost of population-based genetic carrier screening tests is not currently covered by government funding or private health insurance. By contrast, several health insurance providers in the United States cover the cost of such testing. It should be noted that some of the cost of the first trimester trisomy 21 screen is similarly not covered by government funding or private health insurance in Australia. This is commonly offered as part of routine pregnancy care, and the majority of pregnant women choose to pay between A$200–300 to include maternal serum screening and a nuchal translucency measurement by ultrasound. Carrier testing has the advantage that it only needs to occur once in each individual's lifetime rather than in each pregnancy, provided partners remain unchanged. Nevertheless, cost is an important barrier to universal carrier screening. The current arrangement in Australia creates inequity in healthcare, with only those that can afford it being in a position to take up carrier screening.

Patient education is an integral part of informed consent. Time constraints, language and cultural barriers, uncertainty in interpreting results, and lack of supporting services were all identified as important barriers to offering screening. With the number of available screening tests set to increase, it may be that detailed counseling will need to be reserved for those couples found to be at increased risk of specific conditions. Of note, the majority of obstetricians offering CF carrier screening routinely to all patients practiced in the states where there are existing fee-for-service carrier screening programs. Having a dedicated program facilitates screening through the provision of practitioner education, a clear pathway for testing, and support with interpretation and follow-up of results.

Creating psychological harm was the most significant concern that Australian obstetricians had with regards to offering population-based carrier screening programs. In addition, some survey participants commented that those found to be carriers may be stigmatized and subject to insurance restrictions. Carriers for recessive genetic conditions are generally asymptomatic, and each person is estimated to be a carrier for several recessive conditions. The evaluation of existing carrier screening programs has shown that carriers are often initially anxious about their positive test results (Ioannou et al., 2010; Scriver et al., 1984). However, this anxiety subsides, and the long-term follow-up of individuals who have taken part in carrier screening programs has shown that the majority have enduring positive feelings about the experience of being screened (Locock & Kai, 2008; Zeesman et al., 1984). Although commonly cited in professional circles and in the mass media, the concern regarding life insurance implications for those found to be carriers is unfounded (Delatycki et al., 2002).

Australian obstetricians expressed only moderate levels of concern regarding liability arising from not offering carrier screening for genetic conditions in pregnancy. This contrasts with studies of American obstetricians, who cited liability from not offering screening as their most significant concern (Morgan et al., 2004) and there are reports of 'wrongful birth' legal action being taken in the United States over failure to provide CF carrier screening (Hausen, 2012). We are not aware of successful legal action being taken for failure to offer such screening in Australia, but if this were to occur, it is likely that the level of concern would increase considerably.

One of the most notable findings of this survey was that only 6% of surveyed obstetricians felt that pregnancy is the ideal time to offer carrier screening, with most favoring preconceptual screening in adulthood. The ethical considerations in choosing a model for universal carrier screening have recently been reviewed, with CF as an example (Modra et al., 2010). It has been argued that preconceptual carrier screening done outside of the medical context (e.g., in schools or workplaces) is ethically superior as it promotes greater autonomy and maximizes the number of reproductive options open to people identified to be carriers. This model works well for conditions that are limited to certain ethic groups, with attendant high degree of community support and education (e.g., Tay–Sachs disease screening programs in Ashkenazi Jews). However, whether it can be translated to the wider community remains to be seen, and in the absence of such programs, offering carrier testing in pregnancy remains important.

This electronic survey elicited responses from only 21% of those successfully e-mailed. It is likely that the distribution list included many Fellows who do not practice obstetrics, and therefore the true response rate of practicing obstetricians is considerably higher. Nevertheless, a relatively low response rate may be indicative of this issue not being perceived as relevant by Australian obstetricians, which in itself would constitute a major barrier to the development of screening programs. Fertility specialists comprise another group of RANZCOG Fellows who are well placed to perform preconceptional carrier screening and ascertaining their views, as well as the views of general practitioners who deliver a substantial part of pregnancy care, will be equally important.

The field of carrier screening for genetic conditions evokes unique ethical, legal, psychosocial, and privacy concerns. Advances in genetic technology mean that the ability to simultaneously screen for an individual's carrier status for hundreds of inherited conditions using a single sample is already a reality (Levenson, 2010) and the cost of such screening will continue to decline. We have identified specific practical barriers and ethical concerns among Australian obstetricians regarding the implementation of

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population-based carrier screening programs. Addressing some of these concerns may increase support for screening and the findings of this survey have important implications for the future planning of screening programs and genetic services in Australia and internationally.

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References

American College of Obstetricians and Gynecologists (ACOG) and the American College of Medical Genetics (ACMG). (2001). *Preconception and prenatal carrier screening for cystic fibrosis*. Washington, DC: Author.

Berkenstadt, M., Ries-Levavi, L., Cuckle, H., Peleg, L., & Barkai, G. (2007). Preconceptional and prenatal screening for fragile X syndrome: Experience with 40,000 tests. [Evaluation studies]. *Prenatal Diagnosis*, *27*, 991–994.

Cao, A., Pintus, L., Lecca, U., Olla, G., Cossu, P., Rosatelli, C., & Galanello, R. (1984). Control of homozygous beta-thalassemia by carrier screening and antenatal diagnosis in Sardinians. *Clinical Genetics*, *26*, 12–22.

Cao, A., Saba, L., Galanello, R., & Rosatelli, M. C. (1997). Molecular diagnosis and carrier screening for beta thalassemia. *Journal of the American Medical Association*, *278*, 1273–1277.

Castellani, C., Picci, L., Tamanini, A., Girardi, P., Rizzotti, P., & Assael, B. M. (2009). Association between carrier screening and incidence of cystic fibrosis. *JAMA*, *302*, 2573–2579.

Cousens, N. E., Gaff, C. L., Metcalfe, S. A., & Delatycki, M. B. (2010). Carrier screening for beta-thalassaemia: A review of international practice. *European Journal of Human Genetics*, *18*, 1077–1083.

Delatycki, M. B. (2008). Population screening for reproductive risk for single gene disorders in Australia: Now and the future. *Twin Research and Human Genetics*, *11*, 422–430.

Delatycki, M., Allen, K., & Williamson, R. (2002). Insurance agreement to facilitate genetic testing. *Lancet*, *359*, 1433.

Hale, J. E., Parad, R. B., & Comeau, A. M. (2008). Newborn screening showing decreasing incidence of cystic fibrosis. *New England Journal of Medicine*, *358*, 973–974.

Harris, H., Scotcher, D., Hartley, N., Wallace, A., Craufurd, D., & Harris, R. (1996). Pilot study of the acceptability of cystic fibrosis carrier testing during routine antenatal consultations in general practice. *British Journal of General Practice*, *46*, 225–227.

Hausen, J. (2012, February 26). Gardiner couple sues medical providers in wrongful birth case. *Bozeman Daily Chronicle*.

Human Genetics Society of Australasia (HGSA). (2009). *Cystic fibrosis population screening position paper*. Retrieved from http://www.hgsa.com.au/images/UserFiles/Attachments/CysticFibrosisPositionPaper.pdf

Ioannou, L., Massie, J., Lewis, S., Petrou, V., Gason, A., Metcalfe, S., . . . Delatycki, M. B. (2010). Evaluation of a multi-disease carrier screening programme in Ashkenazi Jewish high schools. *Clinical Genetics*, *78*, 21–31.

Kaback, M. M., Nathan, T., & Greenwald, S. (1997). Tay-Sachs disease: Heterozygote screening and prenatal diagnosis. US experience and world perspective. In M. M. Kaback (Ed.), *Tay-Sachs disease, screening and prevention* (pp. 13–36). New York: Alan R Liss.

Levenson, D. (2010). New test could make carrier screening more accessible. *American Journal of Medical Genetics Part A*, *152A*(4), vii–viii.

Locock, L., & Kai, J. (2008). Parents' experiences of universal screening for haemoglobin disorders: Implications for practice in a new genetics era. *British Journal of General Practice*, *58*, 161–168.

Massie, J., Petrou, V., Forbes, R., Curnow, L., Ioannou, L., Dusart, D., . . .Delatycki, M. (2009). Population-based carrier screening for cystic fibrosis in Victoria: The first three years experience. *Australian and New Zealand Journal of Obstetrics and Gynaecology*, *49*, 484–489.

McClaren, B. J., Metcalfe, S. A., Aitken, M., Massie, R. J., Ukoumunne, O. C., & Amor, D. J. (2010). Uptake of carrier testing in families after cystic fibrosis diagnosis through newborn screening. *European Journal of Human Genetics*, *18*, 1084–1089.

McClaren, B. J., Metcalfe, S. A., Amor, D. J., Aitken, M., & Massie, J. (2011). A case for cystic fibrosis carrier testing in the general population. *Medical Journal of Australia*, *194*, 208–209.

Mennie, M. E., Gilfillan, A., Compton, M., Curtis, L., Liston, W. A., Pullen, I., . . . Brock, D. J. (1992). Prenatal screening for cystic fibrosis. *Lancet*, *340*, 214–216.

Metcalfe, S., Jacques, A., Archibald, A., Burgess, T., Collins, V., Henry, A., . . . Cohen, J. (2008). A model for offering carrier screening for fragile X syndrome to nonpregnant women: results from a pilot study. *Genetics in Medicine*, *10*, 525–535.

Modell, B., & Mouzouras, M. (1982). Social consequences of introducing antenatal diagnosis for thalassemia. *Birth Defects Original Article Series*, *18*, 285–291.

Modra, L. J., Massie, R. J., & Delatycki, M. B. (2010). Ethical considerations in choosing a model for population-based cystic fibrosis carrier screening. *Medical Journal of Australia*, *193*, 157–160.

Morgan, M. A., Driscoll, D. A., Mennuti, M. T., & Schulkin, J. (2004). Practice patterns of obstetrician-gynecologists regarding preconception and prenatal screening for cystic fibrosis. *Genetics in Medicine*, *6*, 450–455.

Morgan, M. A., Driscoll, D. A., Zinberg, S., Schulkin, J., & Mennuti, M. T. (2005). Impact of self-reported familiarity with guidelines for cystic fibrosis carrier screening. *Obstetrics & Gynecology*, *105*, 1355–1361.

RANZCOG. (2009). *College statement: Pre-pregnancy counselling and routine antenatal assessment in the absence of*

pregnancy complications. Retrieved from http://www.ranzcog.edu.au/publications/statements/C-obs3.pdf.

RANZCOG. (2010). *Prenatal screening for fetal abnormalities. A statement from the The Royal Australian and New Zealand College of Obstetricians and Gynaecologists.* Retrieved from http://www.ranzcog.edu.au/publications/collegestatements.shtml.

Scriver, C. R., Bardanis, M., Cartier, L., Clow, C. L., Lancaster, G. A., & Ostrowsky, J. T. (1984). Beta-thalassemia disease prevention: Genetic medicine applied. *American Journal of Human Genetics*, *36*, 1024–1038.

Su, Y. N., Hung, C. C., Lin, S. Y., Chen, F. Y., Chern, J. P., Tsai, C., . . . Lee, C. N. (2011). Carrier screening for spinal muscular atrophy (SMA) in 107,611 pregnant women during the period 2005-2009: A prospective population-based cohort study. *PloS One*, *6*, e17067.

Sugarman, E. A., Nagan, N., Zhu, H., Akmaev, V. R., Zhou, Z., Rohlfs, E. M., . . . Allitto, B. A. (2012). Pan-ethnic carrier screening and prenatal diagnosis for spinal muscular atrophy: Clinical laboratory analysis of >72,400 specimens. *European Journal of Human Genetics*, *20*, 27–32.

Wald, N. J., George, L. M., Wald, N. M., & Mackenzie, I. (1993). Couple screening for cystic fibrosis. *Lancet*, *342*, 1307–1308.

Wilkins-Haug, L., Hill, L., Schmidt, L., Holzman, G. B., & Schulkin, J. (1999). Genetics in obstetricians' offices: A survey study. *Obstetrics & Gynecology*, *93*, 642–647.

Zeesman, S., Clow, C. L., Cartier, L., & Scriver, C. R. (1984). A private view of heterozygosity: Eight-year follow-up study on carriers of the Tay-Sachs gene detected by high school screening in Montreal. *American Journal of Human Genetics*, *18*, 769–778.

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