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# Prenatal Diagnosis and Pre-implantation Genetic Diagnosis: Legal and Ethical Issues

Roxanne Mykitiuk

*Osgoode Hall Law School of York University*, [rmykitiuk@osgoode.yorku.ca](mailto:rmykitiuk@osgoode.yorku.ca)

Stephanie Turnham

Mireille Lacroix

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Prenatal Diagnosis and Pre-implantation Genetic Diagnosis: Legal and Ethical Issues

By: Roxanne Mykitiuk, Stephanie Turnham and Mireille Lacroix

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## **Introduction**

Prenatal diagnosis of genetic disorders and foetal anomalies has expanded significantly. Hundreds of conditions can be diagnosed through DNA analysis of foetal cells and ultrasound and maternal serum biochemical screening. The purpose of prenatal diagnosis is to rule out the presence in the foetus of a particular medical condition for which the pregnancy is at an increased risk. This information is provided to the individual or couple to assist in the decision-making process regarding the possible options including: carrying the pregnancy to term, preparing for a difficult delivery, preparing for the birth of a child with genetic anomalies and for special newborn care, or terminating the pregnancy. Preimplantation genetic diagnosis (PGD) involves the creation of embryos outside the body and their subsequent biopsy, in order to test for a genetic disorder. The stated advantage of PGD over prenatal diagnosis or testing is that the genetic diagnosis takes place at a much earlier stage. As a pregnancy has not been established, couples or individuals will not have to consider abortion, which is likely to be a much more stressful and difficult decision than the disposal of affected embryos in their earliest stage of development. It is expected that the range of conditions for which PGD is available will expand as the genes implicated are identified. Prenatal genetic testing and preimplantation diagnosis raise a number of ethical and legal issues that will be discussed below following a brief description of PND and PGD techniques.

## **PND & PGD techniques**

Prenatal testing includes prenatal screening, prenatal diagnosis and preimplantation genetic diagnosis. Screening methods such as determination of maternal age, maternal serum screening and ultrasound are used to identify women who are at increased risk of having a child with a chromosomal anomaly or a congenital anomaly. If a screening test yields an abnormal result, it can be followed by prenatal diagnosis in order to establish whether the foetus does carry a specific genetic mutation or chromosomal anomaly.

### Prenatal diagnosis

Techniques currently used for prenatal diagnosis include:

- amniocentesis,
- chorionic villus sampling (CVS) and
- cordocentesis.

Amniocentesis is performed after 14 weeks of gestational age (usually between 15 and 17 weeks) in order to determine foetal karyotype and to detect the presence of molecular and biochemical abnormalities. A needle is inserted into the amniotic sac of a pregnant woman while ultrasound imaging is used to verify the position of the foetus and the location of the placenta. A small quantity of amniotic fluid is aspirated, and its level of alpha-fetoprotein is measured. The foetal cells contained in the amniotic fluid are also

cultured and analysed using karyotyping, molecular diagnosis, biochemical assays or direct fluorescent *in situ* hybridisation. Results are highly reliable, but are available one to three weeks after the amniocentesis, at 17 to 20 weeks of gestation. This is regarded by some as the most important drawback of amniocentesis: at this stage, termination of pregnancy – currently the only option to prevent the birth of an affected child – entails a greater physical and emotional risk to the woman than a first trimester abortion and is difficult to obtain in many regions of Canada and the United States [Childbirth by Choice, 2003; Henshaw & Finer, 2003]. However, the risks of the procedure itself are low. The risk of miscarriage due to amniocentesis is 0.5 to one percent, while the risk of infection and foetal injury are rare. Minor complications such as leakage of amniotic fluid, and bleeding occur in one to five percent of cases, but usually resolve themselves [Chodirker et al., 2001b; Jorde et al., 2003].

CVS is performed earlier, between ten weeks and eleven weeks six days of gestational age. It involves the collection of chorionic tissue either through a transcervical or transabdominal technique, and its subsequent analysis. The cells obtained can be analysed directly or cultured as in amniocentesis. Like amniocentesis, CVS is highly accurate, but it has the advantage of yielding earlier results. The risk of spontaneous pregnancy loss is slightly higher at one to two percent over baseline risk for transabdominal tissue collection and three to six percent with the transcervical technique. CVS may also entail a one in 3000 risk of facial or limb anomaly due to vascular disruption. In addition, diagnosis may be compromised if the sample has been contaminated with maternal tissue or if there is a discrepancy between the chromosomes

in the foetal tissue and the chorionic tissue. These difficulties can however be easily circumvented [Chodirker et al., 2001b; Jorde et al., 2003].

Cordocentesis is generally performed after 16 weeks of gestation when a rapid diagnosis is required in cases where ultrasound has revealed congenital malformations or growth retardation, to clarify suspected chromosome mosaicism, or to detect viral infections, haematological diseases, inborn errors of metabolism and maternal or foetal platelet disorders. It involves the insertion of a needle into the umbilical cord vessel with ultrasound guidance and the withdrawal of foetal blood. Cordocentesis has the advantage of allowing direct access to the foetus, but entails a slightly higher risk of pregnancy loss, as well as a five to ten percent risk of foetal distress [Chodirker et al., 2001b; Jorde et al., 2003; Simpson and Elias, 2003].

In addition to these techniques, prenatal genetic diagnosis can be conducted through foetal tissue sampling. As is the case in amniocentesis and cordocentesis, the procedure is performed under ultrasound guidance. A needle is inserted and foetal tissue such as skin, liver or foetal urinary tract is collected for analysis. The risks and complications are similar to those linked to cordocentesis. However, the procedure is not commonly practiced. Similarly, the isolation of foetal cells from maternal blood is currently being studied as a technique of prenatal diagnosis. It would have the advantage of non-invasiveness (and therefore extremely low risk) and of being conducted early in the first trimester. It is not yet available in clinical practice [Chodirker et al., 2001b; Health Canada, 2002; Jorde et al. 2003; Shinya et al., 2004].

### Preimplantation Genetic Diagnosis

In contrast with prenatal diagnosis, preimplantation genetic diagnosis (PGD) is conducted before a woman becomes clinically pregnant. It is therefore an attractive option for couples at increased risk of having a child with a genetic condition who wish to avoid the possible termination of an affected pregnancy or who oppose abortion for religious or moral reasons [Simpson and Elias, 2003; Sermon et al., 2004; Verlinsky et al., 2004]. Though it has moved from the research setting to clinical application in recent years, PGD is not widely available, and it is considerably more complex (and costly) than traditional PND methods [Health Canada, 2002; Simpson and Elias, 2003; Verlinsky et al., 2003]. It involves the creation of embryos through *in vitro* fertilisation and the analysis of only one or a few cells from these embryos (or, in some cases, from the ovum) in order to establish a genetic diagnosis.

Cells can be obtained through biopsy at three different stages: prior to fertilisation (biopsy of polar bodies), from blastomeres, and from blastocysts. For polar-body biopsy, an incision is made in the zona pellucida in order to draw the polar body out of the egg. Analysis of the polar body enables scientists to draw conclusions about the genotype of the oocyte. The utility of this technique is limited to testing for aneuploidy or to cases in which risk is known to originate from the mother's genotype since paternal mutations cannot be evaluated [Jorde et al., 2003]. In the case of blastomere biopsy, one or two cells are removed from the embryo three days after fertilisation, when the embryo is composed

of six to eight cells. A greater number of cells can be obtained for analysis through blastocyst biopsy. This strategy entails removing ten to thirty cells from the trophoectoderm rather than from the embryo itself. Biopsy is conducted five to six days after fertilisation, when the blastocyst contains over one hundred cells [Simpson and Elias, 2003; Sermon et al., 2004; Jorde et. al.].

The cells thus obtained can be analysed through various methods. Polymerase chain reaction (PCR) is used to amplify the DNA from a single cell and diagnose monogenic disorders such as cystic fibrosis, sickle-cell anaemia, and Duchenne muscular dystrophy. Specific tests have been developed for approximately 40 monogenic disorders. Though all monogenic disorders could, in theory, be detected through PCR, the cost implications are such that tests exist only for the most common disorders [Sermon et al., 2004; Wells, 23]. Fluorescence *in-situ* hybridisation (FISH) analysis can be used to detect embryo sex and chromosomal aberrations such as aneuploidy, translocations or other chromosomal rearrangements on selected chromosomes. A new technique called comparative genomic hybridisation (CGH) has also been developed to analyse the chromosomal complement of embryos. While FISH can only detect a limited number of chromosomes (between five and nine per cell), comparative genomic hybridisation enables scientists to evaluate all chromosomes simultaneously. However, because it is a lengthy process, and because there is only a very short period of time during which PGD can be performed, CGH is not widely used [Sermon et al., 2004; Simpson et al., 2003; Wells, 2003]. Once cell analysis is completed, embryos that are free from the mutations or chromosomal aberrations tested

for are implanted into the woman's uterus. In the case of polar body diagnosis, the selected ova are fertilised and then implanted.

The use of PGD can improve the outcome of a pregnancy for a couple who use *in vitro* fertilisation in order to conceive and for those who are at increased risk of transmitting a genetic disorder to their offspring [Verlinsky et al., 2004]. However, it does not guarantee the birth of a healthy child. Available data from 2001 shows that following PGD, pregnancies were established in 16 to 24 percent of treatment cycles [European Society of Human Reproduction and Embryology, 2002; International Working Group on Preimplantation Genetics, 2001] and that a small percentage of the children born as a result of these pregnancies had some form of anomaly [International Working Group on Preimplantation Genetics, 2001]. There is a slight risk of misdiagnosis in PGD, often due to allelic dropout (a problem associated with PCR amplification failures) or DNA contamination. However, techniques such as multiplex-PCR and amplification of hypervariable fragments of DNA can lower risks of misdiagnosis [Jorde et al., 2003; Sermon et al., 2004; Wells, 2003]. Though PGD is considered a safe procedure, it remains "costly and technically challenging" [Jorde et al., 2003].

### **Access to PND and PGD**

Prenatal genetic testing is not universally accessible. Many factors affect a pregnant woman's ability to access these services, including economics and clinical risk factors.

**Develop intro here**



## Evidence-Based Access: Standards of Usefulness

Whether or not a woman or couple is offered prenatal testing can depend on whether the health provider believes she meets the relevant risk threshold. Evaluating the clinical necessity of intervention is generally considered an appropriate strategy for allocating scarce medical resources [AMA Code of Ethics, s.E-2.03]. It is also a way to avoid unnecessary invasive tests. Concerns have been raised about recent direct-to-consumer marketing approaches which may cause increased demand on the part of consumers and lead to indiscriminate offers of testing without an appropriate risk assessment [Andrews and Zuiker, 2003; Holtzman and Watson, 1997]. On the other hand, it has recently been argued that the evidence to support the cost utility analysis of the risk thresholds is deficient, and that such thresholds can threaten autonomous choice; accordingly, it is argued that all pregnant women should be offered prenatal testing regardless of risk [Harris et al., 2004]. In any event, risk thresholds remain an important determining factor in access to prenatal testing, and a number of organizations have issued guidelines to assist health care professionals and geneticists in assessing the risk factors. Some risk factors can be identified by asking the right questions, including questions about maternal age, family history of genetic disorders, previous children, ethnic background, and medical and obstetric history [Roop, 2000; SOGC, 1998]. Another way to screen for risk factors is through medical examination, including maternal serum screening and ultrasound. In practice, these various methods are often combined into an overall risk assessment. Evidence-based standards of access arise in the initial screen stage.

Screening cannot detect the presence of a disorder; it only identifies women who are at increased risk of having a child with a genetic anomaly.

### Practice Guidelines

Ethnicity. Certain ethnic backgrounds are associated with an increased risk of genetic disorder. For example, Tay-Sachs disease has been linked to Ashkenazi Jews and French Canadians in Eastern Quebec; and individuals of other than northern European descent are considered at high risk for thalassemia. It is generally recommended that genetic screening be uniformly offered to patients with high risk ethnic backgrounds [SOGC, 1998; AMA Code of Ethics, s.E-2.137].

With respect to CF in particular, a National Institutes of Health (NIH) Consensus Panel originally issued a statement supporting access to CF testing for all pregnant couples, especially those in high risk populations [NIH, 1997]. Following this, a Steering Committee representing the interests of ACOG, ACMG and NHGRI was established to refine the recommendations, leading to creation of general guidelines [Grody et al., 2001], laboratory standards [Richards et al., 2002], and patient education materials [ACOG, 2001a; ACOG, 2001b]. Overall, the materials recommend offering CF testing to couples in high risk ethnic groups who are pregnant or planning pregnancy, and making it available along with information about its limitations to couples in other ethnic groups with lower risk. Given that knowledge of one's carrier status can have implications for family planning [Lafayette et al., 1999], it is recommended that members of high risk ethnic groups be tested for carrier status prior to conception, to allow for genetic

counselling at an early stage [Grody et al., 2001; Chodirker et al., 2001a]. According to Canadian guidelines, it is preferable for both partners to be tested, but knowledge that the woman is a carrier can still be an indication for prenatal diagnosis [Chodirker et al., 2001a].

**Family History.** It is widely agreed that a recurring genetic disorder in a family's history is "a reasonable indication of risk" [Roop, 2000; Holtzman and Watson, 1997].

According to the AMA Code of Ethics, "prenatal genetic testing is most appropriate for women or couples whose medical histories or family backgrounds indicate an elevated risk of fetal genetic disorders" [AMA Code of Ethics, s.E-2.12]. Physicians should therefore take a full family history, which includes inquiries about siblings, parents, parents' siblings, grandparents, and other extended family members where appropriate. However, family history does have limitations, particularly with respect to reliability and the different modes of inheritance [Holtzman and Watson, 1997]. Canadian practice guidelines appear to envision a spectrum, ranging from cases where prenatal testing must be offered (for example, where the woman is a carrier for Down syndrome) to cases where further information or consultation with the patient is required (for example, where one relative is found to have Down syndrome) [Chodirker et al., 2001a].

**Pregnancy History.** Relevant pregnancy history factors include having a previous stillborn birth or a liveborn infant with a chromosomal anomaly. Invasive prenatal testing should always be offered in these cases because there is an increased risk of recurrence. Even when the prior infant had a *de novo* anomaly (i.e. where the parents have normal

karyotypes), prenatal testing is offered because of the potential for parental germline mosaicism [Chodirker et al., 2001a]. Previous environmental exposure can also be relevant [Holtzman and Watson, 1997].

**Maternal Age.** It is traditionally recommended that all women over the age of 35 be offered prenatal testing, given the increased risk of fetal aneuploidy [ACOG, 2001]. However, the particular age cut-off has been described as “somewhat arbitrary,” and the best practice may be to look at all the factors surrounding a woman’s pregnancy [Roop, 2000]. Canadian practice guidelines note that maternal age alone is a relatively poor predictor of fetal anomaly, and it may be inappropriate to rely on it when facilities exist for additional screening methods [Chodirker et al., 2001a]. Notably, there is a trend towards women extending their childbearing years, which means that invasive testing would be offered to an increasing number of women, with questionable utility, in the absence of further risk refinement. A recent study found that there was a reduction in the number of women electing to have invasive testing despite the rising maternal age, and the researchers attributed this to a departure from using maternal age alone as a risk assessment tool.

Advanced paternal age is also associated with an increased risk of genetic anomalies. A frequently used cut-off is 40 years of age at the time of conception, but the ACMG recognizes that risk increases linearly with age. Advanced paternal age is associated with a wide variety of genetic disorders, many of which cannot be detected by ultrasound. Therefore, the ACMG recommends genetic counseling be tailored to the individual needs

of couples [ACMG, 1996].

Ultrasound Examination and Biochemical Markers (Maternal Serum Screening). Genetic assessment is recommended when an ultrasound scan reveals major fetal anomalies.

There are also several minor fetal anomalies, or “soft signs,” that are statistically linked to chromosomal anomalies, but there is controversy about whether these soft signs have enough positive predictive value to warrant genetic testing. [Chodirker et al., 2001a].

The maternal age risk factor for Down syndrome can be refined through the use of maternal serum screening for biochemical markers in the first or second trimester. The ACMG recommends that women who screen positive for trisomy 21 or 18 be offered genetic counselling and amniocentesis [Driscoll, 2004]. The ACMG also recommends that women be offered maternal serum screening to identify pregnancies at risk of neural tube defects and anencephaly. In these cases, the risk assessment is refined by factors such as maternal weight, race, and family history, and an elevated result is an indication for genetic counseling and additional testing [Driscoll, 2004]. There are numerous combinations of markers, and second trimester screening through the “triple test” or “quad screen” has been found particularly useful in detecting Down syndrome and trisomy 18 [Javitt, 2004]. First trimester screening methods use a combination of serum screening and ultrasound nuchal fold translucency screening. These methods have comparable detection rates of second-trimester screens and offer the advantage of an earlier test, but should not be offered routinely unless there is sufficient ultrasound training and quality control [Driscoll, 2004; Dolan, 2004]. Recent studies have found that

“fully integrated screening,” which combines first and second trimester screening methods with risk assessment from maternal age, outperforms second trimester screening alone [Dolan, 2004]. The extent to which these methods are routinely offered will depend on many policy issues. While they offer a superior risk assessment than maternal age alone, there is a concern that some women may rely too heavily on the screen result at the expense of seeking a further diagnostic test. It is also relevant to consider whether a woman who screens positive will be able to access first-trimester diagnostic testing [Javitt, 2004].

Other Medical Factors. Given the variety of genetic disorders that can be detected prenatally, there are other indications of risk that go beyond the scope of this chapter. Health care providers should consult the guidelines and other medical literature for further information. For example, the ACMG has released a number of guidelines with respect to specific disorders, such as Fragile-X [Maddalena et al., 2001], uniparental disomy [Shaffer et al., 2001], and Prader-Willi and Angelman syndromes [ASHG and ACMG, 1996]. There are also clinical practice guidelines for specific testing techniques, such as FISH [Watson et al., 2000].

#### Obligations of the Health Care Provider

The clinical guidelines are significant for healthcare providers because they provide a baseline standard of care for offering access to prenatal testing services. Physicians are under both a legal and ethical obligation to ensure that patients have access to necessary or beneficial services. It is therefore necessary for healthcare providers to be aware of the

clinical indications, to identify them in patients, and to discuss the potential implications with the patient. Under ethical guidelines, physicians are required to discuss prenatal testing with “all women who have appropriate indications” [SOGC, 1998]. The AMA Code of Ethics also notes that women or couples who do not meet the risk threshold may request prenatal diagnosis as long as they are aware of the risks involved [AMA Code of Ethics, s.E-2.12]. However, physicians are not obligated to perform the service if they lack the skills or knowledge or have a conscientious objection, as long as they engage in discussion with the patient or offer a referral. Where a physician feels that he or she lacks the necessary skills or competence to perform a genetic test, it is appropriate to offer referral elsewhere, such as to a clinical laboratory [Grody et al., 2001]. Where a physician has a conscientious objection to genetic testing or abortion, physicians must at least alert the parents of a potential problem, presumably using the risk factors as guidelines, so that the parents may seek genetic counseling elsewhere [AMA Code of Ethics, s.E-2.12]. Canadian guidelines also explicitly recognize the duty to offer referral to an alternative provider when the opportunity for a full and frank discussion is constrained. “Failure to discuss all options with women at risk or, alternatively, to refer them further, is unethical” [SOGC, 1998].

In conjunction with the ethical duty to make testing available, the courts have recognized that there is a legal duty to offer tests from which patients might benefit [Andrews and Zuiker, 2003]. The above-noted clinical utility guidelines have come up in some cases. For example, a physician may be liable for failure to warn of increased risks due to advanced maternal age [Becker v. Schwartz, 1978], failure to sufficiently take into

account ethnic background as a risk factor [Naccash v. Burger, 1982], failure to diagnose a condition in a previous child that would have provided genetic information relevant to a second child [Schroeder v. Perkel, 1981], and failure to offer testing when there is a known family history [Philips v. U.S., 1981] or a previous child born with a genetic disorder [Keel v. Banach, 1993]. Claims in the context of prenatal testing normally arise under “wrongful birth” and “wrongful life” litigation, discussed in detail below, so the outcome of such cases will depend on the particular facts of each case and the extent to which it fits within the framework of these causes of action. The threat of legal action can have a significant impact on the physician-patient relationship. Physicians who practice in states that recognize wrongful life and wrongful birth actions are more likely to offer prenatal testing [Roop, 2000]. The threat of malpractice litigation is a daunting prospect for many health care providers, who may feel the need to practice defensive medicine [Mavroforou et al., 2003]. At the same time, it is important to note that case law should not be relied upon as the source for best practice standards, because “case law is primarily reactive.” It only “speaks to the minimum standard of professional behaviour” and it is likely that the public and medical community will expect an even higher standard in practice [Botkin, 2003].

There remains a question about whether the obligation to offer a test will depend on its predictive validity. The majority of cases have dealt with single-gene conditions and highly predictive tests, and it may be that a different standard will apply for more complex genetic conditions and tests of lower predictive validity [Andrews and Zuiker, 2003; Simmons v. W. Covina Medical Clinic, 1989]. Such an approach may be



shortsighted, given that many patients will value the knowledge obtained from such tests, and will want to hold healthcare professionals accountable for not providing such information [Andrews and Zuiker, 2003].

### Public Policy Concerns

Recent advances in prenatal diagnosis and assisted reproductive technology [ART] have given rise to a number of ethically challenging cases with respect to access rights. While healthcare providers can refer to a general standard of care where guidelines exist as to clinical risk measurement, there is confusion and debate about the appropriateness of offering testing in other cases. Some countries offer specific guidelines on access, but others, including the U.S., have few laws governing ethically complex cases. Instead, it is often left to clinical judgment [Stern et al., 2003; Adams, 2003].

### Sex Selection

The issue of whether parents ought to be able to use genetic testing to select their fetus on the basis of sex is controversial, and can depend on ethical, cultural and religious perspectives. Sex selection can occur through one of three methods: 1) prenatal diagnosis of fetal sex, followed by termination of pregnancy if it is the undesired sex; 2) preimplantation sex diagnosis followed by selection of an embryo of the desired sex; or 3) prefertilization techniques such as sperm separation. However, the latter option is of relative recent development, and its reliability and safety have not yet been proven [Morales et al., 2004].

The primary medical indication for sex selection is to identify fetuses or embryos that may be afflicted with a sex-linked disorder. While the termination of a fetus continues to raise ethical concerns, the use of PGD to prevent transmission of a serious sex-linked disorder is widely considered ethically permissible [ASRM Ethics Committee, 1999; Morales et al., 2004; Csaba and Papp, 2003]. Sex determination may be the only reliable method of identifying sex-linked disorders [Morales et al., 2004].

More ethically problematic cases are those in which prospective parents request sex selection for personal or cultural reasons, such as preferring a child of a certain sex or desiring a sex-balanced family. These motivations raise a host of concerns, including gender bias and discrimination, allocation of scarce medical resources, risk of psychological harm to children, and overall impact on the human sex ratio [ASRM Ethics Committee, 1999]. Sex selection for family balancing purposes is often distinguished from an inherent sex preference. The latter is a particular concern in societies and cultures that have historically shown a preference for male over female children, such as China and India, where screening techniques, abortion, and infanticide have contributed to an imbalanced sex population. While there is no such evidence of sex-preference in North America [Royal Commission on New Reproductive Technologies, 1993; Steinbock, 2002], there remains a concern that allowing sex selection of the first child will reinforce negative attitudes and contribute to sex-discrimination [Morales et al., 2004]. With respect to family balancing, some have suggested that it is justified as long as one or more children are already born, since this would not involve an inherent

favouring of one sex over another [Robertson, 2003]. Appropriate standards are difficult to tease out in this area, for ethical doctrines can be invoked on both sides of the argument. For example, it is argued that modern human rights demand that women not be compelled to maintain pregnancies against their will. On the other hand, human rights are also invoked as the basis for prohibiting sex selection, which is held to violate the right to non-discrimination on the basis of sex [Cook et al., 2001].

Sex selection for non-medical reasons is not officially regulated in the United States, but it is prohibited in other countries. For example, in March 2004, the Canadian federal government enacted Bill C-6, which provides that:

5. (1) No person shall knowingly [. . .]
  - (e) for the purpose of creating a human being, perform any procedure or provide, prescribe or administer any thing that would ensure or increase the probability that an embryo will be of a particular sex, or that would identify the sex of an *in vitro* embryo, except to prevent, diagnose or treat a sex-linked disorder or disease.

Contravention of this provision carries serious penalties including the possibility of imprisonment. Unfortunately, the provision is not as clear as it could be, and leaves loopholes for non-medical sex-selection during prenatal fetal diagnosis.

While it unmistakably prohibits sex-selection for non-medical purposes at the *in vitro* embryo stage, it leaves loopholes for non-medical sex-selection during prenatal diagnosis. For one thing, the provision refers only to embryos, which are defined under

the statute as organisms during the first eight weeks of pregnancy. It appears that screening the fetus after eight weeks for the purposes of sex-selection is not expressly prohibited. There is also no mention of what happens when sex is determined incidentally (for example, during ultrasound for another purpose). Is the doctor either permitted or obligated to disclose this information to the patient?

In the U.S., the Ethics Committee of the ASRM released a policy statement discouraging the use of PGD for non-medical reasons, given concerns about the burdens and costs it imposes [ASRM Ethics Committee, 1999]. The AMA Code of Ethics also censures “selection on the basis of non-disease related characteristics” [AMA Code of Ethics, s.E-2.12]. Several world organizations also discourage sex selection for non-medical reasons [Council of Europe, 1996; World Health Organization, 1999; International Bioethics Committee of UNESCO, 2003]. Nonetheless, these guidelines are not enforceable as law, and there is evidence that a number of clinics are advertising and offering IVF services for the purpose of sex selection.

The emergence of preconception selection techniques, which do not involve discarding embryos or fetuses, has added a new dimension to the ethical debate. The Ethics Committee of the ASRM and others have noted that if pre-fertilization techniques were found to be safe and reliable, it may be ethical to use them for family balancing purposes [ASRM Ethics Committee, 2001; Robertson, 2001].

The sex of a fetus may be determined incidentally to another procedure, such as fetal karyotyping, and a parent may or may not wish to be informed of the sex. As will be discussed below, women have the right to refuse information, just as they have the right to receive it. Therefore, for both legal and ethical reasons, it is best to ask patients in advance whether they want the sex to be disclosed, and to allow them to opt not to know the sex of an embryo or fetus [Mavroforou et al., 2003; FIGO, 1991]. Conversely, the ACOG Committee on Ethics recommends that this information not be withheld from a pregnant woman who requests it, given that she has a right to her own medical information [Morales et al., 2004]. If a physician is uncomfortable with the possibility of indirectly contributing to sex selection, he or she ought to clarify the patient's goals in advance, and explicitly inform patients of any procedures they are not willing to perform [Morales et al., 2004].

#### Testing for Susceptibility and Late-Onset Conditions

A further ethical concern is the use of PGD to test for genes that increase one's susceptibility to disease, such as cancer, or late-onset conditions, including Huntington's disease and Alzheimer's disease. The two situations are somewhat different: late-onset conditions have full penetrance and are generally not preventable, while susceptibility genes do not have full penetrance and may be amenable to treatment. However, it has been argued that the ethical issues are similar in each case, given the substantial, non-trivial burden imposed by the diseases, which may serve to justify testing [Sermon et al., 2004; Robertson, 2003]. The discomfort with susceptibility testing relates to the use of PGD to summarily discard embryos that are only associated with an increased risk, not

certainty, of disease [Genetics and Public Policy Center, 2004]. Concerns about adult-onset testing include the fact that a child could enjoy years of healthy life, and that a treatment or cure may yet be found [Genetics and Public Policy Center, 2004]. Late-onset testing also has an additional “ethical twist” in that the prospective parent will themselves be a carrier of the disease because the genes are dominant. The shortened life span of the parent is a concern because the child may lose the parent while still dependent on them [Robertson, 2003; Towner and Loewy, 2002]. However, this may still be an ethically justifiable procedure, given that people with disability have no less interest in reproduction than others, and that denying services in this case could create a slippery slope to other restrictions [Robertson, 2003].

The tests are currently legal in the U.S., subject only to gene patent restrictions [Robertson, 2003], but this does not mean that practitioners are obligated to provide the service. According to Botkin, the adult onset risk is not sufficient to warrant this type of testing as a standard of care [Botkin, 2003]. Some ethical committees feel that it is too early to offer a clear opinion on the acceptability of these tests, but an ESHRE Ethics Task Force noted that PGD for adult onset conditions is acceptable, in spite of limited knowledge about potential therapy in the time gap before onset [Shenfield et al., 2003]. A UNESCO committee has suggested that susceptibility testing should be given a low priority and restricted to cases of high risk of severe disease [International Bioethics Committee of UNESCO, 2003]. The NSGC has urged caution in the use of adult-onset testing. Genetic counseling is essential, and must include discussion of psychological and social risks and benefits, including the potential for future discrimination and the

possibility of disclosing carrier status of other family members. Parents ought to consider whether the decision to test should be reserved for their offspring to make upon reaching adulthood [NSGC Position Statement, 1995].

### Selecting for Disability

A difficult issue arises when a woman or couple, who themselves possess a disability or a unique characteristic, wish to have a child with that same characteristic. For example, some deaf couples have expressed a desire to use prenatal testing or PGD to select a child with deafness. For them, deafness is not considered a disability, but rather a defining feature of their cultural identity. Similar cases could arise, including dwarves wishing to select a dwarf child, or people with an intellectual disability wishing to select a child with the same disability [Savulescu, 2002].

A deaf lesbian couple attracted much criticism in the U.S. when they attempted to have a deaf child by intentionally using a deaf sperm donor [Savulescu, 2002]. One argument repeatedly raised is that it was a selfish action on the part of the parents. It is argued that intentionally selecting for deafness is not in the best interests of the child, because it restricts the child's future options and thereby threatens her autonomy [Davis, 1997]. Even a member of the National Association of the Deaf had trouble understanding the couple's decision because deaf people "don't have as many choices" [Spriggs, 2002].

On behalf of the parents, it has been argued that deaf people can be considered a minority group who merely suffer disadvantage as a result of societal discrimination, not of

deafness itself. “In this sense, deafness is strictly analogous to blackness” [Levy, 2002].

Second, it is questioned whether a child selected for deafness actually suffers harm as a result. She is not worse off than she would have been—as might be the case if the parents had refused to allow treatment for a correctable condition—because the alternative is that she would never have been born; a different child would have been selected instead.

Third, there is an argument based on freedom of choice. Although some members of society may view deafness, or dwarfism, or intellectual disability in negative terms, these value judgments should not be imposed on the couple making the reproductive decision. This argument reminds one of the terrors of the Nazi eugenic programme, during which couples deemed “unfit” to create the perfect child were forced to undergo sterilization [Savulescu, 2002]. Indeed, parental choices will influence most of the important aspects of a child’s life—from education, to religion, to social interaction—which in turn form the basis for the child’s freedom. It is difficult to draw the line between those parental decisions which are the preconditions for the child’s freedom of choice, and those which foreclose too many options for the child [Levy, 2002].

Few ethical guidelines have been established in this area. UNESCO’s International Bioethics Committee rejects the use of PGD to purposely select a child with a genetic disease, and it includes deafness and dwarfism in its definition of disease. This practice is held to be “unethical because it does not take into account the many lifelong and irreversible damages that will burden the future person” [International Bioethics Committee of UNESCO, 2003]



### Potential Stigmatization of Persons with Disabilities

It is important to recognize that for some, the very practice of PND is problematic and raises challenging ethical issues. This means that healthcare providers and patients can face conflicting messages about the value and acceptability of these tests. The disability rights movement has provided a strong critique of prenatal genetic testing, drawing heavily on concerns about its eugenic implications. Parens and Asch provide a useful summary of the main arguments. First, it is argued that prenatal testing and selective abortion are against public policy because such practices can lead to stigmatization of individuals living with disability. In particular, there is a worry that the aim of genetic testing is “to eradicate disability and reduce the number of births of genetically disabled individuals” [DeVaro, 1998]. This is said to be harmful in the message it sends about the worth of a life with disability and for ignoring the rich life experiences and useful contributions that disabled individuals can bring to society. In response, proponents of prenatal testing object that the purpose of the procedure is not to eradicate disability, but rather to increase the reproductive autonomy of women. Also, prenatal testing does not necessarily entail termination of the fetus; it can provide other useful information and allow for interventions [Chen and Schiffman, 2000; Mahowald, 2003]. However, in practice there is clearly a tension between the goals of enhancing reproductive choice and preventing disability, particularly given evidence that the success of some genetic screening programs is measured in terms of reduction in disability [Parens and Asch, 2003; Beaudet, 1990], and that some women perceive pressure from the social system and health professionals to undergo testing and to terminate for disability [Fox, 2002; Mahowald, 2003]. Second, there is a concern that the “selective mentality” of prenatal

testing, in which parents strive for perfection and lament over disability, fosters a morally deficient view of parenthood [Parens and Asch, 2003]. On the other hand, a distinction can be drawn between parental motivations during prenatal testing of a “potential child” and parental care for a disabled newborn [Mahowald, 2003]. Third, disability rights advocates argue that genetic testing is based on a misunderstanding about what life is like for individuals living with disability. While it is true that some families experience stress and disruption, on average the needs of families with and without disability can be “strikingly similar” [Parens and Asch, 2003].

Some suggest that the practice of selective abortion for disability can be criticized on similar grounds as sex selection. Wolbring notes, for example, that both practices raise concerns about “significant threats to the well-being of children, the children’s sense of self-worth, and the attitude of unconditional acceptance of a new child by parents.” Moreover, concerns about stereotypes, discrimination and oppression of persons of the unwanted sex can apply equally to stereotypes, discrimination and oppression of individuals with the unwanted disability [Wolbring, 2003; Wong, 2002]. According to Wolbring, the only way to justify disability deselection, while at the same time prohibiting sex-selection, is by “arbitrarily” labeling disabilities and diseases as medical problems, thereby applying a different moral standard to them. He suggests that this approach is buttressed by the marginalization of people with disabilities and the corresponding exclusion of disability rights critiques from the bioethics discourse [Wolbring, 2003]. It is therefore important to take seriously the disability rights critique

so as to prevent a purely medical model of genetic testing from standing in for a rights-oriented model.

### Where Do We Draw the Line?

Parens and Asch set up a working group to compare and contrast the disability rights critique and the advocacy of prenatal genetic testing and to offer recommendations. Although the group reached consensus on a number of issues, they reached an impasse at the critical question: “is there a helpful and rational way to distinguish between tests that providers should routinely offer and those they should not?” According to the group, it comes down to whether a test is judged reasonable or unreasonable under the circumstances, and such line-drawing can be an impossible task [Parens and Asch, 2003; International Bioethics Committee of UNESCO, 2003]. It is highly dependent on individual ethical judgment [Garel et al., 2002]. However, many members of society would value some kind of line-drawing, whether for ethical reasons [Henn, 2000] or to limit malpractice liability among the medical profession [Botkin, 2003]. Disability groups also fear that a lack of guidelines will encourage more testing, and potentially greater intolerance of disability [Parens and Asch, 2003; Wasserman, 2003]. Accordingly, there have been some attempts to draw workable lines between tests or conditions [Murray, 1996; Strong, 1997; Botkin, 2003; Council on Ethical and Judicial Affairs, 1993]. For example, Botkin draws a line between childhood conditions that have significantly adverse effects on the parents (“in terms of heartache, worry, time, effort, and money”) and those that do not. Taking a cue from tort law, he suggests that the magnitude of the potential challenge in raising the disabled child can be one criterion for offering prenatal

diagnostic testing [Botkin, 2003]. However, the definition of precisely what constitutes a serious parental challenge, and any other potential considerations, will need to be fleshed out with further ethical guidelines.

### **Abortion Law**

A discussion of prenatal diagnosis would not be complete without a discussion of the legal framework governing access to abortion. Until 1988, abortion was regulated as a criminal act in Canada. The *Criminal Code* established a set of rules pursuant to which women could have limited access to therapeutic abortions in Canada. It provided that an abortion could only be performed by a qualified physician in a hospital accredited by the provincial government. In addition, a therapeutic abortion committee had to certify that continuing the pregnancy would or would likely endanger the patient's life or health. The committee was to be composed of three additional physicians from the hospital in which the procedure was to be performed. Abortions that did not meet these criteria carried the possibility of life imprisonment for the physician and 2 years' imprisonment for the female patient [s. 251].

However, in 1988, following a constitutional challenge, the Supreme Court of Canada struck down section 251 of the *Criminal Code*, stating that it violated the right to security of the person as guaranteed by the *Canadian Charter of Rights and Freedoms* because of the procedural defects its application entailed. One Supreme Court judge also concluded that s. 251 violated women's freedom of conscience and religion, as well as their right to

liberty [R. v. Morgentaler, 1988]. The Court did not conclude that abortion could not be criminalised nor that women had a positive right to access abortion services; these issues remained open. In 1990, the federal government attempted to pass a bill that would have re-criminalised abortion, but this effort was defeated in the Senate in 1991 [Bill C-43]. Subsequent federal governments have not attempted to legislate abortion since then. Consequently, abortion is not a crime in Canada if it is performed by a qualified medical practitioner, with the consent of the woman undergoing the procedure [Rodgers, 2002].

This does not mean, however, that abortion is completely unregulated. On the contrary, it is regulated in the same complex manner as all other medical procedures and therefore subject to provincial and territorial laws governing health care professionals, hospitals and access to health care services [Rodgers, 2002; Farid, 1997]. The provinces and territories have adopted different approaches to the issue of abortion, leading to unequal access to these services across Canada.

For example, the funding of abortion services varies among provinces and territories. Provincial health insurance plans in British Columbia, Alberta, Ontario and Newfoundland and Labrador cover the full cost of abortions performed in both hospitals and clinics. Those in Saskatchewan, Manitoba, New Brunswick, Prince Edward Island and the territories fund only abortions performed in hospitals. Québec and Nova Scotia provide full funding for abortions performed in hospitals, but partial funding only for those performed in clinics [Childbirth by Choice, 2003].

While some provinces such as British Columbia and Ontario have taken steps to protect women's right to access abortion services without undue interference [Access to Abortion services Act; R. v. Lewis; Ontario (A.G.) v. Dieleman], in other jurisdictions legislatures have limited access by imposing funding criteria such as the requirement to obtain certification by a committee of physicians that the abortion is necessary because the health of the woman and/or foetus is at risk [Health Services Payment Regulation; Morgentaler v. Prince Edward Island (Minister of Health and Social Services), 1995; Moulton, 2003].

Access to abortion services is also hampered by the lack of availability of services. In 2003, only 17.8 percent of hospitals in Canada were providing abortion services [Canadian Abortion Rights Action League, 2003]. Abortion clinics provide services to a significant number of women, but they are not found in every province or in the territories. Abortion services, whether in hospitals or clinics, are concentrated in large urban centres, leaving many regions under-serviced. In addition, abortions are not performed at all in Prince Edward Island or in Nunavut [Childbirth by Choice, 2003]. In many jurisdictions, this lack of services results in long waiting lists and compels women to travel to another region or province to obtain an abortion [Childbirth by Choice, 2003]. This may constitute a significant barrier to access given the costs of travel as well as the delay involved.

This has serious implications for couples who wish to terminate a pregnancy after the prenatal diagnosis of a genetic disorder. Moreover, in many regions of Canada, abortions

are performed only up to 12 or 16 weeks' gestation. Though some hospitals do perform abortions after 20 weeks in cases of severe disability or risk to the woman's life or health, these are limited in number [Childbirth by Choice, 2003]. The delay required to conduct prenatal testing and obtain results combined with the waiting list for abortion services may significantly limit the woman's or couple's choices.

Since the landmark case of *Roe v. Wade*, women in the United States have a constitutional right to abortion. In that case, the United States Supreme Court held that the right to privacy included the right to decide to have an abortion. However, this latter right was not absolute; it could be limited by a compelling state interest. The Court went on to devise a rule aiming to balance a woman's right to privacy with the state's interest in protecting her health and its interest in the potentiality of human life. The result was a rule based on a recognition of increasing state interest as the pregnancy progressed.

During the first trimester of pregnancy, the woman's right to privacy was prevalent and the decision to abort was left to the medical judgement of the woman and her physician. After the end of the first trimester, the state could regulate abortion "to the extent that the regulation reasonably relate[d] to the preservation and protection of maternal health." Finally, after the fetus became viable, the state could limit and even proscribe abortion, except where it was necessary to protect the life or health of the mother [*Roe v. Wade*, 1973].

The *Roe v. Wade* decision did not settle the abortion issue in the United States. On the contrary, it opened up a highly polarized debate. States began passing laws restricting

access to abortion through a variety of means, including the restriction of funding, the imposing of counseling and waiting periods, the requirement of parental consent for minors and the prohibition of certain procedures [Green & Ecker, 2004; Hull & Hoffer, 2001; Peterson, 1996; Partial Birth Abortion Act of 2003; Alan Guttmacher Institute, 2004b]. Some of these laws have been successfully challenged, though many have been upheld by the Supreme Court. The Court has somewhat eroded the Roe decision through its subsequent rulings, but its central finding - that a woman has a right to choose abortion before the fetus becomes viable - still holds [Peterson, 1996]. States are therefore free to prohibit all late-term or post-viability abortions, except where the woman's life or health is in danger. Nineteen states have adopted such statutes and seventeen others have adopted statutes that do not meet the Supreme Court's criteria because they permit abortion only where there is a threat to the life of the woman or because they prohibit abortion after a specific gestational age [Alan Guttmacher Institute, 2004a]. The problem in the context of prenatal genetic diagnosis is that almost all these states do not recognize an exception in cases of fetal anomaly. Because of the gestational age at which genetic tests can be safely and effectively conducted and the time required to obtain results, some women may be placed in a situation where their state laws prohibit them from choosing to terminate their pregnancy. This situation may become increasingly common as technology enables the medical profession to push the fetal viability limit earlier in the gestation period.

A number of additional barriers limit women's access to abortion in the United States [Henshaw & Finer, 2003]. The first of these is financial: many states do not provide any



public funding for abortions [Peterson, 1996]. Women or couples who do not have private health insurance therefore have to pay for the procedure themselves. Secondly, the number of physicians who provide abortion services is limited. The violence of some elements of the anti-abortion movement in the 1990s and the lack of training in medical schools has had a significant impact on the shortage of providers. In many regions, women may have to travel more than 50 or 100 miles to find a physician or a clinic [Finer & Heanshaw, 2003; Henshaw & Finer, 2003; Joffe, 2003; Peterson, 1996; Dresser, 1994]. This, compounded with legal barriers such as the obligation imposed on physicians to provide counseling and to require patients to wait 24 hours before the procedure, or alternatively, to evaluate fetal viability, creates a substantial burden for patients who already are in a difficult situation [Henshaw & Finer, 2003].

### **Informed Consent and Genetic Counseling**

**As discussed in Chapter \_\_\_\_**, health care professionals have both a legal and ethical duty to ensure informed consent before providing treatment. In the context of PND and PGD, informed consent recognizes a woman's right to reproductive autonomy, which means both that women have the freedom to choose whether to procreate or not procreate [Strong, 2003], and that they have the right to refuse medically indicated testing [Morales et al., 2004]. Reproductive autonomy is promoted when women or couples are provided information relevant to decisions about whether to continue the pregnancy [Strong, 2003]. The duty demands a full disclosure of information to the patient, including available tests, alternatives, risks and benefits, and outcomes.

Ethical guidelines recognize the importance of ensuring informed consent to prenatal testing [ACOG Code of Ethics, s.I(5); NSGC Position Statement, 1991; NSGC Code of Ethics, s.II(3); AMA Code of Ethics, s.E-2.12]. For example, the ACOG Code of Ethics requires obstetrician-gynecologists to disclose all relevant medical facts, including “alternative modes of treatment and the objectives, risks, benefits, possible complications, and anticipated results of such treatment” [ACOG Code of Ethics, s.I(5)]. The ACOG also recognizes that informed consent implies not only provision of information, but also free choice and active decision-making on the part of the patient; it is “not only a ‘permitting’ but a ‘doing’” [Morales et al., 2004]. Where patients are referred to a clinical laboratory, the responsibility for obtaining informed consent remains with the health professional, though laboratories may be required to document the consent depending on jurisdiction. In turn, laboratories must provide sufficient information to doctors to facilitate the informed consent process [Bradley et al., 2004; Richards et al., 2002; Holtzman and Watson, 1997].

Courts have also noted the importance of providing information relevant to reproductive decisions. As one court noted, “society has a vested interest in reducing and preventing birth defects,” and the failure to perform a procedure “which would have yielded information material to the parents’ decision whether to abort the fetus, constitutes a breach of . . . duty” [Blake v. Cruz, 1984]. Therefore, healthcare providers are under an obligation to provide information, and can face legal liability for failure to enter a full discussion with the patient. Indeed it has been noted that most medical malpractice suits

are based on inadequate communication between healthcare provider and patient, rather than error [Mavroforou et al., 2003]. In the context of prenatal care, the legal duty to provide information has been well-established in “wrongful birth” cases, in which the parents allege that the physician’s negligence deprived them of the opportunity to make a reproductive choice regarding conception or termination.

New and difficult questions about informed consent arise in the context of PND and PGD. Prenatal diagnosis is an evolving and complex procedure, characterized by “inevitable ambiguity, uncertainty, and difficult decision making,” and involving many ethical dilemmas, social implications, and grave potential for diagnostic errors that are not seen in other kinds of medical intervention [Strauss, 2002]. Perhaps the most unique aspect of genetic testing is that the desirability of treatment depends highly upon moral and ethical judgments; in other words, “there is no single “right” answer for all women and couples, only answers that are right for the individual woman or couples, based on personal circumstances and values” [Royal Commission on New Reproductive Technologies, 1993]. Respect for an individual or couple’s values and beliefs is of “paramount importance” [Holtzman and Watson, 1997]. This may give rise to unique legal and ethical models of informed consent. For example, while Canadian courts typically rely on what a “reasonable” patient or physician would decide, it has been argued that a “genetics model of informed consent” has evolved which is more about incorporating the decision-maker’s (i.e. the patient’s) personal values into the process of decision-making [Sharpe, 1997].

The recognition of differing value judgments forms the basis of the principle of non-

directiveness, which is central to genetic counseling. According to this principle, the role of the health professional is:

. . . not to lead clients to make particular decisions or choices (those preferred or recommended by the clinician, the health service or by society) but to help them to make the best decisions for themselves and their families as judged from their own perspectives [Clark, 1997].

The AMA Code of Ethics notes that counselors should not substitute their own personal values and moral judgment for that of the prospective parent [AMA Code of Ethics, s.E-2.12]. It must be remembered that not all women are the same, and that a woman and her physician may have radically different perceptions of risk and burden [Csaba and Papp, 2003]. The counselor or physician should therefore ascertain the patient's personal values and expectations and use them as the framework for the decision-making process. Several reasons for the emphasis on non-directiveness have been identified, including: respect for individual autonomy; a desire to disassociate current genetic practices from the history of eugenics; prevention of over-involvement by health care providers; and protecting professionals from legal action for medical decisions [Williams et al., 2002]. However, as will be discussed below, there are questions about whether non-directive counseling is possible, or even desirable, in practice.

### Barriers to Informed Consent in the PND Context

Before delving into the requirements for informed consent, it is important to examine the unique barriers to informed consent that arise in the context of prenatal diagnosis. Several

studies have unfortunately revealed that many women are not given the tools to exercise an informed choice [Marteau and Dormandy, 2001; Kohut et al., 2002], which no doubt reflects the extent to which health care providers and patients are struggling to grapple with some of these barriers. First, this area of medicine is highly complex, involving a variety of different screens and tests and a multitude of detectable anomalies. The general public may lack knowledge of the intricate science of genetics [Lanie et al., 2004]. Also, given that prenatal testing involves probabilities instead of certainties, patients can feel frustrated by the ambiguity and vagueness of the genetic predictions when attempting to make an informed decision [Royal Commission on New Reproductive Technologies, 1993]. Discussions of prenatal diagnosis will be unique from other types of medical discussions because many prospective parents will have no first-hand knowledge of the disorders being diagnosed. They will therefore be “almost entirely dependent on their counselors for information about disabilities and may have difficulty imagining the various possibilities and options” [Royal Commission on New Reproductive Technologies, 1993]. There is also a danger that health professionals themselves may lack sufficient knowledge, especially as services are increasingly performed by primary care physicians or obstetricians, rather than geneticists [Ormond et al., 2003; Marteau and Dormandy, 2001; Abramsky et al., 2001; Holtzman and Watson, 1997]. Furthermore, the rapid increase in technology means that an increasing amount of information is available for disclosure. This raises concerns about the detrimental impact of information overload on a patient’s ability to understand the process and to make an informed choice.

A second barrier to informed consent is the significant time constraints imposed by the

PND context. Not only does the woman or couple face time pressures while trying to process information, but the health care provider may also lack the time to present information adequately [Marteau and Dormandy, 2001; Holtzman and Watson, 1997]. These time pressures become increasingly important when the woman wishes to consider abortion, because the longer an abortion is delayed, the more risks and trauma it involves for the woman [Royal Commission on New Reproductive Technologies, 1993].

### Informed Consent for PND: What to Include in the Discussion

It is essential to remember that prenatal tests are only *offered* to women and couples. They are not mandatory, and the choice to undergo testing remains with the woman [Kohut et al., 2002]. It is therefore essential that as much information as possible be discussed *prior* to testing, not merely after a diagnosis has been made. It may also be prudent to discuss the genetic counseling service itself, including issues such as confidentiality and nonpaternity, given that many patients will be unfamiliar with the service [Jacobson et al., 2001]. Several elements have been repeatedly identified as central to a well-informed decision with respect to PND.

#### I. Information about the Procedures

Under Canadian law, courts tend to impose a very high standard of disclosure for services that affect reproductive capacity because they are generally not life or death situations [Sneiderman et al., 2003]. Ethical guidelines similarly demand a very comprehensive disclosure of information. The necessary information includes: available methods of

prenatal diagnosis; the difference between screening (e.g. ultrasound) and diagnostic testing (e.g. amniocentesis); risks and benefits of various techniques, including the risk of pregnancy loss; the timing of the procedures; details of the conditions for which procedures screen or test; frequency of abnormal results; accuracy of results, including the implications of false positives and false negatives in screening; the frequency of need for repeat testing; the possibility that abnormalities may go undetected; and, for relatively new and untested procedures, a detailed explanation of their uncertainty and experimental nature [SOGC, 1998; Chodirker et al., 2001b]. Overall, there has been a trend towards providing an increasing amount of detailed information, and it has been suggested that this is unlikely to reverse in the future [Mavroforou et al., 2003].

The method of presentation of such complex information can have a significant impact on the patient's reaction and interpretation. Describing every possible risk and consequence in alarming detail can distort the patient's comprehension and impair the informed consent process.

It is possible to be frank, honest, and direct without being either grim or frightening. It is possible to discuss risk and still leave hope. Pessimism may decrease the effectiveness of the communication and lead to increased patient anxiety [Kelly, 1992].

It is unclear whether physicians are under a duty to provide information about procedures that appear unavailable to the woman. It has been suggested that physicians may now be obliged to inform patients of the inadequacies of the health-care system and of the

availability of better diagnostic or treatment options elsewhere in Canada or in the United States [Osborne, 2003]. Certainly, if the procedure is unavailable due to lack of hospital resources, then the patient should be informed if it is accessible elsewhere. But if the procedure is unavailable because the cost is too prohibitive or because it is only available in distant countries, “it is not obvious that physicians are legally bound to tantalize” the woman with options beyond her reach [Dickens, 2002]. There is also authority from the U.S. for the argument that physicians are not obligated to inform of procedures available in other jurisdictions when such procedures are illegal in the state in question [*Spencer v. ??*]. There may also be no duty to disclose treatment that is not generally recommended [Munro v. Regent of the University of California, 1989] The physician may want to cater the information to options that are within patients’ reasonable access. On the other hand, one may want to risk erring on the side of over-informing rather than under-informing because the couple may be willing to make sacrifices the doctor does not consider reasonable or possible [Cook et al., 2001], and doctors may face liability for failing to inform of alternatives that the patient would have considered.

The distinction between prenatal screening and diagnostic testing is a particularly important piece of information that must not be neglected. The increasing “routinization” of ultrasound screening means that it is occurring with greater regularity than diagnostic testing and is more likely to be performed by a general practitioner or obstetrician than a genetic specialist [Suter, 2002]. Unfortunately, studies have documented the poor levels of information conveyed by non-genetic specialists at early stages of pregnancy [Bernhardt et al., 1998; Marteau et al., 1992]. It is essential that women be given the



opportunity to opt out, without detriment to their further prenatal care [Mavroforou et al., 2003]. Particular attention must be paid to the woman's potential lack of information about these screening procedures. First, many women are not aware of the full extent of genetic information that can be revealed, and some are not even aware that ultrasound can detect fetal abnormalities at all [Kohut et al., 2002]. Women must be properly informed of this possibility because some may elect not to have the procedure for this reason, or they could at least be better prepared to hear the results. If the woman is not properly informed, and a genetic problem is detected, there is a question about whether this should be disclosed. This is a difficult ethical issue, and the best recommendation is that patients be warned of such possibilities prior to testing so that they can express their wishes with respect to disclosure [Ontario Law Reform Commission, 1996]. Second, patients must be fully informed about the limitations of the screening procedure (the fact that not all anomalies can be detected), and about the risks and benefits involved [Driscoll, 2004; SOGC, 1999; Kohut et al., 2002]. Third, while diagnostic testing can detect the presence of a disability, screening only identifies at-risk individuals. Patients should be informed that a screening test is not a replacement for a diagnostic test, and that detection of a fetal abnormality through screening may necessitate a more invasive diagnostic test [Driscoll, 2004]. Otherwise, women who would not choose diagnostic testing will end up facing a very difficult decision after receiving an abnormal screening result. If they reject the further diagnostic test, "the rest of the pregnancy will be fraught with tension, anxiety and worry." This unnecessary anxiety could have been avoided by refusing the initial screening test [Suter, 2002]. Fourth, it is important to clarify the distinction between probabilities revealed through screening (including the risks of false

positives) and confirmatory diagnoses. To enhance understanding of the numbers, evidence shows that quantifying uncertainty in frequencies rather than probabilities, and presenting risks in words rather than numbers is most helpful [Marteau and Dormandy, 2001]. Finally, even women who fully understand this information may face increased anxiety upon a false positive screening result, feeling that “something must be wrong,” and it is recommended that this risk of anxiety be discussed [Suter, 2002].

## II. Information about the Genetic Conditions

It is essential to provide women information not only about the available procedures, but also about the conditions that can be detected through testing. First, many patients may not have sufficient understanding of basic genetics, and it is important to clear up misconceptions (e.g. heredity, genetic terminology) [Lanie et al., 2004]. Second, it is important to be as comprehensive as possible about the types of conditions that may be detected to enable an informed choice about whether they want the information disclosed. It must be remembered that although women are entitled to receive information, this does not mean they are under a duty to do so; in other words, they have the “right *not* to know.” This is significant in the genetic context because a genetic diagnosis may reveal unwelcome information [Dickens, 2002]. Providing fetal information to a parent who did not request it may cause a number of harms, including forcing the couple to make difficult choices about continuing the pregnancy, changing the parents’ attitude towards the pregnancy and the “abnormal” child, and affecting others in the family by revealing genetic information about their own health [Boyle et al., 2003]. As one woman recently stated, “Knowing is not always best” [Kohut et al., 2002]. It is therefore essential to

inform patients about the various anomalies that a procedure may detect, to give them an opportunity to a) accept or reject the procedure as a whole, and b) choose which information they would like disclosed to them should anything be detected [Boyle et al., 2003; FIGO, 1991]. Physicians should also be cautious about attributing a woman's reluctance to discuss fetal anomalies to an informed choice not to know. The reluctance may simply stem from a fear of the unknown, and physicians should attempt to allay this fear [Kohut et al., 2002]. Unfortunately, the breadth of information available means that some information will be conveyed more often than others. For example, it has been found that women are informed less often about the possibility of detecting a SCA, than the possibility of autosomal trisomies, leading to fear and confusion when SCAs are detected [Petrucci et al., 1998].

Providing information about conditions is a difficult balancing act. On the one hand, respect for a patient's autonomy demands that counselors convey as much information as is practical to convey, but on the other hand too much information might be confusing or overwhelming for the patient. Because genetic information is framed in language of probabilities and statistics, practitioners must be especially careful not to be misleading and to communicate at a level appropriate to the patient.

### III. Range of Options Available upon Finding an Abnormal Result

Couples must be informed of the full range of management options available when confronted with an abnormal result. These options include termination of the affected

pregnancy or continuing with the pregnancy while preparing for the birth of the child. [Chodirker et al., 2001b; Strong, 2003]. Studies have shown that information on the first option is given more often than information on the second option, but even that is not always provided [Marteau and Dormandy, 2001].

With respect to abortion, the woman must be informed of the various abortion procedures and their availability, including the time frame during which abortion is legally available, its availability in the local area, and the nearest facilities where it can be obtained [Strong, 2003; Chodirker et al., 2001b]. If a physician has a conscientious objection to abortion, or lacks the necessary resources to perform abortions, this does not justify failing to inform the patient of the option or failing to offer a referral [Strong, 2003; Dickens, 2002; CMA Code of Ethics, AMA Code of Ethics s.E-2.12; FIGO, 1995].

Although abortion is to be discussed as an option, women must be advised beforehand that an agreement to terminate a pregnancy if an abnormality is found is *not* a precondition for testing [Chodirker et al., 2001b; SOGC, 1998; FIGO, 1991]. In other words, “the link between prenatal diagnosis of fetal anomaly and termination of pregnancy is *potential* only” [Bennett, 2001]. This is important because studies show that a decision to undergo prenatal testing is not always linked to the intention to terminate pregnancy [Lafayette et al., 1999]. On the other hand, prenatal testing may not be valuable for all women would who not consider an abortion. Counselors should help women avoid the “potentially ‘toxic’ knowledge” of a fetal anomaly by encouraging them to consider whether abortion is a viable option and what prenatal diagnosis would

mean to them [Suter, 2002].

#### IV: Social Aspects of Disability and Raising a Disabled Child: Stigma, Prejudice, Access to Resources

It is also important to discuss social issues related to disability, including the long-term implications of living with a disease, the prognosis for the disease, and the expected quality of the child's life [Brookes, 2001; Royal Commission on New Reproductive Technologies, 1993]. It can also be helpful to describe what the child might look like [Petrucci et al., 1998]. Unfortunately, health care providers are educated too rarely on the implications of life with disability, and may have difficulty conveying such information [Parens and Asch, 2003]. Different healthcare professionals may also assess quality of life in different ways. There is evidence that most health care trainees focus on medical aspects of quality of life, instead of personal and social variables. Genetic counselling students are more likely than other medical students to prioritize the personal and social factors in discussions of quality of life [Ormond et al., 2003]. The perceived seriousness of disability and the negative social predictions can also vary depending on the person [Marteau and Dormandy, 2001]. This may stem from lack of knowledge and experience in dealing with disability. One mother noted that, "without her experience of caring for a child with a genetic condition, the negative picture presented by her medical practitioners in a later pregnancy would have undermined her confidence in her ability to care" [Brookes, 2001]. It is therefore important for health care professionals to receive education and training in order to present an accurate picture of disability. This will necessarily involve sensitivity training and consultation with a variety of groups to

achieve a balanced picture of life with disability [Ormond et al., 2003; Marteau and Dormandy, 2001; Parens and Asch, 2003].

Parens and Asch note that there is some uncertainty about the ideal moment to engage in this discussion. Prior to the screening test, it may be impractical to convey that much information; after a positive screen, parents may be in shock and may feel threatened by such a discussion. Parents might be most willing to gather information after a positive diagnosis, while on the other hand it may be too stressful to process new information at that point. One clear guideline is that if a woman or couple requests information, they should receive it [Parens and Asch, 2003].

#### V. Social Pressures to Make the “Right” Decision

Informed choice is threatened by various social pressures that influence a woman or couple’s decision in the context of prenatal testing. The very existence of PND makes it possible to view the birth of a disabled child as a woman or couple’s “choice,” thereby making the woman or couple accountable in the event of discriminatory reactions towards the child and themselves [Brookes, 2001]. Social pressures stem not only from direct stigmatization, but also from the lack of economic or social resources for those living with disabilities. It has been recommended that anyone undergoing genetic counseling should be fully informed about the social pressures they may experience, “so that they have an opportunity to consider how such pressures might affect them” [Royal Commission on New Reproductive Technologies, 1993].

At the same time, however, such pressures can originate in the medical community itself. In contrast with the principle of non-directiveness, studies have found that mothers experience a need to “push against” both overt and covert pressure to undergo invasive PND [Brookes, 2001], and that some are left with the impression that they were not actually given a choice [Kohut et al., 2002]. These pressures are of particular danger when procedures such as ultrasound are offered in a routine “matter of fact” manner. Thus, women ought to be explicitly informed that they have the right to refuse or accept any aspect of the offer [Kohut et al., 2002].

#### Emotional and Psychosocial Counseling as Part of the Informed Consent Process

Another unique aspect of PND relative to other medical services is that it is not concerned with treatment and cure, but rather with probabilities and predictions. Given the highly unsatisfactory nature of such information, the anxiety it may cause, and the potential for grave errors, an important aspect of prenatal care in the context of PND is the provision of emotional support. This is based on the ethical principle of beneficence, which requires physicians to reasonably attempt to prevent and remove health-related harms to their patients. When the woman requires help, including emotional support, and the health professional is in a position to offer it, the professional is under a duty to do so [Strong, 2003]. It has been suggested that this type of emotional and psychological counseling imposes upon genetic testing a distinct model of care, based upon a therapeutic “human vision” rather than a mere “medical vision” [Sharpe, 1997]. Not only can emotional distress threaten the patient’s well-being, but it can significantly impair his

or her ability to comprehend the information. There may be a duty to assess and respond to the patient's emotional and psychological needs to ensure that the communication process is not impaired [Sharpe, 1994b; 1996].

#### Pre- and Post-Test Counselling

Before the patient even chooses to undergo testing, counsellors may have a duty to inform her of the risk of anxiety subsequent to the test [Suter, 2002]. After testing, it is the responsibility of the health care provider who initiates referral to ensure that, when test results are received, they are reviewed and reported to the woman as quickly as possible [SOGC, 1998]. Because the initial shock of receiving a positive diagnosis may cause "psychological devastation," physicians must respond to the psychological and emotional concerns of the parents, inform them of the potential for mistaken diagnosis, and inform them of the full range of options available [SOGC, 1999]. Although many physicians receive little or no training in giving "bad news" and feel uncomfortable doing so, there is a growing body of literature that can be consulted regarding effective techniques for delivering "bad news" in these situations [Strong, 2003].

#### Follow-Up Counseling

Following the diagnosis of a severe disorder, or the decision to terminate a pregnancy, the woman or couple may require additional long-term supportive counseling. Follow up calls and letters can be a valuable source of support [Petrucci et al., 1998]. Women may feel a strong sense of guilt following an abortion [Royal Commission on New Reproductive Technologies, 1993], and it also marks the beginning of a grieving process



for many women [Bennett, 2001; Dallaire and Lortie, 1993]. It is recommended that “special attention” be given to the counseling needs of women and couples in cases involving termination after the discovery of a serious fetal disorder [Royal Commission on New Reproductive Technologies, 1993]. For example, a session should be scheduled to counsel the patient concerning future pregnancies and to assess her emotional needs. Moreover, the woman and her partner may grieve in different ways, creating a sense of isolation from each other, and referral for couple’s counseling may be appropriate [Strong, 2003]. Finally, access or referral to self-help groups or associations of people who have had a child with the same disorder may help reduce the feeling of isolation and provide support and helpful advice [Royal Commission, 1993; Dallaire and Lortie, 1993; Petrucelli et al., 1998].

### Structuring the Genetic Counselling Session

The setting of the genetic counseling session plays an important role in the informed consent process. First, women and couples must be given sufficient time to consider the relative advantages and disadvantages of various procedures [Mavroforou et al., 2003]. Canadian guidelines recommend counseling appointments be scheduled at least one day prior to invasive procedures [Chodirker et al., 2001b]. Second, face-to-face encounters may be the best way to communicate information because they allow the counselor to see and respond to the patient’s emotional reactions, to gauge her level of understanding based on body language, and to provide helpful visual aids. The physical setting must be conducive to a discussion; namely, private and relatively quiet [Strong, 2003]. However,

some studies indicate no large difference between telephone and in person counseling with respect to patient understanding and anxiety. Indeed, visual privacy of the telephone may be beneficial for open communication [Sangha et al., 2003]. In any event, telephone discussions can be an important adjunct for women who are geographically distant [Chodirker et al., 2001b; Sangha et al., 2003]. Third, **as discussed in Chapter \_\_\_\_**, written brochures or videos should be made available [Holtzman and Watson, 1997], but should not be relied on as the sole means of transmitting information. Concerns have been raised about the quality of these brochures, and whether they may be too complex, technical, or difficult to read [Royal Commission on New Reproductive Technologies, 1993; Marteau and Dormandy, 2001]. Canadian guidelines note that if provided, they must be regularly reviewed and updated [SOGC, 1998]. Written summaries of the counseling sessions can also be helpful for the patient [Royal Commission on New Reproductive Technologies, 1993], although it has been found that counselors are unlikely to use informed consent documents [Jacobson et al., 2001].

#### Informed Consent and Pre-implantation Genetic Diagnosis (PGD)

While genetic counseling for PND has been studied in detail, little has been written about the recommended course of action for couples considering PGD, presumably because it is relatively new and uncommon procedure [Raeburn, 2001]. In this case, the consultation requires discussion of two topics: in vitro fertilization (IVF) and PGD. There must be a realistic assessment of pregnancy success, side effects and potential risks. It is also important to explain that in PGD only a limited number of diseases are tested for,

meaning that the baby is not guaranteed to be “perfect” [Overton et al., 2001]. It has also been suggested that couples be informed of the differing misdiagnosis rates depending on the implantation method, and that they should decide the degree of misdiagnosis that is acceptable. The discussion must also cover the welfare of future children and the needs of the family [Raeburn, 2001]. Couples should also receive psychological counseling, involving exploration of their reasons for choosing PGD and their overall feelings. Couples who choose PGD may already have suffered stresses, such as an affected child, repeated miscarriage, previous prenatal diagnosis, or termination [Overton et al., 2001]. Finally, the professionals involved at the referring genetic centre and the clinical team from the IVF centre should keep in close contact with one another [Raeburn, 2001].

#### Final Thoughts: Can Genetic Counselling be Non-Directive?

Although non-directiveness in genetic counselling has come to be seen as a “universal norm,” doubts have been raised about how achievable, how desirable, and how accurate the principle is in practice [Bower et al., 2002; Anderson, 1999]. A recent survey of genetic counsellors revealed some of their main concerns. First, the nondirective approach can be confusing for women because they are used to the typical settings in which the health practitioner offers an opinion. Second, some patients are actually seeking direction from practitioners when making important health decisions, to relieve the responsibility from themselves. Third, some women might value the views of others as an information-gathering tool, to test different opinions. Fourth, attempting to give scientific facts and medical knowledge in a value-neutral way is incompatible with the

notion of a caring relationship, and can undermine open communication. The survey highlighted that counsellors did not adopt a uniform strategy for discussions, and much depended on how they framed the information and what information they opted to disclose [Williams et al., 2002].

Aside from the practitioner's behaviour, the general move towards routine prenatal testing has an impact on the directiveness of counselling. Both the strong value our society places on knowledge, information, and technology, and the increasingly routinized way in which prenatal testing is offered places informed "choice" at risk. In particular, these factors deemphasize the emotional and psychological ramifications of undergoing such testing [Suter, 2002]. Furthermore, genetic counseling and genetic procedures (especially prenatal screening) are now performed by a variety of individuals other than genetic counselors, such as obstetricians and midwives. These non-genetic health professionals are less "steeped" in the culture of nondirectiveness and may not understand that genetic testing imposes a unique standard of care for communication [Suter, 2002; Williams et al., 2002; Sharpe, 1994A; 1996]. Evidence does show that the counselling provided by geneticists tends to be more positive and less directive than that provided by other health professionals [Marteau and Dormandy, 2001].

### **Confidentiality**

Health care professionals have a legal and ethical duty to protect the confidentiality of personal health information they receive from or about their patients [AMA Code of

Medical Ethics; CMA Code of Ethics; Feinberg, Peters & Willson, 1984; Picard & Robertson, 1996; Marshall & Von Tigerstrom, 2003]. What does this entail for health care professionals who provide prenatal testing and preimplantation genetic diagnosis services? As a general rule, the wishes of patients to keep their information confidential should be respected. Couples or women who choose to undergo prenatal testing or preimplantation diagnosis may wish to keep this a secret. They may feel that their health risks and the risks to their potential children concern only themselves. Alternatively, they may worry that their choice to undergo such tests, to consider a termination of pregnancy or the elimination of embryos, or to voluntarily have a child who will be disabled could be considered morally questionable by others. Parents may also want to keep the results of these tests, and the information it reveals about them, to themselves for a number of reasons, for example for fear of stigmatization and discrimination or because of feelings of guilt. One parent may also oppose the disclosure of prenatal test results to the other parent because he or she believes it is in the other's best interest or because he or she fears abandonment. If a child is born with a genetic disorder, parents may prefer to give an alternative explanation for the child's disability.

Nonetheless, this information could be relevant for a number of third parties, including relatives of the couple and, in particular, those who are of reproductive age or younger (siblings, cousins, nephews and nieces, etc.). Because genetic information is individual and familial, a test result indicating that a foetus carries a genetic mutation or chromosomal aberration could indicate that blood relatives are at increased risk of having a genetic disorder themselves or of having a child with a disorder. The refusal of a patient

to share that information creates a dilemma for the healthcare professional; his or her duty to protect the patient's confidentiality collides with his or her duty to prevent harm [Lucassen & Parker, 2004; Offit et al., 2004]. Various organisations have developed policies and guidelines to assist healthcare professionals to resolve these issues. The American Society of Human Genetics adopted the position that genetic information "should be protected by the legal and ethical principle of confidentiality" but that disclosure should be permitted in exceptional circumstances, namely when (1) attempts have been made to encourage a patient to disclose the information but have failed; (2) it is highly likely that the non-disclosure of information will lead to a serious and foreseeable harm; (3) the at-risk relatives are identifiable; (4) the harm is preventable or treatable or medically acceptable standards indicate that early monitoring will reduce the genetic risk; and (5) the harm that may result from the failure to disclose outweighs the harm that may result from disclosure [American Society of Human Genetics, 1998]. Though these criteria are broad, they do provide much needed guidance to healthcare professionals who are confronted with these issues. However, healthcare professionals should be aware that though a disclosure may be considered ethically justifiable, it may not be legally justifiable. In some settings, the non-consensual disclosure of a patient's personal information may be authorised only if it is required by law.

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