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Medical Geneticists Confront Ethical Dilemmas: Cross-cultural Comparisons among 18 Nations

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Summary

To provide a basis for international discussion of ethical problems, we studied responses of medical geneticists in 18 countries to questionnaires about 14 clinical cases and five screening situations. Of 1,053 asked to participate, 677 (64%) responded. There was ≥75% consensus on five cases involving (1) disclosure of (1) conflicting diagnostic findings, (2) disclosure of ambiguous results, (3) disclosure of controversial interpretations, (4) protection of mother's confidentiality in cases of false paternity, and (5) nondirective counseling about 45,X and XYY syndrome. A majority (51%–60%) would disclose the diagnosis to relatives at risk for Huntington disease or hemophilia A, against the patient's wishes; would disclose which parent carries a translocation causing Down syndrome; and would disclose XY genotype in a female. As reproductive options for patients with disorders not diagnosable prenatally, 84% would discuss artificial insemination by a donor, 66% would discuss in vitro fertilization with donor egg, and 46% would discuss surrogate motherhood. In all, 85% would perform prenatal diagnosis for (or would refer) parents who refuse abortion, 75% for maternal anxiety, and 42% for selection of fetal sex. Screening questions showed that 72% believed that workplace screening should be voluntary and that results should be confidential.

Introduction

Medical genetics promises to be an area of intense ethical conflict in the next 10 years, as mapping the human genome makes possible a new array of screening tests for inherited susceptibilities. Now is the time for geneticists to come to terms with the ethical problems posed by the new discoveries. Their views will clearly carry weight in the debates to come. Although individual geneticists have set forth their views (see Pfeiffer et al. 1982; Schroeder-Kurth 1982; Berg 1983; Crawfurd 1983; Czeizel 1987), there has been no systematic study of working approaches to clinical problems.

Working approaches are bridges between the general principles underlying medical ethics (e.g., respect for persons, avoidance of harm, and justice) and the concrete situations of individual cases. Working approaches bring abstract principles down to earth and attempt to resolve conflicts between them. For example, clinicians frequently face conflicts between preserving patient confidentiality (respect for persons) and preventing harm to relatives at risk for serious genetic disorders.

Fletcher et al. (1985) have proposed that medical geneticists around the world would benefit from collective reflection on their preferred approaches to difficult moral choices. They suggested that an evolving international consensus, based on respect for patient autonomy and avoidance of harm, would provide the following working approaches: (1) full disclosure of test results, (2) disclosure of psychologically sensitive information in the context of comprehensive counseling and patient education, (3) protection of reproductive options through nondirective counseling, (4) protection of patients' privacy, especially from institutional third parties, (5) use of prenatal diagnosis only to prevent harm from genetic disorders, and (6) volun-

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tary, not mandatory, screening (except for newborns when early treatment is available). In order to assess the strength of consensus about these working approaches and to provide a baseline for future discussion, we undertook a survey of geneticists' views.

Methods

On the basis of fieldwork at 25 genetics centers in 12 countries, we developed 14 clinical case vignettes illustrating the problems that geneticists found most difficult to resolve. These are described in table 1, as asked in the questionnaire, together with the reply choices. The cases fell into the following categories: full disclosure of test results (cases 1–3), disclosure of psychologically sensitive information (cases 4, 8, and 9), directiveness versus nondirectiveness in counseling about reproductive options (cases 5, 10, and 11), protecting patient privacy versus preventing harm to relatives at genetic risk (cases 6 and 7), and indications for prenatal diagnosis (cases 12–14). We also included a set of five questions about screening and access to results and a set of 12 questions about goals and approaches to counseling, reported elsewhere (Wertz and Fletcher 1988*a*). Respondents were asked *what* they would do, from a list of possible responses, and *why*, in their own words, they had chosen this particular course of action. Questionnaires were administered in English, because most genetic specialists worldwide have received training in English-speaking nations.

For study, we chose nations representing a range of geography and cultures. We included only those with at least 10 practicing clinical geneticists at the doctoral level, including one willing to distribute and collect the anonymous questionnaires. Our contact geneticists tried to include all medical geneticists at the doctoral level but omitted genetic counselors at the master's level because this emerging professional specialty is found mainly in the United States and Canada.

Of the 1,053 geneticists asked to participate, 677 (64%) returned anonymous questionnaires by the close of the study, in February 1987 (table 2). In all, 87% held M.D.s, 16% held Ph.D.s, and 3% held other degrees; 82% were members of their national genetics society, and 77% were board certified or accredited where certification in genetics was possible (Canada, Hungary, the United Kingdom, and the United States). Respondents had a median of 14 years practice in genetics and spent an average of 45 h/wk in genetics; 65% were male, and 82% were married, with a median of 1.5 children. Religious backgrounds were 40% Protestant, 18% Catholic, 17% Jewish, 12% none, and 13% other. As a whole, they were nonpracticing, attending a median of one religious observance a year. In all, 49% characterized themselves as politically liberal, 15% as conservative, and 36% as both equally. In the United States, a comparison between 274 respondents and 208 nonrespondents listed in the 1986 combined *Membership Directory* of the Genetics Society of America, The American Society of Human Genetics, and the American Board of Medical Genetics revealed no statistically significant differences between respondents and nonrespondents in type of degree, gender, geographical area, or subspecialty. In the absence of an accepted scientific criterion for consensus, our working criteria for consensus were those frequently used in legislative processes. We used a "3/4's rule" (3/4 of the respondents in each of 3/4 of countries) to define a "strong consensus" on an international basis.

Results

Cases with Consensus

There was strong international consensus on five (35%) of the 14 clinical cases (table 3); four cases concerned full disclosure of test results, and one case concerned nondirective counseling. Few geneticists reported any ethical conflict in disclosing test results in the first three cases. In case 2, disclosure of ambiguous test results, 66% (60% outside the United States) would also disclose the fact that their colleagues had disagreed about the meaning of these results.

Case 4 (false paternity) presents a dilemma between the duty to tell the truth and the duty to prevent harm to the family and to the already vulnerable child who has the disorder. Overall, 96% of respondents believed that protection of the mother's confidentiality overrode disclosure of true paternity. Of these, 81% (79% outside the United States) said that they would tell the mother in private, without the husband present, and let her decide what to tell him; 13% (16% outside the United States) would tell the couple that they are both genetically responsible, and the remaining 2% would ascribe the child's disorder to a new mutation, a one-in-a-million occurrence. As their reasons for such answers, 58% cited preserving the family unit, 30% cited the mother's right to decide, and 13% cited the mother's right to privacy.

Case 5 involves protecting reproductive options, including termination of pregnancy or carrying to term. There was strong consensus that counseling should be

Table 1

Clinical Cases for Ethical Resolution

Issue	Case Vignette (from questionnaire)	Reply Choices	Ethical Principles[a]
1. Full disclosure of test results: conflicting diagnostic findings (table 3)	Maternal serum alpha-fetoprotein has been elevated in your patient on two occasions, but level II ultrasound discloses no abnormality, despite careful examination of the fetal head, spine, abdomen, and kidneys. The fetal karyotype is normal. Amniotic alpha-fetoprotein is elevated, and acetylcholinesterase is borderline. These results raise the possibility of a small neural tube defect. What do you tell the parents?	1. Tell them that there *may* be a small neural tube defect and advise abortion. 2. Tell them that there *may* be a small neural tube defect and advise them to carry the pregnancy to term. 3. Tell them that there *may* be a small neural tube defect, explain the imperfect state of our scientific knowledge and possible other explanations for the test results, but refuse to give any advice about what they should do.	Autonomy (right to know) vs. nonmaleficence
2. Full disclosure of test results: ambiguous/artifactual results (table 3)	Laboratory analysis of amniotic fluid cells suggests that the fetus may be a trisomy 13 mosaic. There is disagreement among the medical geneticists responsible for the analysis as to whether the laboratory results are artifacts of culture—in other words, false positives. Given the present state of knowledge, there is no way of resolving this disagreement scientifically within the legal time limit for termination of pregnancy, because the results of repeat tests will not be available until after 24 wk gestational age. You were not responsible for the laboratory work in this case and have not taken one side or the other. You are, however, the medical geneticist responsible for dealing directly with the prospective mother. What would you do?	1. Tell her that there is *no* abnormality. 2. Tell her that there *is* an abnormality. 3. Tell her that there *may be* an abnormality. 4. Tell her that your colleagues disagree about the test results and that there *may be* an abnormality. 5. Avoid mentioning the results of this particular test at all.	Autonomy (right to know) vs. nonmaleficence
3. Full disclosure of test results: new/controversial interpretations (table 3)	Repeated maternal serum alpha-fetoprotein tests reveal a value that is *below* the norm. Although some studies have found low maternal serum alpha-fetoprotein values to be associated with Down syndrome, geneticists are not in agreement about how a low value should be interpreted. What do you tell the family?	1. Tell them that the tests indicate a possible Down syndrome fetus and urge them to have prenatal diagnosis. 2. Tell them that the maternal serum alpha-fetoprotein value is low but that research on this topic is so new that we do not know how to interpret the test results. 3. Tell them that geneticists are not in agreement about the interpretation of test results but that some geneticists think there may be a possibility of Down syndrome, explain the relative risks, and then let them decide whether to have prenatal diagnosis. 4. Do not tell them about the test results.	Autonomy (right to know) vs. nonmaleficence

4. Full disclosure of psychologically sensitive information: false paternity (table 3)	You are evaluating a child with an autosomal recessive disorder for which carrier testing is possible and accurate. In the process of testing relatives for genetic counseling, you discover that the mother and half the siblings are carriers, whereas the husband is not. The husband believes that he is the child's biological father.	1. Tell the couple what the laboratory tests reveal about the child's parentage. 2. Tell the couple that they are *both* genetically responsible. 3. Tell the couple that the origin of the child's disorder is not genetic. 4. Tell the couple that you have not been able to discover which of them is genetically responsible. 5. Tell the couple the facts about the child's parentage and try to get the name of the child's biological father so he can be told that he is a carrier. 6. Tell the mother alone, without her husband being present.	Autonomy (husband's right to know; mother's right to privacy), nonmaleficence (preserve family unit and prevent harm to child and mother)
5. Nondirective vs. directive counseling: fetuses with low burden disorders (table 3)	Prenatal diagnosis has revealed an abnormal fetus. The prospective parents come to see you in genetics clinic. In this question you are asked how you would react to two different fetal conditions—Turner syndrome (XO) and XXY—in your professional capacity.	1. Advise them to carry the pregnancy to term. 2. Advise them to have an abortion. 3. Refuse to make any suggestion about what they should do. 4. Provide complete information about the disorder, including prognosis, potential treatment and education, and possible problems the child may present for parents and society, taking care to present both sides of any topic that is controversial, and then let them make their own decision. 5. Give them the information described under reply choice 4 in an optimistic form so that they will favor carrying the pregnancy to term. 6. Give them the information under reply choice 4 but stress costs and burdens so that they will favor abortion without your having suggested it directly. 7. Give them information under reply choice 4 but also describe the emotional difficulties associated with terminating the pregnancy and then tell them to make their own decision.	Autonomy (right to decide) vs. nonmaleficence (prevent harm to fetus or society) or justice (interests of fetus should be weighed equally with those of living persons)

(continued)

Table 1

Clinical Cases for Ethical Resolution

Issue	Case Vignette (from questionnaire)	Reply Choices	Ethical Principles[a]
6. Patient confidentiality vs. duties to relatives at genetic risk: Huntington disease (fig. 1, *left*)	A client recently diagnosed as having Huntington disease refuses to permit disclosure of the diagnosis and relevant genetic information to siblings who may be at risk for Huntington disease. In your professional capacity as a medical geneticist, what would you do?	1. Respect the client's desire for confidentiality. 2. Provide the information to siblings, whether or not they ask for it, only after all reasonable efforts to persuade the client to consent to voluntary disclosure have failed. 3. Provide the information to siblings *only* if they ask for it and after all reasonable efforts to persuade the client to consent to voluntary disclosure have failed. 4. Provide the information to siblings, taking care to insure that only information *directly relevant* to the relatives' risk is provided, regardless of client's desire. 5. Send the information to the client's referring physician and let that physician decide about disclosure.	Autonomy (patient's right to privacy) vs. nonmaleficence (duty to warn third parties of harm)
7. Patient confidientiality vs. duties to relatives at genetic risk: hemophilia A (fig. 1, *right*)	A client with a child recently diagnosed as having hemophilia A refuses to permit disclosure of the diagnosis and relevant genetic information to her relatives who may be at risk for conceiving children with hemophilia A. The information could be useful to these relatives both because the female carrier can usually be detected and because hemophilia A is usually diagnosable prenatally. As a medical geneticist, what would you do about disclosure of this information?	1. Respect the client's desire for confidentiality. 2. Provide the information to relatives whether or not they ask for it, only after all reasonable efforts to persuade the client to consent to voluntary disclosure have failed. 3. Provide the information to relatives, whether or not they ask for it, taking care to insure that only information directly relevant to the relatives' risks is provided, regardless of client's desire. 4. Provide the information to relatives only if they ask for it and only after all reasonable efforts to persuade the client to consent to voluntary disclosure have failed. 5. Send the information to the client's referring physician as part of the medical record and let that physician decide about disclosure.	Autonomy (patient's right to privacy) vs. nonmaleficence (duty to warn third parties of harm)

8. Full disclosure of psychologically sensitive information: parental translocation (fig. 2, *right*)	You identify a parent of a Down syndrome child as having a balanced translocation. What is your approach to disclosure of this information to the parents? Select the *one* that best describes your response.	1. *Before* drawing samples of the parents' blood for karyotyping, tell them that tests may identify one of them as a carrier and ask them whether they want to know who is the carrier. If both say they do not want to know, do not tell them. 2. Ask the parents whether they want to know *everything* about the source of the child's abnormality, including their own carrier status, and, if they say yes, tell them which parent carries the extra chromosomal material; if they say no, do not tell them. 3. Wait for the parents to ask which of them is the carrier and, if they ask, tell them. 4. Provide full disclosure to the couple whether or not they ask for it. 5. Provide full disclosure to the couple whether or not they ask for it *and* also provide full disclosure to all of their relatives who are at risk for having a Down syndrome child. 6. Do not disclose information about carrier status even if asked. 7. Tell them they are *both* carriers. 8. Tell them that *one* is a carrier and then give them the choice of whether or not they wish to be told which one.	Autonomy (right to know) vs. nonmaleficence
9. Full disclosure of psychologically sensitive information: XY female (fig. 2, *left*)	A woman undergoes diagnosis for infertility. Tests reveal that she is chromosomally male (XY). What would you do?	1. Tell her that the reason for her infertility is well understood, explain to her the biology of gender, and then explain that she is chromosomally male. 2. Give her reasons for her infertility, without telling her that she is chromosomally male. 3. Tell her that you do not know the reason for her infertility.	Autonomy (right to know) vs. nonmaleficence
10. Directive vs. nondirective counseling: discussing new reproductive options for carriers of tuberous sclerosis (table 4)	Evaluation of a child produces findings consistent with a diagnosis of tuberous sclerosis. On examining the parents, you find evidence that the father is carrying the tuberous sclerosis gene, even though he is of normal intelligence. After a discussion of the risk of having another child with tuberous sclerosis who might be severely affected, the couple asks you whether recurrence of the disorder can be prevented. What course of action do you take with regard to telling the couple about EACH of the following options: taking their chances, artificial insemination by donor, vasectomy, adoption, contraception	1. Advise them to do this. 2. Advise them *not* to do this. 3. Explain that this is a possibility, without giving any advice 4. Explain that this is a possibility and describe the risks and potential problems involved. 5. Do not discuss this. 6. Discuss this only if the clients ask you about it.	Autonomy (right to know or right to decide) vs. nonmaleficence

(*continued*)

Table 1

Clinical Cases for Ethical Resolution

Issue	Case Vignette (from questionnaire)	Reply Choices	Ethical Principles[a]
11. Question 10 repeated, except that the mother carries the gene	Options are taking their chances, contraception, tubal ligation, surrogate motherhood, adoption, donor egg and in vitro fertilization		
12. Indications for prenatal diagnosis: parents would refuse abortion (table 5)	A woman age 42 years requests prenatal diagnosis for Down syndrome. She and her husband are already the parents of a Down syndrome child. She tells you that they are opposed to abortion and that she will carry the fetus to term even if it is diagnosed as having Down syndrome. They would like to have prenatal diagnosis, however, in order to give themselves additional time to prepare for the birth of another affected child. What would you do?	1. Grant their request for prenatal diagnosis. 2. Refuse their request for prenatal diagnosis. 3. Try to dissuade them from having prenatal diagnosis and, if they still insist on having it, refuse their request. 4. Refer them to another medical geneticist or genetics unit offering this service.	Autonomy or beneficence vs. justice (use of expensive or scarce medical resources) or nonmaleficence (prevent harm to fetus)
13. Indications for prenatal diagnosis: maternal anxiety (table 5)	A woman age 25 years with no family history of genetic disorders and no personal history of exposure to toxic substances requests prenatal diagnosis. There are no genetic or medical indications for its use in this case. Nevertheless, she appears very anxious about the normalcy of the fetus and persists in her demands for prenatal diagnosis even after being informed that in her case the potential medical risks for the fetus, in terms of miscarriage, may outweigh the likelihood of diagnosing an abnormality. Assume that your clinic has no regulations that would prevent you doing prenatal diagnosis for her. What would you do, as a professional?	1. Perform prenatal diagnosis. 2. Refuse to perform prenatal diagnosis. 3. Try to dissuade her from having prenatal diagnosis and, if she still insists on it, refuse her request. 4. Refer her to another medical geneticist or genetics unit offering the service.	Autonomy (right to the service) or beneficence vs. nonmaleficence (prevent harm to fetus) or justice (use of scarce resources)
14. Indications for prenatal diagnosis: sex selection, in absence of X-linked disease (table 5)	A couple requests prenatal diagnosis for purposes of selecting the sex of the child. They already have four girls and are desperate for a boy. They say that if the fetus is a girl, they will abort it and will keep trying until they conceive a boy. They also tell you that if you refuse to do prenatal diagnosis for sex selection, they will abort the fetus rather than risk having another girl. The clinic for which you work has *no* regulations prohibiting use of prenatal diagnosis for sex selection. What would you do?	1. Grant their request for prenatal diagnosis. 2. Refuse their request for prenatal diagnosis. 3. Try to dissuade them from having prenatal diagnosis and, if they still insist on having it, refuse their request. 4. Refer them to another medical geneticist or genetics unit offering the service.	Autonomy (right to decide) vs. nonmaleficence (prevent harm to fetus, society, or women) or justice (use of scarce medical resources)

[a] Definitions of ethical principles are as follows: (1) *autonomy* (respect for the person) is the duty to respect the self-determination and choices of autonomous persons, as well as to protect persons with diminished autonomy, e.g., young children, mentally retarded persons, and those with other mental impairments; (2) *nonmaleficence* is the obligation to minimize harm to persons and, wherever possible, to remove the causes of harm altogether; (3) *beneficence* is the obligation to secure the well-being of persons by acting positively on their behalf and, moreover, to maximize the benefits that can be attained; (4) *justice* is the obligation to distribute the benefits and burdens fairly, to treat equals equally, and to give reasons for differential treatment that are based upon widely accepted criteria for just ways to distribute benefits and burdens.

Table 2

Participating Countries

Country	No. of Geneticists Asked to Participate	No. (%) of Respondents
Australia	14	12 (86)
Brazil	51	32 (63)
Canada	73	47 (64)
Denmark	28	15 (54)
Federal Republic of Germany	55	47 (85)
France	35	17 (49)
German Democratic Republic	25	21 (84)
Greece	11	7 (64)
Hungary	18	15 (83)
India	40	27 (68)
Israel	17	15 (88)[a]
Italy	26	11 (42)[b]
Japan	74	51 (69)
Norway	10[b]	6 (60)
Sweden	26	21 (81)
Switzerland	10[b]	5[b] (50)
United Kingdom	50	33 (66)
United States	490[a]	295[a] (60)
Total	1,053	677 (64)
Total, excluding United States	563	382 (68)

[a] Highest in column.
[b] Lowest in column.

nondirective. The exceptions were the German Democratic Republic, Hungary, and France—where 43%, 60%, and 65%, respectively, would advise carrying an XYY fetus to term or would give optimistically slanted information—and Hungary and India—where 40% and 46%, respectively, would advise aborting a 45,X fetus or would give pessimistically slanted information. In all, 14% would give optimistic information or advice about XYY, and 7% about 45,X; 20%, however, considered a child with either disorder to be within the range of normal.

Cases without Consensus

There was no strong international consensus in the remaining nine cases (figs. 1 and 2 and tables 4 and

Table 3

Cases with Strongest Consensus (≥75% of respondents in ≥75% of countries)

Case No. and Resolution of Case	% of Total Respondents Agreeing	95% Confidence Interval	% of non-U.S. Respondents Agreeing	Minimum % Agreeing in Any One Country
1. Disclose conflicting diagnostic findings	98	96–100	98	89
2. Disclose ambiguous/artifactual results	97	95–99	97	80
3. Disclose new/controversial interpretations	94	91–97	91	76
4. Protect mother's confidentiality on discovering false paternity	96	94–98	97	90
5. Counsel nondirectively about fetuses with low-burden disorders:				
XO (Turner syndrome)	88	84–92	84	40
XYY	84	80–88	84	43

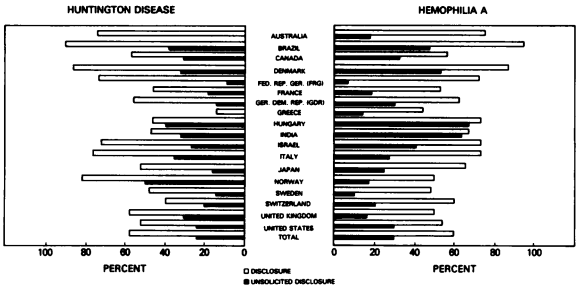

Figure 1 Patient confidentiality vs. duties to third parties: % of geneticists (n = 677) disclosing diagnosis to relatives at risk, against patient's wishes. White bars denote disclosure only at relatives' request; black bars denote disclosure even if it is not requested.

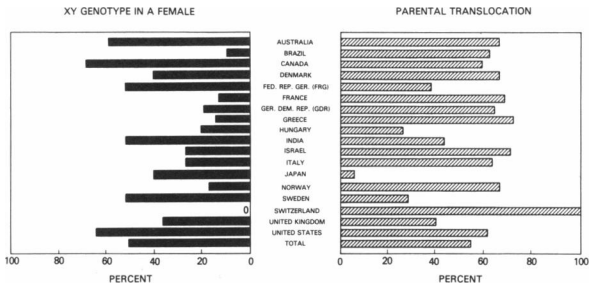

Figure 2 Full disclosure of psychologically sensitive information: % of geneticists (n = 677) who would disclose.

5). These cases involved conflicts between ethical principles and/or between the interests of different parties.

Two cases reflected conflict between the geneticist's duty to preserve patient confidentiality and the duty to warn third parties (relatives at risk for genetic disorders) about potential harm (fig. 1). In each case, the person with a mutant gene (Huntington disease or hemophilia A) has refused to permit disclosure of the diagnosis to relatives at high risk for the same disorder. There were no significant differences in responses to the two cases: 58% (63% outside the United States) in the case of Huntington disease and 60% (66% outside the United States) in the hemophilia A case believed that duties to relatives at risk should override the patient's desire for confidentiality (fig. 1, white bars). These included 24% and 29%, respectively, who would

Table 4

New Reproductive Options (cases 10 and 11)

Country	% of Geneticists (n = 677) Spontaneously Presenting the Option of		
	AID	IVF of Donor Egg	Surrogate Mother
Australia	100[a]	92[a]	46
Brazil	50	59	34
Canada	98[a]	59	37
Denmark	80[a]	80[a]	47
Federal Republic of Germany	72	33	16[b]
France	81[a]	44	0[b]
German Democratic Republic	55	25[b]	5[b]
Greece	67	80[a]	25[b]
Hungary	47	46	31
India	54	43	48
Israel	100[a]	80[a]	39
Italy	80[a]	64	27
Japan	26	21[b]	10[b]
Norway	83[a]	33	33
Sweden	90[a]	55	30
Switzerland	100[a]	80[a]	20[b]
United Kingdom	94[a]	63	27
Overall, excluding United States	72	52	28
95% Confidence limits	68–78	46–58	23–33
United States	96[a]	83[a]	67
Overall, including United States	83[a]	66	46
95% Confidence limits	79–87	46–58	23–33

[a] Strong consensus in favor of presenting.
[b] Strong consensus against presenting.

Table 5

Indications for Prenatal Diagnosis

Country	% of Geneticists (n = 677) Who Would Perform Prenatal Diagnosis (% referrals to other geneticists)		
	Patients Expected Refusal of Abortion	Maternal Anxiety Only	Parental Desire for Sex Selection, in Absence of X-linked Disease
Australia	92[a]	67 (17)	17[b] (8)
Brazil	81[a] (3)	44 (30)	30 (9)
Canada	89[a] (2)	70 (15)	47 (17)
Denmark	87[a]	87[a]	13[b]
Federal Republic of Germany	85[a]	80[a] (7)	4[b] (2)
France	56	57 (19)	13[b] (6)
German Democratic Republic	52	5[b]	10[b]
Greece	57	43	29
Hungary	47	20[b]	60
India	63 (7)	56 (15)	52 (15)
Israel	93[a]	80[a]	33 (20)
Italy	91[a] (18)	82[a] (9)	18[b]
Japan	48 (4)	18[b] (2)	6[b]
Norway	50	50	17[b]
Sweden	91[a]	91[a]	38 (10)
Switzerland	100[a]	100[a]	0[b]
United Kingdom	91[a]	89[a] (9)	24[b] (15)
Overall, excluding United States	76[a] (2)	61 (8)	26 (8)
95% Confidence limits	70–82	55–67	21–31
United States	96[a]	89[a] (11)	62 (29)
Overall, including United States	85[a] (2)	73 (10)	42 (17)
95% Confidence limits	81–89	68–78	37–47

[a] Strong consensus in favor of performing.
[b] Strong consensus against performing.

seek out and tell the relatives even if the latter did not ask for information (fig. 1, black bars); 34% and 31% would tell the relatives only if the latter asked. Overall, 32% would preserve the confidentiality of the Huntington disease patient, and 10% would refer the matter to the patient's family physician for decision; for hemophilia A patients, 27% would preserve confidentiality, and 12% would refer.

Two situations involved disclosure of psychologically sensitive information that might harm the patient (fig. 2). Both cases present a dilemma between respecting patient autonomy (the right to know or not know) and avoiding harm. In case 8, disclosing which parent had a balanced translocation might enable the couple and relatives at risk to use reproductive options that would prevent the birth of another Down syndrome child, but it could also cause guilt in the carrier or threaten the marriage. In case 9, disclosing the XY genotype in an infertile woman could severely damage her self-image, but it could resolve doubts about fertility. Both cases involve conflicts between the geneticist's duty to tell the truth (which may also be phrased in terms of the patient's right to know) and the duty to do no harm. In both cases, patients have asked for the information about etiology but have not asked specific questions about their carrier status or genotype. On average, respondents were equally divided about disclosure, with 54% (46% outside the United States) saying that they would disclose, unasked, which parent was a carrier and with 51% (41% outside the United States) disclosing XY genotype. In case 8, 43% would tell the couple that the information exists and would give them the

choice of knowing or not knowing. In only two countries, Canada and United States, a majority (68% and 64%, respectively) would disclose XY genotype. More geneticists have clinical training in counseling in these countries than elsewhere in the world, and many thought that they could reveal the truth in a manner sufficiently sensitive to prevent harm.

Although both cases involve the same principle (patients' right to know), geneticists in some countries (Brazil, France, the German Democratic Republic, Greece, Italy, Japan, Norway, and Switzerland) responded very differently to the two situations. Their responses suggest that the specifics of the cases exerted more effect on their thinking than did general ethical principles. They replied in terms of how each disorder, as well as gender and family roles, would be understood in their cultural context.

Cases 10 and 11 (table 4) concern directiveness in counseling about options—e.g., artificial insemination by a donor (AID), in vitro fertilization (IVF) of a donated egg, and insemination of a surrogate mother with the husband's sperm—to carriers of a serious genetic disorder not diagnosable prenatally (tuberous sclerosis). The percentages listed represent those who would present each option without being asked and who would discuss it at length without giving directive advice. In the seven countries (Brazil, the Federal Republic of Germany, the German Democratic Republic, Greece, Hungary, India, and Japan) where there was no strong consensus for AID, either the legal status of the child is in doubt or the procedure has been illegal until recently.

The United States was the only country where a majority (67%) would present surrogacy as an option. In six countries (the Federal Republic of Germany, France, the German Democratic Republic, Greece, Japan, and Switzerland), some of which have laws against it, there was a strong consensus against presenting it. Although IVF with a donated egg has had a low success rate, many respondents said that they regarded this as ultimately less likely to cause harm than surrogacy. There was strong international consensus for presenting adoption as an option, but there was no international consensus about presenting taking chances, contraception, or sterilization.

The final three cases concern indications for prenatal diagnosis (table 5). Each of these cases presents a dilemma between respect for patient autonomy, avoidance of harm to a normal fetus, and fairness in distribution of costly or (in some countries) scarce medical resources. The final case (sex selection) also raises societal concerns about the place of women.

In case 12, that of a couple who refuse abortion, 60% of all respondents (53% outside the United States) stated that performance of prenatal diagnosis should not depend on the use that patients intend to make of the information, and 34% stated that such patients might change their minds about termination and thereby justified performing prenatal diagnosis. Refusals were largely based on lack of resources.

In case 13, that of prenatal diagnosis for maternal anxiety, 56% of those in favor cited patient autonomy as their major reason, and 46% cited removal of anxiety. Few would require a psychiatric consultation to confirm the anxiety. Among those opposed to prenatal diagnosis for anxiety, 61% cited possible harm to the fetus as their major reason, and 70% cited waste of resources.

The final case (case 14), that of prenatal diagnosis for sex selection, was the one that respondents said gave them the greatest ethical conflict. Responses from the United States and Canada contrast markedly with findings from surveys conducted in 1972–73 and 1975, when only 1% (Sorenson 1976) and 21% (Fraser and Pressor 1977), respectively, of geneticists were willing to perform prenatal diagnosis for sex selection. In the United States, 68% of those who would either perform prenatal diagnosis or refer would do so out of respect for parental autonomy. To some, sex selection appeared to be an extension of families' right to determine the number, spacing, and quality of their children. Others said they wished to avoid paternalism or to be nondirective (Wertz and Fletcher 1988*b*, 1989*b*).

In their reasoning, 30% of all respondents (same rate in the United States) said that they opposed the abortion of a normal fetus or that the interests of the fetus should be weighed equally with those of living persons. In Hungary, all 15 of those offering prenatal diagnosis would do so in order to prevent the otherwise certain abortion of a normal fetus.

Of the 605 persons who gave reasons for their action, only 4.7% cited the position of women in society, 0.5% cited maintaining a balanced sex ratio, 0.6% cited limiting the population, and 4.9% cited setting a precedent that would harm the moral order. The exception was India, where 61% cited at least one of these social issues. In all, 28% of respondents cited issues related to justice, such as wise use of medical resources or diversion of services away from patients at genetic risk.

Genetic Screening

There was a strong consensus regarding four of the five screening questions (table 6). Respondents agreed

Table 6

Genetic Screening and Access to Results

Case Resolution	% of Total Respondents Agreeing (n = 677)	95% Confidence Interval	% of non-U.S. Respondents Agreeing	Minimum % Agreeing in Any One Country
No access for employer, school, life insurer, or health insurer to results of presymptomatic tests for Huntington disease, without patient's consent	93	90–96	93	60 (India)
No access at all	45		52	
No access for health, life, or workers' compensation insurers to results of screening for susceptibility to work-related disease, without worker's consent	89	84–92	90	65 (India)
No access at all	40		44	
No access for employer to results of screening for susceptibility to work-related disease, without worker's consent	81	77–85	85	41 (India)
No access at all	22		30	
Mass screening for carriers of cystic fibrosis should be voluntary	75	70–80	77	0 (German Democratic Republic)
Screening in the workplace for genetic susceptibility to work-related disease should be voluntary	72	67–77	68	25 (German Democratic Republic)

NOTE.—The full texts of screening questions and reply choices appear in the work of Wertz and Fletcher (1989*d*).

both on protection of privacy from employers and insurers and on voluntary screening for cystic fibrosis, but they fell short of a consensus that screening in the workplace should be voluntary. Although there was more consensus about screening than about clinical cases, all the examples of screening were futural, exceeding scientific capabilities at the time of the survey. We cannot be certain that such a high degree of consensus will remain when these screening tests become actualities. When fully developed, the tests may be accompanied by unforeseen technical, economic, and social problems that will reduce the degree of consensus (Wertz and Fletcher 1989*d*).

Characteristics Related to Choices

For each question or case vignette, geneticists' professional and personal background characteristics were entered into stepwise logistic regressions, with choice of action as the dependent variable. Gender was the major variable related to choice, across nationalities (Wertz and Fletcher 1989*a*). There were significant gender differences in choices of action in 4.17 (30%) of the 14 clinical cases (cases 1, 5, 8, and 13 and one part of case 11) and in moral reasoning in an additional case (case 4). Women were more likely than men to say that they would counsel nondirectively about conflicting diagnostic findings or about fetuses with low-burden disorders; disclose parental translocations, unasked; and perform prenatal diagnosis for an anxious woman. Men were more likely than women to say that they would present surrogacy as a reproductive option. In their moral reasoning, women were more likely to discuss marital conflict in cases of false paternity and to discuss patient autonomy for an anxious woman seeking prenatal diagnosis. Respondents' type of degree, specialty, years in practice, hours per week in practice, number of patients per week, age, marital status, religious background, religiosity, and political preferences were largely unrelated to choices of action. Cultural differences emerged after controlling for professional and personal background, even among countries sharing a common language. For example, the United States, Canada, and the United Kingdom differed at the $P < .05$ level on 5.4 (38%) of the cases (cases 2, 3, 9, 13, and 14 and two parts of case 11), with more geneticists in the United States than in either the United Kingdom or Canada leaning toward disclosure of test results or reproductive options and toward performance of prenatal diagnosis for maternal anxiety or sex selection. These differences may reflect in part the greater litigiousness

of patients in the United States, as well as the consumer revolt against paternalism. A full discussion of cultural differences among all 18 countries appears in the work of Wertz and Fletcher (1989*c*), which devotes a chapter to each country. Cultural views about the status of women, about the seriousness of the disorder in question, about the role of the extended family, about the moral value of the fetus, and about the shame attached to genetic disorders may all affect geneticists' decision making.

Discussion

We propose the following seven standards for working approaches to ethical problems. Some of these (standards 2–4) reflect consenses about cases in our survey. Others (standards 1 and 5–7) do not reflect a consensus among survey respondents but represent the thinking of our contact geneticists and co-authors in the 18 countries (Wertz and Fletcher 1989*c*).

1. *Fairness of Access to Genetic Services*

In the future, the most pressing problem in medical genetics will be increased demand for services. Geneticists in all countries in our study expressed concern that services will not be distributed equally and that the spread of private, for-profit clinics will give privileged access to patients who can pay out-of-pocket. In the United States, genetic services must be seen in the context of larger inequalities in prenatal care. An estimated 25% of all mothers, or 939,000 women annually, receive late or no prenatal care (U.S. Department of Health and Human Services 1989, pp 42 and 46). Women who receive prenatal diagnosis are disproportionately white, well educated, and financially well-off. If this trend continues, being genetically handicapped could become a mark of social class. Even countries with national health insurance experience some social and geographical inequalities in use of genetic services.

2. *Full Disclosure of All Clinically Relevant Information*

Psychologically sensitive information, such as XY genotype in a female, should be disclosed only in the context of full and supportive counseling and patient education.

3. *Respect for Parental Choices, Including Decisions to Abort or to Carry to Term a Fetus with a Malformation or Genetic Disorder*

Abortion rights, including the right to abortion on the basis of fetal defect, are under attack in many countries in this study. Geneticists in all countries considered it essential to protect rights of choice.

4. *Protection of Patients' Privacy from Institutional Third Parties*

Geneticists around the world agreed that third parties, such as health insurance companies and employers, should not have access to personal genetic data unless consent has been obtained. Realizing the potential power of institutions to engineer consent by denying employment or health insurance, a substantial minority of geneticists believed that third parties should have no access at all. This may not be feasible in countries where private health insurance is a major industry, unless legislators can establish that genetic information deserves special protection not currently afforded to other types of medical information. An alternative would be to permit institutional access to genetic information but to have extensive legal protection for individuals that would prevent discrimination. Another alternative would be government underwriting of health insurance for those at genetic risk. Policies governing disclosure of results of HIV testing may set the pattern for disclosure about genetic tests.

5. *Use of Prenatal Diagnosis Only to Give Parents Information about the Health of the Fetus*

Any other use, such as for sex selection (except for X-linked disease), should be avoided. As many survey respondents pointed out, "sex is not a disease." Sex selection discredits the public image of prenatal diagnosis and of medical genetics, lends support to the campaigns of anti-abortionists, and sets a precedent for parental choices on "cosmetic" grounds.

6. *Voluntary, Not Mandatory, Screening—Except for Newborns When Early Treatment Is Available (President's Commission for the Study of Ethical Problems in Medicine and Biomedical and Behavioral Research 1983, pp 47–54)*

In the case of newborns, the primary purpose of screening should be to help the newborn; carrier detection is secondary.

7. *A Need for Further Study and Discussion of the Complex Issue of Patient Confidentiality in Cases Where There is a High Risk of Serious Harm to Relatives at Genetic Risk*

Survey respondents ranked this as the issue that presented the second most difficult ethical conflict (behind sex selection). As pointed out above, there was no consensus. About one-third would not favor breach-

ing confidentiality, about one-fourth would take an active approach toward informing relatives, and about one-third would inform the relatives only if the later asked.

According to the President's Commission for the Study of Ethical Problems in Medicine and Biomedical and Behavioral Research (1983, p. 44), confidentiality may be breached only in exceptional circumstances that meet the following four conditions: "(1) reasonable efforts to elicit voluntary consent to disclosure have failed; (2) there is a high probability both that harm will occur if the information is withheld and that the disclosed information will actually be used to avert harm; (3) the harm that identifiable individuals would suffer would be serious; and (4) appropriate precautions are taken to ensure that only the genetic information needed for diagnosis and/or treatment of the disease in question is disclosed." Berg (1989, p ix) argues that "members of families with genetic diseases have a collective responsibility to their groups that should not easily be ignored." It might be argued that, if patients will not assume this responsibility, then doctors should be legally permitted (but not required) to assume it for them.

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