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Chromosomes and Communication: The Discourse of Genetic Counseling

Amniocentesis is one of the new reproductive technologies, and a new health profession, the genetic counselor, has been developed to communicate medical information to patients about its risks and benefits. In this article I examine the language of genetic counseling as it communicates and miscommunicates not only medical information but also structural power arrangements, social knowledge, and popular meanings about medically defined disability. The analysis—based on two years of fieldwork in New York City hospitals observing amniocentesis intake interviews, interviewing 30 genetic counselors, and visiting scores of pregnant patients at home—focuses on the multiple understandings that arise at the intersection of professional and popular knowledge in contemporary American life.

In 1958 Jerome LeJeune (or, according to an alternate telling of the origin tale, a female researcher in his lab) peered through the microscope at samples of smooth connective tissue taken from three patients with Down’s syndrome, and identified the 47th chromosome, the presence of which marks that condition. In the late 1960s, several teams of doctors reported identifying the same extra chromosome in amniotic fluid extracted from pregnant women’s uteruses. This discovery opened up the possibility of diagnosing Down’s and other inherited disabilities prenatally.

The practice of amniocentesis for prenatal diagnosis of inherited disabilities increased as techniques for identifying, staining, and studying chromosomes developed. At the same time, the DNA revolution transformed the fields of cytogenetics and cell biology, and by the late 1980s, more than 200 inherited conditions—most of them extremely rare autosomal recessive diseases—could be diagnosed prenatally. Research frontiers in human genetics and molecular biology became inextricably tied to the routinization of amniocentesis.

The rapid proliferation of information in human genetics has implications not only for health workers in the field of reproductive medicine but for pregnant women and their families as well. How were those technical and ever-shifting implications to be explained to the lay public? The profession of genetic counseling developed to translate the discourse of human genetics into usable and more popular language.

In this article I explore the conversation between professional genetic counselors and their multilingual, multicultural clients. The work is based on two
years’ field study in New York City, following a team of genetic counselors through their hospital-based rounds, interviewing 35 genetic counselors about their work, observing a cytogenetics laboratory, and interviewing pregnant women and their families. A discussion of genetic counselors and the development of genetic counseling as a new health profession introduces the analysis. An interpretation of genetic counseling sessions, using a language-based perspective, presents the core of my argument. This perspective allows me to understand genetic counseling as a discourse in which multicultural clients and science-based health professionals communicate and miscommunicate. The power and limits of technical medical language are explored in the next section.

While the language of science claims to be universal, it must, in fact, confront the local idioms with which diverse groups and individuals respond to its powerful messages. While such salient social distinctions as class, ethnicity, race, and gender deeply influence clients’ communication choices, they do not determine them. Thus a counseling session is also a context in which meanings are actively and interactively produced. The fourth section concerns variability in the meanings of disabilities, and the ambiguities of “choice.” I conclude with a discussion of the power of genetic communication: counseling discourse has the power to create and highlight some problems, while masking and silencing others. Throughout the analysis, I implicitly describe the discourse of genetic counseling as caught in a contradiction: developed to provide pregnant women and their families with more choices, it also inadvertently and unself-consciously replicates and extends the existing social hierarchies that limit choices.

**The Genetic Counselor**

In 1969, program developers at Sarah Lawrence College in Bronxville, New York, planned a new master’s-level science degree, and two years later, the first formally trained genetic counselor was born. “Genetic counseling,” a label coined in 1947, initially stood for a position of ethical neutrality, favoring personal choice in the century-old eugenics debate concerning society’s responsibility to encourage or discourage reproduction in certain individuals and families (Kevles 1985; Reed 1974). Prior to the invention of the modern genetic counselor at Sarah Lawrence, research pediatricians, geneticists, immune biologists—that is, medical doctors and researchers, the majority of whom were men—would counsel families with genetically disabled members about recurrence risks and disease management. Once amniocentesis developed as a clinical service, the need for an “interface” also developed: someone to convey the risks and benefits of the test, to translate scientific possibilities into personal calculations for potential patients. That “gatekeeper” between science and social work, between epidemiology and empathy, became a woman. Women students seemed especially suited to a field that was designed to counsel pregnant women. And “counseling” was a field in which “female qualities” seemed particularly appropriate.

Since the program at Sarah Lawrence was mounted through the School of Continuing Education, the first wave of genetic counselors tended to be well-educated housewives living near the college, often wives of doctors, lawyers, and businessmen. Many had raised their children and were ready to go back to school. They brought with them enormous resourcefulness and a specific set of upper-
middle-class family values. The program promised an amalgam of hard science and counseling skills, while also holding out the possibility of part-time work. Advances in genetic science thus provided for the first wave of genetic counselors an exciting way to leave home and enter the world of public work.

Genetic counseling quickly became a profession-in-formation (Rollnick 1984). While Sarah Lawrence remains the largest and perhaps the most respected program in the country, there are now nine others located in seven states, the District of Columbia, and Canada. While the curriculum varies somewhat, all provide training in human genetics, medical genetics, counseling skills, and supervised clinical internships. Some programs include a seminar on bioethics or ethical issues in genetic counseling, with a strong orientation toward individual choice. According to the counselors I have interviewed, however, there is not much discussion of the cultural constraints and resources with which different pregnant women and their families may be operating. Most programs require two years’ study. Board certification for practitioners is under way, and genetic counselors are insured by the medical centers for which they work. The National Society of Genetic Counselors (NSGC) estimates that there are currently about 600–700 practicing genetic counselors, including those who have completed a two-year MS program and served supervised internships, as well as those “grandfathered” (in this case, “grandmothered”) into the profession through prior experience in nursing, social work, and related fields.

Genetic counseling remains a “woman’s field.” Fewer than 1% of the graduates of genetic counseling programs are men, and many of those are employed in administration. Current students and recent graduates are less likely to have attended elite women’s colleges, are more likely to have considered a premed track and rejected it, and are somewhat more diverse in class, ethnic, and racial background than the first wave of graduates. While consciousness of minority issues in counseling is growing, the number of NSGC counselors from Afro-American, Hispanic, or Asian-American backgrounds remains small—not more than 5%.

As part of an ongoing study of the social impact and cultural meaning of prenatal diagnosis, I observed five genetic counselors working for New York City’s Department of Health during their counseling sessions one to two days a week for more than two years, sitting in on more than 200 intake interviews. As an observer and sometimes as a participant-translator, I have had extensive informal conversations with the five women who allowed me to observe their work. I have also interviewed 30 other genetic counselors who work in New York City, at least one from each medical center that offers amniocentesis. While most of this sample would identify themselves as white and middle-class, at least five spoke Spanish as their first language, and one was Afro-American.

In New York City, unlike many other parts of the country, a combination of state and city programs both fund and offer prenatal diagnosis to women, regardless of their ability to pay for the test. The City Laboratory (through which my fieldwork was conducted) collects samples of amniotic fluid drawn from pregnant women, approximately one-third of whom are Hispanic, one-third Afro-American, and one-third white. The Lab has contracts to perform cytogentic analyses for 24 urban hospitals, both municipal and voluntary, in all boroughs and neighborhoods. Women seen by the city’s genetic counselors are roughly 50% private
patients and 50% clinic patients (that is, poor and working-class women). The city’s genetic counselors are “circuit riders,” rotating among six to eight hospitals that serve the gamut of urban clients. Genetic counselors must thus confront a polyglot diversity of patients as they attempt to explain the test. In the process, they help to shape experiences of knowledge and power in the reproductive lives of women, many of whose backgrounds are significantly different from their own.

As a new professional, the genetic counselor tends to be highly self-conscious of her ethical and scientific roots. The counselors I interviewed were very much aware of the anxiety, as well as the relief, that their services invoke. Most had thought deeply about why someone might reject as well as accept amniocentesis and possibly abortion. Moreover, many were curious and insightful about the problems of cross-cultural communication that my interview schedule posed.

The Discourse of Genetic Counseling

A genetic counseling session almost always precedes the use (or non-use) of amniocentesis. Counselors meet with their pregnant patients (and any supporters the woman brings with her) in the hospital where the test is offered. In the course of an hour’s visit, counselors convey a great deal of medico-scientific information, ask and answer questions, and prepare women to take the test. The interaction is conversational: in a session, meanings are actively and interactively produced by patients and counselors together. But the discourse of genetic counseling is resolutely medico-scientific, revealing and creating some meanings that mask or silence others. Medical language commands great authority in the interview and cannot always respond to the resources and questions particular women bring to this encounter. Miscommunication as well as communication, silence as well as conversation, characterize a genetic counseling appointment.

Genetic counselors generally begin communication with pregnant patients with three goals in mind: to convey significant information about the risks of birth defects and the availability and nature of amniocentesis; to take a health and family history; and to communicate with the patient well enough so that her questions and concerns can be addressed. In order to accomplish these tasks, most genetic counselors begin by “setting up a dialogue.” Many begin intake sessions by posing some variant of the question, “Do you know why you are here talking to me?”

From the beginning, interactions are context-sensitive to the responses and resources patients bring to the interaction. Middle-class, scientifically educated pregnant patients may respond, “We’re planning to have amniocentesis,” thus pushing the counseling script into high gear. Implicit in that answer is not only knowledge of appropriate medical indications for the test but also the existence of “the couple” as a decision-making unit. But a Dominican mother of three may answer, “por culpa de me edad” (literally, “for the fault of my age”), thus presenting the counselor with several options. She can assume that the woman “knows,” since “age” is the factor that sends her here. Or she may respond to the possible implications of “culpa,” explaining that older women having babies presents no shame, just medical risks. But wherever the counselor begins her routine explanation, she is likely to have to adjust her language to the language and assumptions of her pregnant patient (and sometimes the patient’s mate).
In adjusting (or not adjusting) to the patient’s language, genetic counselors are bound by the limits of their own communicative resources. Although many languages are spoken by the pregnant women whom counselors see, most counseling sessions are conducted in English. There are at least five counselors in New York hospitals who are native Spanish speakers, however, and another five who are comfortable counseling in Spanish. Many others have learned a bit of medical Spanish and work through translators, ranging from trusted assistants who understand their agendas (a secretary in the office of a clinic nurse) to catch-as-catch-can interpreters (from the 10-year-old child brought along by the patient to a husband, brother, or male neighbor, embarrassed to find himself discussing prior miscarriages and abortions). Availability of fluent translation is a significant problem: depending on the hospital’s catchment area, patients may be close to 100% Spanish speaking, 50% French/Creole speaking, or about 25% monolingual Spanish speaking.

“Native language” only approximates the variety of communicative differences that genetic counselors confront. “Hispanic” glosses a range of Spanish-speaking cultures, especially at the present time in New York City. Some genetic counselors distinguished “Hispanic” (which often meant Puerto Ricans and Dominicans, the “old” migrants) from new migrants, who might be “middle-class” Colombians and Ecuadorians or the “field mice” of Central America (by which the speaker meant “the poor, rural, and humble”). Although exact cultural differences among Spanish-speaking groups may be unknown, most counselors recognize something of the diversity in educational levels, familiarity with medical terminology, and religious observance that different nationalities may represent.

Nominal or deep fluency in another tongue may aid but does not ensure direct communication for science speakers. Language differences may signal communicative ambiguities far beyond the question of literal translation. Local metaphors of pregnancy, birth, and parenthood do not necessarily translate easily into the realm of medical discourse. Two native Spanish-speaking counselors pointed out the far-reaching impact of their conversations with pregnant patients.

This knowledge is more than genetic. They learn about things that were completely hidden—where the eggs are, what sperm does, how children get to look like their parents. They have ideas, but this is female physiology, it is knowledge, not just information. For this, they come back.

When I see confusion, I go to work, I tell them in language they will understand, language of the streets. They are comfortable here, it is a good place to visit. They come back to see me whenever they come to the hospital.

Of course, not everyone is equally open to the complex relation between native tongue, knowledge, and communicative power. Two counselors, one of whom conducts group sessions for patients in Spanish, expressed irritation that so few of their clients “bothered to learn English”:

They’re here 10, maybe 15 years. They learn enough English to pick up their welfare checks. Why don’t they just learn the language? My grandparents did.

And if speaking Spanish is a contested domain, French and Haitian Creole are virtually terrae incognitae. Only one genetic counselor feels comfortable counseling in French; none knows any Creole. The lack is significant: in at least
one city hospital, Haitians make up about 50% of the patients referred for counseling. In translating for counseling sessions, I discovered that there is no recognition of Down’s syndrome or mongolism among recent immigrants from the Haitian countryside. No word exists in Creole for the condition. In principle, the incidence of Down’s syndrome is invariant world-wide. But in a country with the worst infant mortality statistics in the Western Hemisphere, babies may die from many causes, and this one may go unrecognized as a “syndrome.” Nonrecognition of the label may also reflect other cultural and political experiences. Haitians living in New York City have already confronted alternative definitions of their children’s vulnerabilities. As one Haitian Evangelist father told me, while firmly rejecting amniocentesis on his wife’s behalf,

What is this retarded? They always say that Haitian children are retarded in the public schools. But when we put them in the Haitian Academy [a community-based private school], they do just fine. I do not know what this retarded is.

In his experience, “chromosomes” seem a weak and abstract explanation for the problems a Haitian child may face.

The Use of Technical Language

Language and cultural understandings are linked in other ways as well. Statistics and medical terminology are genres of communication, not simply neutral vocabularies. Both convey and delimit the quantity and quality of information that a counselor provides. The language of genetic counseling is resolutely statistical; it is an axiom of good counseling that a patient must be told her risks before she can decide to take or refuse the test. Yet “statistics” implies an abstract mathematical universe that may not be shared by clients who have little formal schooling. The majority of genetic counselors confront this problem by simplifying the numbers and adding information if it is requested.

[To someone perceived as unable to handle numbers] At your age, the risk of having a baby with mongolism is about one in a hundred.

[To someone perceived as uneducated, but attentive] Pregnant ladies your age have a one in 106 chance of having a baby with this condition. That means that of every 106 pregnant ladies your age, 105 will have no problems, and one will have a child with the problem.

[To someone perceived as scientifically sophisticated] At 35, a woman’s risk of bearing a liveborn child with Down’s syndrome is one in 385; at 40, it increases to one in 106; at 45, it is one in 30.

Likewise, miscarriage rates following amniocentesis must be shared by using probabilities.

This is a very safe test, but there’s always some risk to any test in medicine. The risk of losing the baby after amnio is very small, but it isn’t zero.

Amnio adds three miscarriages per 1,000 to those having the test. Of 1,000 women your age 16 weeks pregnant who don’t have amniocentesis, 32 will not have a liveborn child at the end of the pregnancy, through miscarriage or stillbirth. Of 1,000 women who have the test, 35 will lose the pregnancy.
But code switching and simplification of numbers only mark the professional side of the interaction. Such strategies may sit comfortably with information-seeking, medically compliant patients, especially those with some advanced education (that is, middle class), but they often gloss over the reality of less privileged women. Low-income Afro-American women, for example, often expressed a sense of statistics based on personal experience which varied radically from the perspective of middle-class couples. When a woman has given birth to four other children and comes from a family of eight, and all her sisters and neighbors have had similar histories, she has seen scores of babies born without recognizable birth defects. It requires a leap of faith in abstract reasoning to contrast these experiences with a number produced by a lady in a white coat proclaiming that the risk of having a baby with a birth defect is steadily rising with each pregnancy. Among middle-class professional families, where childbearing is likely to be delayed, the counselor is discussing a first, or at most a second pregnancy. Children are likely to be scarce throughout the network of the professional couple. To them, one in 300 sounds like a large and present risk, while for the low-income mother of four, the same number may appear distant and small.²

The vocabulary of biomedicine describes pregnancy and birth, abortion and disability in ways that may result in a tug-of-war of words. In a 45-minute intake interview, code switching occurs rapidly, as counselors feel out their clients: “babies” vie with “fetuses” for space in “wombs,” “tummies,” or “uteruses”; “waters” or “liquid” or “fluids” may be “taken out with a needle,” or “withdrawn through an insertion”; the “test” or “procedure” may involve “looking at the inherited material” or “examining chromosomes.” And, in the worst case scenario, women must decide to “terminate” or “abort” an “affected” or “sick” pregnancy in which “Down’s syndrome” or “mongolism” has been diagnosed. In the stand-off between medical and popular language, the more distant idioms may provide reassurance by suggesting to some pregnant women that their experiences are part of medical routine (Brewster 1984), but for others, medical terminology may muffle anxiety-provoking choices until they are expressed through dramatic disruption.

So I was sittin’ and listenin’, listenin’ and sittin’ and all the time gettin’ more and more preoccupied. The counselor kept on talkin’ but she never did say it, so finally I had to just say it, right while she was still talkin’. “You can’t take the baby out then [i.e., so late in pregnancy], can you now?” I finally asked. [Veronica Landry, 36, Trinidad-born factory worker]

Much of the scientific information that counselors want to convey is technical and invisible. Most counselors therefore work with visual aids, especially with less educated patients, attempting to map with charts, graphs, and karyotypes what patients cannot see for themselves. Many show pictures of children with Down’s syndrome, and almost all discuss the sonogram accompanying amniocentesis, in which “you can see the baby moving around.”

Developed to present the mysteries of the womb, the workings of heredity, and the universe of epidemiology graphically, such icons of professional knowledge are not, to be sure, self-evident. They require interpretation during which health professionals not only reveal some of their arcane wisdom but also shape the perceptions of the client.
I saw the sonogram of the twins, and I was thrilled. But I really couldn’t read it, I didn’t know what it meant. They had to interpret it for you, to say, “Here’s a heart, these are arms.” Afterwards, it made me queasy—they made the babies real for me by telling me what was there. If they hadn’t interpreted, it would have just been grey blobs, and now I’m more frightened to get the results of the amnio back. [Daphne McCarle, 41, U.S.-born professional].

It was nothing, really, it looked like nothing. Then they showed it to me, and made it something. [Ileana Mendez, 37, Ecuadorian-born babysitter]

Virtually all counselors have a minimal “threshold” of information they need to explain. For some, it includes the concepts of chromosomes and genes; for others, it is the idea of heredity, especially hereditary health problems. For most, it is the 2–3% risk of a birth defect involved in any pregnancy and the increased risk of chromosomal problems associated with childbearing later in life. Whatever her personal goal, no counselor is satisfied if she feels that a patient has failed to grasp her minimal scientific scenario. But medically significant lessons may mask the social experiences and meanings that disabling conditions hold, a point to which I return below. Scientific discourse silences, as well as expresses.

In their training, counselors are taught to illustrate chromosomal problems by reference to the 47th chromosome, which is clinically expressed in Down’s syndrome and its accompanying mental retardation. While this condition is almost universally recognized, the content of that recognition varies considerably. Many families share the counselor’s concerns about the limitations on independence that mental retardation represents in our culture. Yet in families who have had direct experience with children with Down’s, consciousness of disabilities is more finely honed: Down’s children may be mildly, moderately, or profoundly retarded; they may also suffer from heart or esophageal problems, hearing loss, or increased risk for leukemia. “Mental retardation” provides an iconic description that blurs differences among Down’s children, even as it categorizes them. Yet counselors do not routinely offer information on the social realities that accompany this condition (or any other). Many counselors are open, in principle, to questions concerning support for families with genetically disabled members. However, they have little or no experience in discussing either the social stigmas of Down’s syndrome or the infant stimulation programs that a newborn with the syndrome requires. Unless propelled by a patient’s question, they do not transcend the medical frame of the discussion by offering to arrange a visit with a family whose child has the diagnosed condition about which the woman is concerned. Thus the counseling session is likely to reinforce a medical, rather than a social, definition of the problems of childhood disability.

Indeed, the assumptions of medicine weigh heavily throughout an intake interview. Genetic counselors elicit health histories, using a standard questionnaire. From a counselor’s point of view recent immigrants, especially from very poor countries, are likely to exhibit shallow knowledge of their own heredity. The cause of a father’s death or the name of an uncle’s form of mental retardation may be unknown, especially if births and deaths have occurred after the immigrant left home. And while immigrants may vividly recall some health experiences—high fevers, exposure to x-rays—others seem irrelevant, or are named in a language they do not speak. Many Haitians, for example, routinely answer “no” to all questions concerning family histories of heart and kidney disease, diabetes, and
venereal diseases. Their negative answers may well be ambiguous: serious conditions may be unreported or unnamed, since they are virtually untreated for all but the most privileged elite. For example, a community health outreach worker from Jamaica, now working in Brooklyn, told me this part of her life story.

Sickle cell, do I counsel sickle cell? Sure I do, now. But then, I didn’t know what it was. My brother, he died of it back then. We didn’t know, no one told us. What’s the difference? No transfusions back there, anyhow.

To the counselor, the “no” may result in one of a number of misinterpretations—for example, an absence of health problems in the family, or a lack of interest or intelligence on the part of the patient. These interpretations are overdetermined by the individual nature of a medical health history, in the absence of an epidemiological and cultural context.

The codes, genres, and assumptions of biomedicine construct the limits of the conversations genetic counselors may have with their patients. Biomedical claims of universality silence other cultural resources and world views. The language of biomedicine also limits communication by locking counselors into a discourse in which technical language dominates, despite a sincere desire on their part to reach out to patients. Counselors are caught between the need to sound authoritative and the desire to “glide on the patient’s wavelength,” as one counselor described the situation.

Genetic counselors, as new professionals, must lay claim to a monopoly over the information that they offer. Such a claim must impress the rest of the medical hierarchy, in whose ball game they are new players. At the same time genetic counselors are extremely interested in the public image of their field as a new field, and many are eager to further public education about genetic issues. Individually, many enjoy speaking at popular health forums, being interviewed by journalists, and serving as consultants to media projects which they hope will disseminate scientific knowledge effectively. This tension between monopolizing an arcane body of information and popularizing it is inherent in any new health profession: members need to “sell their services” to both the medical establishment and the individual clients for whom they serve as translators (Brown 1986). But this tension cannot easily be resolved in favor of popular language: medical discourse is authoritative, it pervades the hospital setting, and it claims universality. Patients are often multilingual, and usually have no lexicon, or have an unacceptable lexicon, for the expression of clinical and epidemiological facts.

This contradiction between speaking like doctors and talking with patients surfaces in nonverbal communication as well. Some counselors wear white coats “to appear like medical professionals,” for example, while others forego that symbol of antiseptic separation, hoping to “make the patient right at home.” One counselor described her dress code to me.

On clinic days I like to dress comfortably, so they will feel at home, casual with me. I dress up like a professional when I see private patients; they expect more from their medical providers.

The inscriptions of professional status extend to briefcases, charts, and visual aids, like those of both health care professionals and their private clientele. But the poor carry inscriptions with them, too. Among non-English speakers,
there are often electricity bills, check stubs, or personal letters, carried to communicate home addresses. The ubiquitous Medicaid and clinic cards that define payment status and rights serve as passports into medical domains, marking the client status.

Disability, Variability, and “Choice”

In a genetic counseling intake interview rich, many-layered, and powerful messages are being communicated. Officially, information about a new reproductive technology, its risks, limitations, and possible benefits is being conveyed. Unofficially, the power to define a pregnancy, fetus, disability, and maternal responsibility for fetal health are all under negotiation. Basic knowledge about human heredity, reproduction, and their control simultaneously offers the potential for self-control and social control, as several of the Spanish-speaking counselors tried to indicate. The counseling situation is structured so that women (and their families) are given a set of choices about the kinds of babies they might, or might not, accept bearing.

I was hoping I’d never have to make this choice, to become responsible for choosing the kind of baby I’d get, the kind of baby we’d accept. But everyone—my doctor, my parents, my friends—everyone urged me to come for genetic counseling and have amniocentesis. Now, I guess I’m having a modern baby. And they all told me I’d feel more in control. But in some ways, I feel less in control. Oh, it’s still my baby, but only if it’s good enough to be our baby, if you see what I mean. [Nancy Smithers, 36, U.S.-born lawyer]

Such choices are, of course, far more than individual, for they emerge from the embeddedness of each specific pregnancy in ethnic, class, racial, religious, and familial experiences. As the excerpt above indicates, Creole-speaking Haitian immigrants from the countryside may not recognize Down’s syndrome. In this case, being “offered the test” for a condition that holds no cultural meaning may make no sense. Recognition of a medical condition may also be contextualized within other powerful discourses. For example, an Ecuadorian evangelical and a Colombian Catholic, both opposed to abortion but still wanting the test, made similar points to me: “Science can reveal God’s miracles, let you know what He has in store.” Their desire for the test might be interpreted as a bridging of religious and secular-scientific cultures, an expression of the necessities and possibilities of living as fundamentalists in a multicultural world. And even for black Americans raised with an awareness of medical discourse, its choices may seem inappropriate: there is no inevitable bridge between a positive diagnosis and an abortion. One pregnant black woman who was adamant about her anti-abortion stance, for example, wanted the test in order to know whether she ought to move back to Georgia, where her mother would help her raise a disabled child. Another, having recently given birth to a baby with Down’s, said to the genetic counselor, “My kid’s got a heart problem. Let me deal with that first, then I’ll figure out what this Down’s business means.”

Mental retardation is the key focus when genetic counselors speak about Down’s and offer amniocentesis, but it may not be the most significant factor in the consciousness and decisions of many of their clients. Among the Spanish speakers I interviewed, physical vulnerability, especially if it was highly visible,
seemed a much more urgent problem for family life. Low-income Puerto Rican parents I interviewed at an infant stimulation program said of their daughter with Down’s syndrome, “She’s growing really well. We were only concerned that she wouldn’t grow, that she’d be really small. But now that she can walk, and she’s growing, she seems like a normal child to us.” Thus, the “choice” any pregnant woman makes to take or reject the test, and to keep or end any specific pregnancy, flows from the way that pregnancy is embedded in the totality of her life.

If Down’s syndrome represents the iconic case for genetic counseling, other potential diagnoses are more ambiguous and present complex problems for communication. The sex chromosome anomalies (XXY, or Klinefelter’s syndrome; XYY; Triple X; and Turner’s syndrome, or XO) all spell problems with growth and fertility, but none is “incompatible with life,” as medicine would express it. There are controversial claims concerning mental retardation, learning disabilities, and antisocial behavior with these conditions, but all are contested because there is no baseline population from which to make scientific comparisons. Only people who are diagnosed as having a clinical problem will ever have their chromosomes “read.” And even when the ethical complexities of collecting baseline data on anomalous sex chromosomes are sorted out, epidemiological patterns cannot predict whether affected individuals will express many symptoms of the condition and whether the clinical expression will be severe or mild.

Thus, when one of these diagnoses emerges in a test tube of fetal cells, its meaning is open to interpretation. One genetic counselor encountered two patients, each of whom chose to abort a fetus after learning that its status included XXY sex chromosomes (Klinefelter’s syndrome). One professional couple told her, “If he can’t grow up to have a shot at becoming the President, we don’t want him.” A low-income family said of the same condition, “A baby will have to face so many problems in this world, it isn’t fair to add this one to the burdens he’ll have.” A Puerto Rican single mother who chose to continue a pregnancy after getting a prenatal diagnosis of Klinefelter’s said of her now four-year-old son,

He’s normal, he’s growing up normal. As long as there’s nothing wrong that shows—he isn’t blind or deaf or crippled—he’s normal as far as I’m concerned. And if anything happens later, I’ll be there for him, as long as he’s normal looking.

From a parent’s point of view, most diagnoses are inherently ambiguous. An extra chromosome spells out the diagnosis of Down’s syndrome, but it does not distinguish mildly from severely retarded children. Nor does it indicate whether this particular fetus will need open heart surgery. A missing X-chromosome indicates a Turner’s syndrome female but cannot speak to the meaning of fertility in the particular family into which she may be born. Homozygous status for the sickle cell gene cannot predict the severity of anemia a particular child will develop. All such diagnoses are interpreted in light of prior reproductive histories, community values, and aspirations that particular women and their families hold for the pregnancy being examined.

Values, Decisions, and Power in Genetic Counseling

The ethical complexity of diagnoses is something all genetic counselors confront. Counselors are trained to be empathic as they convey statistics and to prac-
tice Rogerian therapy—that is, to use a therapeutic style that is noninterventionist, aimed at helping the patient make up her own mind. This counseling model assumes that the professional and the patient mutually participate in a decision-making process. Their task is short-term and well-defined by two key questions: should the woman accept amniocentesis, and if a positive diagnosis is made, should she end or continue the pregnancy? As distinct from a directive model (which counselors attribute to physicians), the counseling protocol assumes that the patient can and must decide for herself.

Yet counselors all know how hard it is to keep their own feelings out of a given situation.

We’re supposed to ooze empathy, but stay aloof from decisions.

Oh, I know I’m supposed to be value-free. But when you see a woman on welfare having a third baby with one more man who’s not gonna support her, and the fetus has sickle cell anemia, it’s hard not to steer her toward an abortion. What does she need this added problem for, I’m thinking?

So I try to put it in neutral, to go where she goes, to support her whatever her decision. But I know she knows I’ve got an opinion, and it’s hard not to answer when she asks me what I’d do in her shoes. “I’m not pregnant,” I say, “remember that.”

A social worker who trained me at Sloan-Kettering taught me something important: to clear my own agenda before I walk into the room, to let the patient set the agenda. It’s the hardest lesson, and the most important one.

Despite this consciousness, counselors stand in a contradictory position with respect to their clients’ decisions. For they are always making choices about what sort and how much information a pregnant woman needs and can use, as well as the form in which she can best absorb it. At the same time, most of the information that the woman receives comes directly from the counselor, since she is unlikely to have a folk model of most of the diseases and risks associated with amniocentesis. This is not true of any other aspect of pregnancy or pregnancy loss, where medicalized views are often countered by ideas and images shared among communities of women. Here, communication about the health or illness of a potential child is shaped in a vocabulary that is exclusively medical, a grammar that is technological, and a syntax that has yet to be negotiated. As in so much of modern biomedicine, the genetic counselor really is the gatekeeper between science and social experience, regulating both the quantity and quality of the information on which decisions will be made.

At first glance, then, genetic counselors appear to control and shape communications because they hold near-exclusive access to the medical information that patients have come to discuss. But patients are not silent partners in these encounters, no matter how few sentences they utter. Ten percent of private patients and 50% of clinic patients break their appointments for genetic counseling, and somewhere between 20% and 50% of those counseled decide not to have amniocentesis. While their reasons are varied, they certainly include a disbelief or nonacceptance of the medical premises behind testing for fetal disabilities. The conflict often takes the form of a clash of cultural assumptions, with pregnant women saying, “God will protect my baby,” and counselors saying, “Most babies are born healthy, but 2–3% of all babies are born with birth defects.”
Sometimes an interaction will reveal an especially clear instance of a shift in the meaning of motherhood, with technocratic and traditional images uncomfortably located in the same sentence. As a low-income Chinese-American woman said when queried about her own desires, after her husband had signed her up for the test,

My mother, my grandmother, they all had babies in China, and nobody did this. They wouldn’t do it now, if they were here. Now is modern times, everyone wants to know everything, to know as soon as possible, in advance, about everything. What kind of information is this? I don’t know, but I will soon have it, faster than I can understand it.

Most counselors insist that ‘‘I’m not here to sell amnios’’; ‘‘I don’t feel like a success or failure according to whether or not she takes the test.’’ Their interests lie in ‘‘informed consent,’’ that peculiarly American medical-legal document that attests to an individual’s acceptance of information properly conveyed. Such an individual contract model is highly appropriate to a litigious society without a national health plan, where the only remedy for lack of information or services, or for harmful information or services, is the malpractice suit. When viewed in this larger context, the counselor provides protection for the doctor and the medical center, ensuring their invulnerability to legal suits despite the chaotic conditions of an ‘‘information revolution’’ in which the techniques and interpretations of genetic diagnoses are continuously in flux.

When viewed culturally, however, the process of obtaining ‘‘informed consent’’ is not simply the exchange of information-for-signature negotiated at the intake interview. It is based on all the assumptions, fears, and norms concerning healthy and sickly children with which any given woman undertakes a pregnancy. It includes the meaning of illness in family history; the shame and pride attached to the bearing (or non-bearing) of children; beliefs about fertility, abortion, femininity, and masculinity; and the social consequences and prejudices surrounding disability (including the ‘‘courtesy stigmas’’ borne by those close to disabled people) (Goffman 1963).

In this larger context, knowledge and power are not reducible to medical terminology, despite medicine’s hegemonic claims. For surely this new technology has potentials that are at once both emancipatory and socially controlling, depending on the context in which its use is shaped and practiced. Genetic counselors, no less than their polyglot patients, are heirs to a eugenic script in which aspirations for the liberation of women and children necessarily confront the current conditions under which family life is enacted. If, and only if, the discourse on disability and reproductive rights is lifted out of the medical context and negotiated as part of popular culture will it become possible to speak in other languages.

We may occasionally catch glimpses of the effects of such displacements of medical culture by examining amniocentesis and disability among children as they are inscribed by mass media. To my question, ‘‘Where did you first learn about amniocentesis?’’, many women without advanced formal education answered ‘‘Dallas,’’ ‘‘St. Elsewhere,’’ or the National Enquirer. An Ecuadorian domestic worker told me she’d learned about spina bifida (for which amniocentesis is offered) from ‘‘Jerry’s Kids’’ (the Jerry Lewis telethon). Several patients
gave articulate, up-to-date descriptions of children with Down’s syndrome after Phil Donahue devoted a show to them. Middle-class amniocentesis patients often arrived armed with new questions after genetics stories appeared in the science section of the New York Times. As teaching and learning about inherited disabilities and even amniocentesis increasingly permeates the world of mass culture, medico-scientific discourse will have to confront its own popularization and challenges to it. Medicalized melodramas and prime-time primers on genetics have entered the discourse of reproduction and disability.

A more popular and widespread consciousness about both birth defects and the technologies aimed at their screening may benefit both pregnant women and counselors. Pregnant women might then come to a decision from a more nuanced and knowledgeable position, with opinions partially constructed in frameworks larger than the medical. Counselors might be relieved of some of the burdens of protecting the medical hierarchies within which they now work. Under those circumstances, we might begin to discuss the stakes that women and their supporters hold in particular pregnancies as technological transformations reconstruct the intersection of disability and reproductive rights.

NOTES

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1The first NSGC workshop on “counseling the culturally different” was conducted by a team of minority genetic counselors at the annual meetings in 1985. Members of that team estimated that no more than 30 minority counselors are trained and practicing nationally. Both the definition of “minority” and of “practicing” are open to interpretation, however. Latin Americans from privileged backgrounds “become” minorities when they enter the American system of racial and ethnic stratification. And several “minority” counselors have moved into medical school or health care administration for reasons they describe as both political and financial.

2Moreover, many low-income minority women expressed a healthy skepticism about “white” medicine and hospital procedures, as well as a fear of being treated like “guinea pigs” in experiments. While this skepticism was not linked directly to rejection of statistical explanations, it adds to the distance clients may feel from counselors.

3This is not true of genetic counselors who work in pediatric, as opposed to obstetrical, services. Increasingly, however, most counselors work exclusively in prenatal care, and information about support services for families with disabled children is vested in social workers. Counselors’ lack of knowledge about the social aspects of disability becomes an implicit bias, however, since it limits the resources available to a pregnant woman and her supporters, once a positive diagnosis of fetal disability has been made and she must decide to end or continue her pregnancy.
Rothman (1986) argues forcefully that all prenatal diagnoses seem ambiguous to pregnant women.

Folk models for children with Down’s syndrome—highly stereotyped and badly out of date—surely exist for most people. And some Afro-Americans have opinions and images about the consequences of sickle cell anemia. But virtually no one I have observed or interviewed knew about neural tube defects (e.g., spina bifida), for which the test is also being done, or about sex chromosome anomalies. Among the 40 women with whom I have spoken who subsequently received positive diagnoses of fetal disability, only two ever felt competent, in hindsight, to predict the health status of the fetuses they were carrying.

These numbers are drawn from the counselors and medical centers in which I did direct observations, and they vary enormously from facility to facility. A small percentage of “no shows” have miscarriages before they are scheduled to speak with the counselor, but most have opted out of the appointment. In some clinics patients feel well-served and can communicate directly with a nurse or paramedic about their desire to see or not see the genetic counselor. In others suspicion, anger, or resignation dominate the experience of medical care, and appointments are coercively assigned and often broken in resistance. Those who choose not to have the test after counseling include women who discover that it cannot diagnose the problem they came to discuss, as well as those deemed “appropriate candidates” for amniocentesis but who don’t want it. Their reasons may include religious and spiritual beliefs; conflict with other family or community members about the meaning of pregnancy, testing, and having babies; fear of the test, especially fear of needles among Hispanic women; and the possibility of miscarriage.

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