Genetic Testing

Care, Consent, and Liability

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Dedication

This book is dedicated to my wife, Honey, who in the face of a chronic, debilitating disease, always has displayed an indefatigable attitude and inexhaustible courage, laughter, love, and joy.

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Neil F. Sharpe

Dedication

To my wife Ludy, and our children Veronica, Timothy, Emily, and Stephanie: for their patience and understanding in the face of many hours at work on this book.

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Ron Carter

Methods

To facilitate understanding, test information should be presented in:

- Simple, commonly understood terms [Edwards et al., 1996; Cosmides and Tooby, 1996; European Commission, 2004].
- The language the patient best understands.
- An empathetic approach with a focus on the patient's, and family's, concerns. Patients often wish to discuss the psychological, socioeconomic, and familial concerns [Geller et al., 1998; Marteau and Croyle, 1998; Menahem, 1998; McConkie-Rosell et al., 2001; Esplen et al., 2001; Lion et al., 2002; Speice et al., 2002; Parsons et al., 2003]; however, the health care provider may be more focused on factual test results [Levinson and Roter, 1997; Michie et al., 1997b; Roter et al., 1997; Levinson et al., 2000; Carroll et al., 2000; Phillips et al., 2000; McConkie-Rosell et al., 2001; Bensing et al., 2003].
- A variety of formats including the use of graphs and charts [Kessler and Levine, 1987; Marteau, 1989; Shiloh and Sagi, 1989; Redelmeier et al., 1993; Grimes and Snively, 1999; Sandhus et al., 2001; Edwards et al., 2001, 2002; Woloshin et al., 2002].

Terms, Tone, and Follow-up

Health care professionals may overestimate the burden and negative effects of a genetic disease or disorder [Blaymore et al., 1996; Bogardus et al., 1999; Kirschner et al., 2000; Abramsky et al., 2001]. A patient's perception and understanding can be influenced by the health care professional's tone of voice and facial expressions [Shapiro et al., 1992; Ambady et al., 2002].

Health care professionals need to understand that communication of test information only by telephone and/or by letter [Tluczek et al., 1991; Phelps et al., 2004] and/or electronic means [Cepelewicz et al., 1998; Hodge et al., 1999] without access to face-to-face discussion [AMA Report on Scientific Affairs, 1999] can increase the risk of misunderstanding. Discussion about risk management options [Lerman and Croyle, 1994; Kash, 1995; Lerman et al., 1995, 1997; Burke et al., 1997] with referral for genetic counseling is recommended.

Patients may be uncertain as to the meaning and implications posed by terms such as *rare* and *carrier* and may wish to discuss when is an appropriate time, if at all, to discuss and disclose carrier status with their children [Fanos and Johnson 1995; McConkie-Rosell et al., 1999, 2002; Michie and Marteau, 1999(a); Ciske et al., 2001; Tercyak et al., 2001b; Edwards et al., 2002].

Follow-up visits and a written letter [McConkie-Rosell et al., 1995; Michie et al., 1997c; Hallowell and Murton, 1998; Lobb et al., 2004] summarizing the genetic counseling, the test results, and their significance, helps to facilitate understanding.

CULTURE AND COMMUNICATION IN THE REALM OF FETAL DIAGNOSIS: UNIQUE CONSIDERATIONS FOR LATINO PATIENTS*

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Amniocentesis and other prenatal genetic tests have become well-established features of modern prenatal care but place a considerable decision-making burden on the expectant mothers who are offered them. The genetic and emotional issues involved are complex and the "best" course of action sometimes ambiguous. It is often assumed that ignorance, poverty, and illiteracy, more acceptably termed *cultural* factors, lead women from certain ethnic minority backgrounds to turn down amniocentesis at higher rates than ostensibly less "ignorant" women from European American backgrounds. However, the extent to which *both* amniocentesis acceptance and refusal by ethnic minority women is influenced by miscommunication between providers and patients—and the extent to which such miscommunication stems from *cultural factors*—has rarely been explored [Resta, 1999]. We focus on Latinos because they represent a large and rapidly growing population in the United States (38.8 million as of July 2002, with a growth rate of 9.8 percent), and because they face a higher risk of neural tube defects than most other U.S. groups [Stierman, 1995]. There have been few studies of why Latinos are more likely to decline prenatal testing and why they accept when they do. This section is intended to highlight some of the ways communication and miscommunication may impact decision making.

Most babies born in the United States today are screened for the most commonly occurring birth anomalies, including neural tube defects and certain chromosomal disorders [ACOG, 1996]. In California, where our studies were conducted, women who screen positive are referred to a state-certified prenatal diagnosis center where they are offered a consultation with a certified genetic counselor and additional testing, typically high-resolution ultrasound, and if indicated, an amniocentesis. (Prenatal genetic services tend to be organized differently in different parts of the United States [see, e.g., Rapp 1999; Hunt et al., 2004].) Prenatal genetic consultations in California typically follow a fairly standard protocol that includes obtaining the woman's reproductive and family medical history, describing her options for additional testing, reporting the mathematical probability of a fetal anomaly based on the screening test result, and outlining the risks associated with the amniocentesis procedure. The consultation can also include a discussion of the benefits of reassurance and preparedness that fetal diagnostic testing can provide, the woman's right to accept or decline it, and her right to terminate the pregnancy if an anomaly is found.

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Our data come from an ethnographic study of amniocentesis decision making by women from Latino backgrounds who were referred for prenatal diagnosis because they had screened positive for a birth anomaly [Preloran et al., 2001]. The study population consisted of 156 women and 128 male partners who completed a face-to-face, semistructured interview. In addition, we analyzed sociodemographic and other background data from 25 women the clinics classified as "no shows" because they did not return for a follow-up visit and a second chance to be offered amniocentesis (their behavior represented de facto refusal, although we did not have the chance to ask them about their reasons). We also interviewed 60 medical personnel including genetic counselors, geneticists, family practitioners, ob/gyns, perinatologists, nurses, health educators, and the translators whose work included offering prenatal genetic information or services to pregnant Latinas in southern California. We systematically observed 73 clinical encounters where such information or services were offered or provided and 12 prenatal education classes at primary or prenatal care facilities and recorded detailed notes based on our observations.

The cases we offer highlight some of the complexities associated with offering prenatal genetic information and services; the unit of analysis is the actual interaction between the patient, her clinicians, and any others present. We focused on two broad questions in developing our analyses: (1) What factors contributed to the clients' decisions to accept or decline amniocentesis? (2) Were the reasons clients gave for their decisions based on information received directly from a clinician or from other sources? In our examples of amniocentesis refusal based on miscommunication, our intent is not to imply that refusal is wrong, nor that women who relied on biased or incorrect information from male partners or other family members were necessarily victims of undue or unwelcome family pressure. Most such women said they wanted their relatives' help in interpreting the information they received. Likewise, the sources of misinformation we describe should not necessarily be considered the sole—or even the most important—factor in the women's decisions. Like any other, clinical communication is an inherently fragile endeavor [Mattingly and Lawlor, 2001]. Although counselors may genuinely wish to help patients make informed decisions, the priority biomedicine puts on neutrality may indeed deter them from directly challenging patients' views that they know or believe to be wrong.

MISCOMMUNICATION AND PRENATAL GENETIC CARE

There was a distinct and significant connection between women's understandings of the significance of having screened positive and their amniocentesis decision. Whereas most that declined attributed their positive test result to concrete but transitory physical or mental conditions, most who said yes attributed it to mere chance [Browner and Preloran, 2000b]; only a tiny proportion thought there was actually anything wrong with the pregnancy. The explanations for screening positive given by those who accepted were usually a reiteration of the medical information communicated during the genetic consultation; in contrast, the reasons given by those who refused diverged much more widely. This led us to consider the possibility that miscommunication may have influenced the women's decision processes.

It was no surprise to find that misunderstanding appeared to play a role in some women's amniocentesis refusals. Some genetic counselors expressed frustration at what they felt was their inability to convey their message to certain patients, a situation they attributed to patients' low levels of education or ethnic backgrounds. At the same time, we observed many instances of successful communication between prenatal genetic service providers and clients. Counselors almost invariably followed the standard protocol regardless of clients' educational or ethnic background. They detailed the advantages and disadvantages of the amniocentesis procedure and sought to make it clear that any decision (i.e., to accept or decline the test, to continue or terminate the pregnancy) would be the patient's. Most counselors talked sensitively about the often difficult issue of abortion as well as other matters such as caring for a child born with a disability, as well as religious, economic, and practical issues. *Mutuality*, in which the clinician provides expertise and shares the responsibility of medical decision making with the patient [Roter and Hall, 1992], was the approach most counselors sought to emulate.

Most women in our study understood that they had been referred for genetic counseling because certain substances measured by a blood test were either too high or too low. But we also found that clients' interpretations of what this meant *for them* often differed from those of the genetic counselor. To cast some light on the origins and characteristics of these misunderstandings, we have selected several cases that exemplify the recurrent sources of miscommunication we observed. The cases are offered next.

Idioms and Jargon

Health care providers, no less than other professionals, become fluent in a professional discourse. Steeped in specialized terminology and jargon, it may be inevitable that clinicians occasionally lose sight of the fact that terms and usages familiar to them can have different meanings for patients and other laypersons. Prenatal care providers are particularly well practiced in discourse aimed at reassurance intended to manage the anxiety of women undergoing testing. Yet communication is compromised if patients misunderstand either the idioms or the reassurances at the core of health care providers' professional language. In our research we found these kinds of misunderstandings could sometimes be traced back to information clients were given even before their screening test, when they attended prenatal education classes given as part of the State of California Expanded AFP program.

These prenatal care instructors, usually nurses or health educators, are acutely aware of the anxiety most pregnant women experience if they screen positive and have learned to underscore the fact that screening positive does not *necessarily* mean there is a problem with the pregnancy. Sometimes, however, even the most conscientious of these efforts can backfire. In one class we observed, participants were told that they would be offered a "standard" test designed to measure, among other things, a

"protein" produced by the fetus that is also present in the mother's blood. Normal amounts, said the instructor, indicate a "very good chance" that "the baby is healthy." She then continued: "Knowing that everything is normal is a great relief, and don't be alarmed if your test comes out positive. We will call you if it's positive. You will be informed if your baby's protein came out high or low. That means that your baby has low or high protein compared with other babies that were born healthy. That is all." Before dismissing the class she repeated: "I see that most of you have signed the consent form for the test. Good! You will be called only if your test is positive. If we don't call you, don't worry... And if we call you, please, you don't have to worry either. We may offer you other tests that are more complete or more accurate. Because, you know, this test [XAFP], the one that some of you are going to have today, doesn't say much. It's like a red flag that indicates 'be careful, pay attention.' So this is why you might be offered other tests that will reassure you that everything is fine" [Browner et al., 2003, p. 1937].

In this example, the educator did not distinguish between the fetal "protein" for which clients would be tested and the popular meaning of protein, as found in food, to maintain energy. Drawing on the more conventional meaning of protein, many clients attributed their own "low" fetal protein test results to weakness brought about by a poor diet [Browner and Preloran, 2000b] and believed that by changing their eating habits, their pregnancies would no longer be at risk. The educator also failed to explain that a "positive" medical test indicates possible fetal abnormalities. As we will see in the following examples, these misunderstandings combined with providers' repeated efforts to reassure patients that the screening test was "standard," "painless," and "nothing to worry about," may have made a positive test result seem less important. Convinced there was little need to worry, some saw no compelling reason to undergo amniocentesis, which in the minds of many was an exceedingly risky procedure [Browner et al., 1999].

Suggestions

In addition to misconstruing clinicians' reassurances, we found it not uncommon for clients to misconstrue the nondirective nature of genetic counseling. Accustomed to receiving prescriptive medical advice and carrying out "doctor's orders," these clients took the genetic counselor's reluctance to directly recommend an intervention as a sign that the intervention was not truly needed, rather than it being a standard part of prenatal care [Williams et al., 2002a; ACOG, 1996]. For example, one participant we call Maria, age 24, and with no disabilities in her family, was living on a small income (less than $20,000 a year for a family of four). She had first come to the United States 6 years prior to the interview when she was 18 and pregnant by another partner. This was Maria's third pregnancy. She had completed elementary school, worked as a babysitter, and believed that she was going to have a healthy baby because "it moves happily like my first born." The following comes from our face-to-face interview and explores Maria's reasons for declining amniocentesis.

When we asked whether she had thought things over before deciding to decline, she replied that there had been no need because *she already knew* she did not want the test. When called and invited to attend a genetic consultation, she told the "nurse" on the phone that she was not interested in an amniocentesis, but the nurse had insisted that even if she planned to refuse the test, she would be better off attending the consultation "for the [good of the] baby [because] the blood was a little low [and it was better] to have an ultrasound first, and later the test of the needle [amniocentesis]."

Asked what she thought having her "blood a little low" meant, Maria explained she had been told it was from "the baby's protein... because I haven't been eating well." She said she was told she should eat more and that she could have an amniocentesis. When we asked directly why she had declined the amniocentesis, she explained that the clinicians "did not pressure me at all" (*"No me obligaron para nada"*), which she interpreted to mean that they themselves saw no real need for the test. From Maria's perspective, the offer was quite casual: "They said, 'I suggest you consider it, but it is up to you to decide.' They did not recommend it at all; it was only a suggestion. I'm sure I could catch up with my weight if I make an extra effort to eat for two."

In our larger sample, we found that women like Maria usually drew on two types of information when trying to make sense of a positive screening test result: the prenatal classes they had attended and personal experiences of friends and family [Browner and Preloran, 2000b; Mittman et al., 1998]. Both sources led some to decline testing. The medical setting, in its effort to be nondirective and highly reassuring, sometimes resulted in recommendations that were perceived as only casual suggestions. In the home and community setting, the ubiquity of what some counselors referred to as amniocentesis "horror stories," which magnified risk of fetal harm, also inhibited test acceptance. Information drawn from these two sources led some to conclude that amniocentesis was not sufficiently justified in their own cases [Browner et al., 2003, p. 1939].

Cultural Sensitivity

California genetic counselors who typically deal with an ethnically diverse client population generally recognize that they must be sensitive to their clients' cultural backgrounds. Occasionally, however, a counselor's efforts to be culturally sensitive can inadvertently interfere with direct and open communication and, in the process, jeopardize informed choice. Our observations reveal that many counselors are reluctant to challenge the mistaken beliefs of their clients when they appear to be rooted in the client's ethnic or cultural background. The following excerpts come from observations of a formal genetic consultation and that of a more casual encounter between a patient and her genetic counselor.

Genetic Consultation

The participants, 24-year-old Lidia and her husband Rodrigo, 25, have lived in California for 3 years. She is a homemaker and he works in building construction; this is their second pregnancy. Their family medical history is uneventful, except that

one of Rodrigo's brothers was apparently diagnosed with spina bifida as a child in Mexico, although the couple reported that he showed no long-term effects. Rodrigo also confessed to some skepticism about U.S. medical care because he has noticed fewer "mongoloid" and paralyzed children in Mexico than in the United States. During the 35-minute consultation, the genetic counselor avoided discussing what she deemed to be her client's ethnic beliefs because she feared she would jeopardize rapport. She eventually timidly suggested that they check the brother's diagnosis with his mother and even offered to talk to the Mexican doctor in an effort to further clarify the brother's diagnosis. (The couple did not take the counselor up on her offer.)

After an inconclusive ultrasound, the counselor told the couple she was offering amniocentesis mainly to rule out the possibility of spina bifida. Rodrigo responded that it was unnecessary because 20 years ago, his mother's doctor had told her the baby's problem would abate if she took "very good care of herself." Rodrigo proudly explained that was just what she did "and my brother walked normally like you and me." The counselor tried to clarify Rodrigo's misunderstanding by explaining that the severity of the condition depends on the location of the spinal defect and suggested that an older diagnosis could be less accurate. But her explanations were insufficient for the couple to reverse their decision.

Opportunistic Observation

Elena is 20 years old with no disabilities in her family. This is her first pregnancy. She completed secondary school in Mexico, was living with her grandmother, and had no plans to establish a domestic relationship with the child's father. Elena came alone to the genetic consultation and remained mostly silent throughout. She asked for a few minutes alone when it was over, to which the counselor replied, "Take your time." Returning to the waiting room, Elena asked the receptionist for the location of a pay phone and, when she returned a short time later, asked to see the counselor again. She told her that she had discussed the test with her grandmother who had convinced her to agree because "it is the only way to be 99 percent sure that my baby will be 100 percent healthy." Neither then, nor subsequently, did the genetic counselor explain that a negative amniocentesis was not a guarantee that the baby would be "100 percent healthy." The counselor had only said that the test is "99 percent accurate," which Elena reinterpreted to mean she could be "99 percent certain" that the fetus would be fine.

The above are examples of amniocentesis decisions based on clients' misinformation not clarified by clinicians. In the first, the counselor did not try to dispel the mistaken notion that spina bifida may be reversed during pregnancy if the woman takes good care of herself and did not emphasize the increased risk associated with her client's family history and AFP positive test result. In the second, the counselor did not clarify that a normal amniocentesis did not 100 percent guarantee a perfectly healthy baby.

Regarding the first case, when we subsequently asked the counselor to explain her reticence, she said that when clients bring contradictory "ethnic" data to a genetic consultation, she seeks a balance between "respect" for their cultural beliefs and her professional objective to convey the fact that there may be genetic or developmental explanations for the condition in question. In the second case, the counselor said, "I know she agreed for the wrong reason. I know it was because her grandmother talked her into doing it; but I didn't want to intervene. First, I know that among Latinos, family relations are sacred and felt it was not my place to contradict the grandmother. Second, I found no harm in the family advice." These two providers and others like them were uncomfortable with directly challenging clients' cultural beliefs because they assumed it could offend their clients. But, as we later argue, avoiding open discussion of these very issues seems to lead to a serious gap in communication and possible consequences for informed decision making.

TRANSLATION AND SECOND-HAND INFORMATION

Multilingualism is a stark and increasing reality in many U.S. medical settings, and its challenges can take on special salience during pregnancy, when complex information is communicated and important decisions must be considered (see also discussion on linguistic differences in Chapter 7, Informed Consent). Since little is known about the impact of translators on amniocentesis—or other types of medical decisions—we focused attention on this issue. Most U.S. medical settings offer translation services, either through an on-site interpreter (who may or may not have formal training and/or primary job responsibilities such as clerical or nursing) or a telephone translation service. Patients with limited English also may be accompanied by a family member or friend who ostensibly speaks English better than the patient. Either alternative can present its own difficulties, as will be seen below in the examples offered to illustrate how a translator can, in fact, impede comprehension and/or compromise informed decision making.

On-Site Translators

Our data derive from two sources. Part I is from observation of a genetic consultation with Rosalia, in her midtwenties, with no family history of disabilities. Part II comes from our face-to-face interview with Rosalia, immediately after the genetic consultation. Rosalia had resided in the United States for 3 years prior to the interview and suffered continuing economic setbacks. She had completed high school in her native country, was married to a man 20 years her senior, but attended genetic consultation alone. Although she said she had been studying English, at the time of the interview, she spoke it poorly and requested the help of an interpreter. A secretary was called away from her usual duties to translate (but was not unaccustomed to providing such service in that setting).

Genetic Consultation

Assisted by the translator, the genetic counselor asks Rosalia about her family's medical history. The interpreter literally translates the conversation. Rosalia reports

no problems on her father's side but indicates that her younger half-sister's (with her mother's second husband) legs were "semiparalyzed" as a child. Hearing this, the counselor probes to learn more about the half-sister, but her questions, when translated literally (e.g., "*estaba retardada*?"), sound harsh and crude, and Rosalia appears discomfited. To each question about her sister's condition, she repeatedly answers that she is walking perfectly well now. Eventually, the counselor turns to the translator to discuss how the questions can be asked without further upsetting the client. This exchange is not translated and throughout the continuing consultation, the translator looks mainly at the counselor, only once making eye contact with the client. At one point, Rosalia repeats that her sister has improved—as if she wants to convince the genetic counselor that her sister's medical condition should not be of concern: "She is walking well now," Rosalia asserts, "She only has to use special shoes." But the interpreter says only: "She said her sister is O.K. now." As we later show, the client seems to notice that her words are not being faithfully conveyed, enlarging her sense that what she says is being disregarded.

The counselor then explains that the ultrasound showed the pregnancy to be dated correctly and she would therefore like Rosalia to consider amniocentesis. Rosalia appears doubtful. She says she would "perhaps prefer to consider" the test later because right now she is not feeling all that well, adding that she recently had the flu and, in fact, had been eating poorly when she had the triple marker screening. The counselor explains that "the flu" would not cause a positive screening test and urges Rosalia to decide as soon as possible because her pregnancy is very advanced. When Rosalia does not reply, the counselor begins to describe the amniocentesis procedure, adding that it allows the doctors to analyze the chromosomes for the presence of Down syndrome and spina bifida. She then asks Rosalia if she remembers what they had previously discussed about these conditions. Rosalia shakes her head. In response, the counselor asks if Rosalia wants to review the information again, but she quickly says no. The counselor repeats that Rosalia can have the test today and avoid having to make another special trip. The patient responds: "If you want to do it because of my sister, she is well now. My mother says she was a little behind (*quedada*) when she went to school, but she is walking well now. She only has to use special shoes." The interpreter again translates this by saying: "She said her sister is O.K. now." Instead of responding, the counselor sighs and changes the subject, suggesting that Rosalia use the office phone to talk over the situation with her husband.

Interview after Consultation

When we ask Rosalia if she would be willing to discuss her genetic consultation, she readily agrees. When we ask if it was difficult for her to understand the information the counselor conveyed, she says it was, but adds that she disliked the way that the counselor and the translator interacted with her: "They were talking to each other, not to me . . . I told her about my sister, she only needs special shoes. She [the translator] didn't say 'shoes'; I think she didn't say anything [about that]." Rosalia said that she knew the English word for shoes from her English as a second language classes and

complained that they were "very impolite" and excluded her from the interaction. She adds that she much prefers her neighborhood clinic where everyone speaks Spanish and is friendlier and less intimidating. Despite our best efforts to ascertain how well Rosalia felt the information about fetal diagnosis was conveyed, she repeatedly shifts back to the antagonistic emotions she experienced [Browner et al., 2003, pp. 1940–1941].

Family and Friends as Translators

These data come from interviews with two couples: one who accepted and one who declined amniocentesis. Both husbands attended the genetic consultation and served as their wives' translators. Although both couples described their amniocentesis decisions as jointly made, in each case, the men shaped the information they gave to their wives to achieve their own desired outcome.

Elisa, 29, is a housewife married to an insurance salesman. Because her English is rather poor, she wanted her husband to translate the genetic consultation. She explained that she felt comfortable in his doing so because he had been very involved in the pregnancy, accompanying her to all her prenatal visits including the genetic consultation offered after she had screened positive. Regarding the amniocentesis offer, she said, "At the [genetics] clinic, he explained all the pros and cons of the test and later [after the genetic consultation] when we were relaxed at home, I could see things more clearly." There, she said, they had talked "about all the advantages of the test," [the most important being] the opportunity to be reassured," something Elisa said her husband told her the counselor had mentioned several times during the consultation. She added that her husband already knew the advantages of "being prepared" because as an insurance agent, he witnessed many families struggle "to pay medical bills for chronically ill patients." Elisa said that talking with her husband enabled her to see that the "small risk" of amniocentesis was worth taking.

Lia, 28, was employed part-time and married to Pedro, a full-time college student. Both had been raised Catholic, left the Church, and had recently returned to become active adherents to their faith. They were thrilled by the prospect they would soon become parents, having unsuccessfully tried for several years to conceive. Both were fearful they might loose the pregnancy, but Pedro seemed the more anxious of the two about the miscarriage risk associated with amniocentesis. He indicated he would abide by his wife's decision about amniocentesis but also told us he was committed "to helping her to decide by considering all the facts, especially that she might lose this baby." He said he advised her to take time and think "[because] now that we belong to the Church, we can't do whatever we want and in the Church, they say one could kill his own child with that test. I made her realize that that is the truth. If not, look at what happened to my friend [his co-worker's wife who had a miscarriage that he attributed to an amniocentesis]." As the interview was ending, Pedro spontaneously returned to their amniocentesis decision: "For me it represents a lot of responsibility. I am alone, I lost my entire family, I only have her [Lia] and now this child. [For me] it was love at first sight, and when I learned about that test, I was afraid. She

was also. My co-worker told me that when that happened to them [miscarriage], his wife cried and cried—so, I told Lia better not to have the test, because if she loses the baby, what is she going to do? She struggled years to get pregnant."

While, as Roter and Hall [1992] observe, power differentials render client–clinician relationships inherently problematic, these differentials are intensified when actors lack a common language. With these examples, we demonstrate sources of miscommunication that go beyond simple errors in translating words. Skilled interpreting is a creative technique demanding the communication of ideas and concepts as well as the accurate translation of words. Listeners depend upon pauses, word emphasis, eye contact, and a trusting attitude to understand the message. In the first example, those conditions were not fully met. On the one hand, the Spanish-speaking secretary was insensitive to the harsh connotations of a literal translation. On the other hand, she lost the trust of the patient when her translations were not faithful or comprehensive enough. In our second and third examples, women received genetic information through their partners, who clearly had their own agendas. Because research on the use of friends and family as translators in medical encounters is virtually nonexistent, we have included these examples to highlight some of the issues.

CONFIDENCE AND TRUST

Although the relationship between counselor and client is a professional one, it is not wholly impersonal. The degree to which a client can trust and feel comfortable with the counselor can have significant bearing on the effectiveness of communication between them [Rapp, 1999]. Anecdotal data from genetic counselors [Alvarado, 1999; Caldwell, 2000; Tatsugawa, 1998] suggests that minorities in general, and Latinos in particular, may be highly skeptical of the motivation for offering genetic tests and the results that follow. They are unlikely to air that skepticism or engage in a full and frank discussion of the implications of the tests offered unless they trust the counselor. But women from minority backgrounds may find it difficult to develop a trusting relationship with health service providers of different cultural backgrounds, especially ones with whom they have had no prior relationship [Browner et al., 2003, p. 1941]. The following cases of two women, one who declined and the other who accepted amniocentesis, helps illustrate the significance of trust in medical decision making.

Woman Who Declined Amniocentesis

Rosa was 27 and had lived in the United States for the past 2 years. She said she and her family had all been quite fortunate with regard to their health but described her background as unusual in that she had been "touched by adversity." She confided that she had been abused as a child, did not complete elementary school, ran away from home, landed in a relationship at age 22 with a man who tried to kill her, and 2 years later became pregnant—but lost the baby at birth. When she decided "to cross the border" in search of a better life, it was also to get away from abusive relationships. One year later, she found a new partner in the United States and again became pregnant. The partner was currently under arrest, and Rosa was living with

a friend who participated in the genetic consultation and took care of her "out of charity." The friend was also instrumental in motivating Rosa to seek prenatal care at a free clinic close to their home. There, she was offered triple-marker screening and, when she screened positive, additional testing including amniocentesis, which she declined on the spot.

Asked why, Rosa explained that she intended "to have this baby because it is the only family I have" and that she found the risk of amniocentesis unacceptable. Besides, she added, she does not understand why she was offered "the needle test." She proceeded to ask the ethnographer's opinion and, when told that the test is offered for diagnostic purposes, she insisted that in her case, there was no need. She added that in the neighborhood clinic where she attended prenatal education classes, she was informed that "the baby's protein was low, but all other things were fine." In addition, she trusted the doctor who saw her there because he was "a good doctor . . . he helped my neighbor with her baby. I can trust him." Probing about her experience at the genetics clinic, Rosa expressed doubts: "They said the baby could be mongoloid. But how could she [genetic counselor] know only because of that [measurement of AFP]?" When the ethnographer asked whether Rosa had voiced her concern to the counselor, she said no "[because] she doesn't listen to things. I told her about the [neighborhood] clinic [where I was told my baby was fine] and I told her about my [financial] problems, but she didn't pay attention. [In contrast] in the [free] clinic, each time I go there, they check me and [they check] my baby [too]. . . . They said the baby is fine; nobody there told me I needed to have the needle test."

Woman Who Accepted Amniocentesis

Ana, a 28-year-old clinic clerk, is pressed into duty to translate for Rocio, the patient, a 45-year-old woman with a history of hereditary family illness; one daughter, born with Down syndrome, died at age 8 [Browner and Preloran, 2004]. Rocio attributed her daughter's medical problems to an injection, which the genetic counselor said that she doubted. Although Rocio came to the genetics consultation mentally prepared to accept amniocentesis, she changed her mind twice before ultimately agreeing to have the test. This case is a clear example of how the dynamics of ambivalence, trust, and compliance interact in the context of a decision about fetal diagnosis.

During our subsequent interview, we ask Rocio the standardized question: "If doctors recommend a test, such as the amniocentesis, do you think a person should agree because doctors know more than the patients?" At first she categorically answers, "No," but a bit later softens her response adding, "Well, as much as we can, we should follow the doctor's advice," quickly adding "[agreeing is acceptable] but we have to be careful, because sometimes they [doctors] lie because they don't know 100 percent what is going on [referring to her inconclusive ultrasound]."

Asked about the genetic consultation, Rocio explained that she had problems communicating with "the blonde" [the genetic counselor] to clarify her doubts and concerns. She found her "too critical": "If she does not believe in me, I won't believe in her;" and she found her words of comfort false: "If everything is going to be O.K. why would they send me to do that [amniocentesis]?" She said she much preferred

the directness of the translator with whom she "could talk." Asked if her preference was due to the fact that they could converse in Spanish, Rocio said it went beyond that. She said that Ana did not challenge her explanation of the cause of her daughter's problem and openly told her that things could go wrong such as miscarriage, but she should agree to amniocentesis if she wanted to be reassured. Rocio said that Ana's "frankness inspired my confidence" (*esa franqueza de ella me dió confianza*). She added that being so indecisive, she felt relieved and supported when Ana "pushed her a little" to agree to amniocentesis if she wanted to "stop worrying."

These two cases illuminate how the dimension of "trust" can shape the course of a prenatal genetic consultation. Rosa felt that what she had told the genetic counselor about her diet and financial circumstances was dismissed and she reciprocated in kind: "Well, if she doesn't believe me, I don't believe her." Rocio used virtually the same words when describing her own encounter with "the blonde" [genetic counselor], whom she found critical and skeptical: "If she does not believe in me, I won't believe in her." Establishing trusting relationships may be particularly challenging in cross-cultural clinical encounters. In Rosa's case, she gave more credence to clinicians she felt she could trust, such as her neighbor's doctor at the free clinic close to home, than the suggestions of the anonymous genetic counselor. In Rocio's case, although a typical genetic consultation usually lasts about 45 minutes, hers spanned almost 5 hours. During that time, we often observed Ana listening patiently to Rocio, comforting her during the ultrasound procedure, and bringing her orange juice while she was waiting to see the genetic counselor for a second time. Ana's open, warm, and friendly style of communication won Rocio's trust.

DISCUSSION: COMMUNICATION, MISCOMMUNICATION, AND GENETIC COUNSELING

The traditional doctor–patient relationship is rapidly changing, no less in the field of genetics than elsewhere in medicine; the goal today is to forge partnerships aimed at better enabling patients to make informed decisions. But, acting as partners demands mutual knowledge, trust, and unerring communications that are sometimes difficult to achieve, especially when interactions involve more than two parties.

The genetic counselors in our study were genuine advocates for their clients' views and sought to take into account their clients' values about abortion, their socioeconomic situation, their need for concrete medical explanations, and even their capacity to handle anxiety. Indeed, the counselors in our "cultural sensitivity" examples were, if anything, guilty of displaying *too much* deference to their clients' beliefs.

Most clients rated their genetic consultations very highly, even when an interpreter was required [Browner et al., 1999]. The open dissatisfaction one participant expressed (in the "they're talking to themselves" example) was almost an exception. Women assisted with translation by friends and family also reported they were generally satisfied with the experience. Yet we found clear evidence of misunderstandings, even among women who reported satisfaction. This was most evident when it came to women's assessments of risk and choice and their understandings of

the nature of genetic conditions and the meaning of fetal "protein." Genetic testing can also create new ambiguities and uncertainties [Lock, 1998] and may challenge patients to generate creative responses. Many of the women in our study who refused amniocentesis used the opportunity of the positive screening result to forge their own strategies for dealing with the anxieties produced by the positive screen [Browner and Preloran, 2000a].

Prenatal instructors aim to inform women about the benefits and limitations of screening tests. They adopt a discourse of reassurance to reduce anxiety but, by doing so, can also contribute to misunderstandings. Instructors frequently repeated assurances such as, "This test [XAFP] is only a red flag; it doesn't mean too much." Such statements led some women who screened positive to believe they were either at a very low risk or no risk at all. In their own minds, they assumed that prompt remedial actions—usually modification of diet and lifestyle aimed at minimizing stress—would control or eliminate any risks indicated by the test.

For many women, the refusal of amniocentesis was based on perceptions reinforced by the genetic counseling that followed their prenatal classes. Counseling suffered from the fact that patients and health providers, both attuned to a certain model of medical practice, tend to expect a definitive diagnosis and prescription for treatment and cure. Patients do not, however, know or perhaps comprehend that in the field of genetics direct recommendations are usually avoided. Thus, when counselors refrain from making suggestions or recommendations and remain neutral, miscommunication may well occur [Browner et al., 2003].

Our evidence suggests that many of those who declined amniocentesis believed that physical or psychological conditions were responsible for their positive screening result. Clients who confused fetal protein with standard protein, as the word is commonly understood, were especially prone to believe that simple dietary changes could improve their condition. This misinterpretation allowed women to feel more in control and made the risk (*peligro*) much less threatening. This type of misunderstanding is not exclusive to women of Mexican origin; researchers working with other ethnic groups have reported similar findings [Cohen et al., 1998]. Neither is it only manifest among women who refused the test. Some women who accepted amniocentesis also expressed doubts about the biomedical explanation that screening positive was a random event. These women usually looked for more concrete explanations for the test result and sometimes blamed such factors as their spouse's alcohol or drug use for the threat of a genetic abnormality.

If misunderstanding has its consolations, genetic counselors face the difficult task of removing these misunderstandings and possibly depriving clients of the benefits they provide. As we saw in the excess of cultural sensitivity example, while counselors may be aware their clients are emotionally invested in mistaken beliefs, they are sometimes reluctant to address them for fear of undermining trust and rapport. But this is to confuse the means with the end: Rapport is but an aid to open communication, and it should not be compromised in order to create or maintain rapport. In other cases, counselors were quick to point out to clients that their lay theories, preconceptions, or expectations were mistaken or irrelevant. But as we saw in the trust example, clients of Mexican origin, many with limited or no prior knowledge of genetic testing, seem to not be predisposed to accept the guidance of genetic counselors over advice from

other, more familiar sources. Counselors thus find themselves in the difficult position of having to earn the right to have their counsel heard.

Our examples show that clients who do not feel their views have been heard and respected are much less likely to listen to and respect the counselor's guidance. Miscommunication is often the result of asymmetric communication: The client is asked to accept what the counselor has to say before the counselor has heard the client. In one of our confidence and trust examples, the client complained that the counselor "doesn't listen to things"; in the suggestion example, the client mentioned that the counselor "didn't pay much attention." On the other hand, in Rocio's case, another of the confidence and trust examples, she accepted in large part due to the "little push" by the translator who inspired her "confidence"; in the "second-hand information" example, the client complained that her translator disregarded what she had to say. All of the foregoing suggests that the best way for counselors to improve comprehension and win their clients' ears and understanding is to give a better listen to their clients' words.

CONCLUSION

Our research on communication between genetic counselors and pregnant Latinas and their partners revealed predictable sources of miscommunication, including the use of idioms and jargon, inadequate translation, counselors' efforts to maintain neutrality (the edict of nondirectiveness), and a reluctance to directly contradict what they feel are clients' erroneous worldviews. Cutting-edge diagnostic technologies, as much in the prenatal domain as elsewhere in medicine, have challenged the long-established hierarchical relationship between patient and clinician. Providers are expected to share their knowledge and expertise with clients and they no longer have the final word in patient care. Free and open communication, as well as an awareness of the many opportunities for miscommunication will be key to the success of this newly emerging medical paradigm. Since "informed" consent is increasingly recognized as a basic right and goal in medical care, such consent is best achieved when patients have a good understanding of the means and ends of the care they are being offered. This analysis is offered in the spirit of moving closer toward those objectives.

COMMUNICATION: CLINICAL DIAGNOSIS

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HISTORY

Individual III:1 in the pedigree is a 4-year-old-boy of Chinese origin who has been referred to the Genetics Clinic to confirm a suspected diagnosis of neurofibromatosis

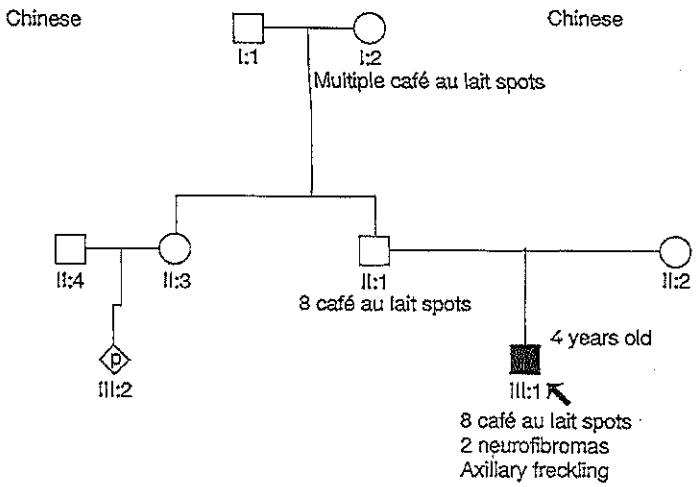

Figure 2.1 Pedigree of family discussed in case history. Numerical identifiers refer to individuals discussed in the text. Arrow indicates proband.

type 1 (NF1). He presents with eight café au lait spots, two neurofibromas, and axillary freckling. Based on his clinical presentation, a diagnosis of NF1 is confirmed. Examination of his parents reveals that his father (II:1) has eight café au lait spots and reportedly the paternal grandmother (I:2) has multiple café au lait spots. II:1's sister (II:3) is currently pregnant. (see Fig. 2.1.)

Background and Diagnostic Criteria for NF1

Neurofibromatosis type 1 is one of the most common dominantly inherited conditions with a prevalence at birth of 1 in 3000. The clinical manifestations are extremely variable, and the diagnosis is based on the presence of two or more of the following clinical findings [diagnostic criteria established by National Institutes of Health (NIH) consensus conference, 1987]:

- Six or more café au lait spots over 5 mm in greatest diameter in prepubertal individuals and over 15 mm in greatest diameter in postpubertal individuals
- Two or more neurofibromas of any type
- Intertriginous freckling (axillary or inguinal)
- Optic glioma
- Two or more Lisch nodules
- Distinctive osseous lesion (e.g., sphenoid dysplasia)
- First-degree relative with NF1