MEDICAL HUMANITIES REPORT

Routine Prenatal Genetic Screening in a Public Clinic: Informed Choice or Moral Imperative?

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INTRODUCTION

Recently the great strides that have been made in our understanding of the human genome have been accompanied by soaring expectations about the potential health benefits of such information. Media accounts about each new morsel of genetic knowledge are often reported in reverent, almost science-fiction-like tones, intimating that the conquest of most human disease and suffering is only a few steps away. These high hopes notwithstanding, at present the clinical application of genetic testing for most conditions is primarily limited to risk modeling: that is, identifying an increased statistical likelihood that an individual with a particular genetic profile may develop a given condition.

To date, prenatal diagnosis is one of the few areas where this increasing genetic knowledge has moved into the mainstream of health care, and is having a tangible impact on a wide cross-section of the population. Improved techniques for screening for Down Syndrome, Neural Tube Defects, and Trisomy 18 make it possible to detect most occurrences of these conditions in the second trimester of a pregnancy. The maternal serum triple-marker blood test, or "Triple Screen", is now a routine part of prenatal care in the United States. It must be emphasized that this is only a screening test, and as such only has the power to indicate an "increased risk" that a fetus is affected by the condition in question. Further testing is required in order to determine the diagnosis. It is this condition, of being classified as having an "at-risk" pregnancy, which will be examined in this paper.

Kenen (1996) has argued that being classified as having an "at-risk" health status includes a set of expected role performances and norms. In the case of prenatal genetic screening, these roles would consist of accepting what Kenen has termed a "diagnostic invitation" and the "gift of knowing."

Because there are no effective therapies for the conditions identified through routine prenatal genetic screening, the benefits of this "gift of knowing" are limited. In fact, the only prevention for these conditions, once identified in a developing fetus, is termination of the pregnancy (Markel 1997). While providing women with such knowledge about their pregnancies does indeed have the potential benefits of informing their reproductive decisions or allowing them to prepare for delivering and caring for an affected child, knowledge is not always power. Having this information may be highly anxiety provoking, confronting the woman with morally fraught issues such as the acceptability of abortion, the value of a disabled child and whether mothering such a child should be viewed as discretionary.

Due to the danger that further testing may inadvertently cause a spontaneous abortion of a healthy fetus, coupled with the ambivalence that surrounds elective abortion in this country, the

current ethos of clinical practice calls for the invitation for further testing to be extended in morally neutral terms. Despite this intended neutrality, the very act of offering prenatal diagnosis intrinsically puts forth that possession of this knowledge will be beneficial and empowering (cf. Browner et al. 1999). It is our contention that, for a woman who has not sought out this information, it may be a distinctly disempowering experience to unexpectedly and abruptly find her pregnancy has been classified as "at-risk," based on a routine blood test.

We conducted a study of a group of low income Latinas in Texas who, through the course of routine prenatal care, had received abnormal blood screening results for birth defects. Our analysis of their experiences challenges the assumption that they perceive the offer of further testing in morally neutral terms. In this paper we show that having a pregnancy classified as "at-risk" was experienced by many of these women as a moral imperative to take action "for the good of the baby." Many accepted further testing not so much because they valued the knowledge it would produce, but simply because they felt compelled to remove the pregnancy from the quasi-disease state of being classified as "at-risk". We will argue that, rather than empower them, the unsolicited knowledge provided by prenatal genetic screening carried a moral mandate for them to accept further testing in order to restore their pregnancies to a normal, healthful status.

BACKGROUND

Currently in the United States, women determined to be "at-risk" for Down Syndrome, Neural Tube Defect, or Trisomy 18 based on either their age, prior family history, or an abnormal Triple Screen test result, are routinely offered amniocentesis. Amniocentesis is performed by withdrawing fluid from the amniotic sac with a needle, and then culturing the cells to determine the presence of these genetic and chromosomal defects. While this is a highly accurate test, it bears a number of important risks of its own, most notably that it may cause a miscarriage in about 1 in 200 women.

The protocol of using Triple Screen blood tests followed by amniocentesis has proven capable of identifying most cases of these anomalies (NIH 1996). However, one notable disadvantage of the Triple Screen is that it has a very high false-positive rate. As many as 99% of the pregnancies classified as "at-risk" by the Triple Screen are in fact free from these anomalies. Still, in the eyes of the pregnant woman, once her pregnancy has been classified as "at-risk," she is confronted by the problem of a pregnancy with an ambiguous health status.

Prenatal genetic screening is unusual as a medical service, in that its value does not lie in managing or curing illness, but instead in simply producing information that generates difficult choices for the patient. Women determined to be "at-risk" are generally provided pertinent information in a "genetics counseling session," and then asked to choose whether or not they want the amniocentesis. The information needed to make an "informed" decision of this sort is of a complex technical nature, contrasting relative risks, and involving terms and ideas which are often not part of the everyday experience of the patient. This may be particularly true for low-income patients with limited formal education or limited English language skills.

From the health provider's perspective, the patient's decision process regarding accepting or declining amniocentesis is thought to be one of weighing the risk of miscarriage against the value they place on knowing the health status of the fetus. Our research suggests that the patient may not be considering so much these two issues, as she is responding to the prospect of going through the rest of her pregnancy with her baby remaining in an ambiguous state of health.

SETTING AND METHODS

The research was conducted in an outpatient Gyn/Genetics clinic of a large university teaching hospital in south Texas. Services there are provided mostly by medical students and residents, and are overseen by an attending physician. Patients are referred to Gyn/Genetics from a variety of public clinics throughout the city, when they have been determined to be "at-risk" due to their age, family history or an abnormal blood screening. It should be noted that because this blood screening is "routine" in prenatal care, although the women have most likely signed a consent form before being screened, quite commonly they are unaware that they have had such a test, and arrive at the Gyn/Genetics clinic with no clear idea of why they are being sent there. Due to the complications of scheduling and the difficulty of getting patients to return, they are encouraged to decide on the spot whether or not they want the amniocentesis, so that it can be performed preferably that same day.

We interviewed twenty-nine patients from this clinic. All were low-income, self-identified Latinas who had been referred to the clinic for an abnormal Triple Screen test and were subsequently offered amniocentesis. Interviews were conducted in Spanish or English, as the women preferred, and focused on reproductive history, current pregnancy experience, understandings about fetal abnormalities and testing procedures, how decisions about amniocentesis were made, and general experiences and satisfaction with the genetics consult.

FINDINGS

For most of the women in our study, finding that her pregnancy was considered to be "at-risk" came as a shock. Most had not been told by their referring clinic that the reason they were being sent to another clinic was the possibility of a birth defect. Many came to Gyn/Genetics thinking it was a routine Ob/Gyn visit, or having only been told that they had a "bad blood test."

Despite the notably vague way in which these referrals were often made, nearly all of the women understood from the outset that something might be wrong with their babies, and reported feeling very anxious and frightened at finding that out. They consistently described feeling fear and sadness, with many recounting a moment of shock followed by crying, prayer, and loss of sleep.

Interestingly, in their overall discussion of their experiences, the "at-risk" concept was transformed into a very personal and embodied sense of something actually being wrong, and needing to be set right. It should be noted that all of these interviews were done after the women had already received their amniocentesis results, and all but one had gotten a clean bill of health for her baby, so they already knew that this had been a "false alarm." Still, the language they used to describe how they had felt when they found out something *could be* wrong, indicates that many remembered this as finding out something was wrong. Comments like these were common: "I felt very sad that something was wrong with the baby;" "I was very alarmed, and thought the baby would probably come out with a defect;" and "The Triple Screen test came up that my baby was sick, I think they said he had Down Syndrome."

Thus, it seems that for many, an abnormal screening test produced an alarming sense that their baby was in danger. It seems the idea of being "at-risk" in itself was perceived as a disorder. Once they were told that their pregnancy was "at-risk" the women were offered the opportunity to have an amniocentesis to confirm whether or not there was in fact an abnormality.

In principle, the purpose of prenatal testing is to enable the woman to choose to either terminate the pregnancy, or prepare for the birth of an affected child. If this were the case, one would expect to find that those who would not consider an abortion, or who were prepared to care for any child they may have, would be unwilling to take the risk of an amniocentesis. However, like previous research in this area (Browner et al. 1999) we did not find this to be true.

Most of the women in our study who *did* accept the amniocentesis said that they would not consider an abortion for themselves, and would want the child regardless of the outcome of the test. Only about a fourth of them said they wanted to be prepared for such a birth, and only three individuals said they accepted the amniocentesis so they might choose to terminate an affected pregnancy. Clearly, most of these women cannot be presumed to be motivated by the supposed goals of informing termination decisions, or preparing for the birth of an affected child. What then might better explain their decision to accept a test that could potentially harm their baby?

Nearly all said that until receiving the blood screening result they had felt their pregnancy was normal and healthy, but that being told their pregnancy was "at-risk" made them very anxious, leaving them with a sense that their baby was in danger. Once this sense of disruption was introduced, accepting the amniocentesis became the only way they could know that the baby was O.K., and return to a sense that they were having a normal pregnancy. As one woman explained it, "It was the most effective way to stop being as frightened as I was, to have something more concrete, an answer." Another commented: "I said to myself, if I don't do this, I'll have this doubt over me the whole time I'm waiting for the birth, and that would be a heavy burden, really, that's why I did it."

The women who accepted amniocentesis were almost unanimous in saying that they wanted to know for sure what was going on, to get rid of the worry and to be reassured of the health of their baby. In responding to the sense of malady that the "at-risk" status had engendered, many viewed accepting the amniocentesis as a way of promoting the welfare of the baby. All but one who had accepted amniocentesis expressed the idea that it was in her baby's best interest to do so. Comments like the following were common: "I was doing it for the good of the baby;" "The doctor said it was good for me and good for the baby;" "I did it for my own sake, and for the baby's sake."

During the time they waited for the amniocentesis results, which took a minimum of two weeks and sometimes longer, most of the women continued to feel anxious. But at the end of the whole experience, once they had received the results, they all said they were no longer anxious, and felt a return to a sense of well-being about their pregnancy. Interestingly, the one woman who had received a positive diagnosis of Down Syndrome also reported feeling relief once the result was confirmed.

It would seem that for all of these women, the pathology of the "at-risk" health status was effectively dispelled by proceeding with the amniocentesis. The disruption engendered by the abnormal blood test was resolved once the pregnancy moved out of a state of ambiguity.

DISCUSSION

In considering the meaning of being classified as "at-risk," Gifford has argued that, for the patient, the ambiguity of an "at-risk" health status "results in the creation of a new state of being... that is somewhere between health and disease" (Gifford 1986, 215). The women in our study did indeed seem to find themselves in such a "gray area." It should be noted that these women had not sought out diagnostic information about their pregnancies, but instead were suddenly confronted with this gray area in the course of routine prenatal care. For most of them, finding out they were "at-risk" had an iatrogenic effect, transforming what for them had been a normal, healthy pregnancy, into an quasi-disease state which required action to reinstate a sense of well-being.

One is led to wonder if the patient's "right to know" is counterbalanced by a "right not to know" in this nascent field of prenatal genetic diagnosis (Press and Browner 1995). Does the notion of "informed consent" have any salience at all for women who are abruptly confronted by an unsolicited choice about accepting a test, when that test presents the only avenue for resolving

this imposed ambiguity? The common clinical assumptions about how patients make prenatal testing decisions may be inaccurate for many women. We suspect this may be particularly true for those who have not sought out this information and have limited knowledge about the disorders in question and the test being offered. For these women, accepting further testing may be less based on a desire for more information about the health status of their baby, than on a sense that they are morally obligated to do whatever is necessary to remove an "at-risk" status from their pregnancy.

NOTE

An earlier version of this paper was presented at the Annual Meeting of the American Anthropological Association. San Francisco, CA. November 2000.

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InkLinks *is a regular column in which readers reflect on issues related to the lead article. This month, three readers respond to the lead article about the darker face of prenatal screening.*

A New Mother: As if Screening Could Make the Womb a Safer Place

Alice Dreger
Lyman Briggs School

When I was pregnant with Kepler, I decided not to do a sonogram. Counterintuitive as it seems, apparently sonograms do not improve fetal or maternal outcomes in medically uneventful pregnancies. I was having a medically uneventful pregnancy. Our midwife, Meta Bray, was fine with this. But one day, as I was checking out of the OB office where Meta worked at the time, the receptionist who was scheduling my next appointment told me I should also schedule my sonogram. I told her we had decided not to do one. Her response was a look of shock and the declaration, "I'll have to talk to Meta about that!"

Many people, medical and otherwise, had a similar reaction: that it shouldn't really be my choice, or maybe that I was a crazy luddite. People kept demanding, "But isn't it better to be safe?", as if the sonogram would make the womb a safer place for Kepler. Ruth Hubbard, in her recent visit to MSU, reminded us that besides considering the risks and benefits of technologies we should also think about the burdens. Natalie Angier and others have shown the burdens of sonograms, especially on the developing mother-child relationship, are substantial. But unquantifiable, they are often ignored.

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A Childbirth Educator: Doesn't Anguish Count as an Outcome?

Libby Bogdan-Lovis
Center for Ethics and Humanities in the Life Sciences

In the past, I worked in multiple capacities towards educating women about different facets of the childbearing year (pregnancy, labor, delivery, postpartum). During this tenure I was education coordinator at a clinic, and it was at this time that the AFP neural tube defect screening test was introduced through a spirited marketing campaign. At a clinic inservice, an enthusiastic diagnostic company rep described the simple new blood screen that provided impressively rapid results. Carefully relegated to the end of the inservice was information on the high rate of false positives. A positive reading would then necessarily need to be followed up with another test, a very accurate (but more invasive) amniocentesis. We were reminded that a woman would be well into the second trimester of pregnancy when the results of the amniocentesis would be available.

I later had my first clinical encounter with a woman receiving news of a positive AFP screen. As a consequence of that test result, the woman faced a dilemma presented by the recommended follow up amniocentesis. Abortion was not an option for her. Moreover, she felt certain that the test was wrong and that her baby was unaffected by the list of neural tube defect possibilities presented in the literature. Her partner, the father of the baby, felt equally certain of the veracity of the test results, and he desperately wanted her to undergo amniocentesis to confirm his worst fears. Armed with this evidence, he hoped that he might then successfully convince her to end the ill-fated pregnancy. This couple's shared anguish lingered until she later gave birth to a healthy baby.

Their personal agony caused me to ponder the experiential content missing from that original AFP inservice. How might that enthusiastic marketing rep have conveyed the depth of human suffering associated with the simple, rapid result, AFP screen? Was this couple's experience located somewhere in those emotionally sterile charts and graphs presented? The introduction of additional testing is commonly viewed as an unqualified good. If it can be known, it should be known. Isn't it better to know more?

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A Doctor: The Obligation to Have a Conversation, and to Listen

Barbara Supanich
CHM Clinical Faculty, Traverse City

As a family physician and a family practice residency educator, I have discussions with my patients or with residents on a daily basis regarding the indications or need for screening labs, x-rays, or ultrasounds.

Women come into a physician's office with a variety of expectations regarding screening tests. Some come with a very clear understanding of the pros and cons of a test and expect to have a conversation, which clarifies their current understanding. Some come with very little knowledge about the test(s) and want to know more, and some come asking for a test because it was recommended by a friend or family member, but they do not know anything about the test or the pros and cons related to the test.

Physicians and others involved in providing health education to patients have an obligation to engage in ethical decision making with our patients and/or family members. In particular, women who are receiving prenatal care will have a variety of prenatal screening tests offered to them. I think that *how* conversations occur between the woman and her clinician is of critical importance.

First, although as physicians, we are obligated to inform pregnant women of the availability of various prenatal screening tests (chorionic villi sampling, the triple test, amniocentesis, etc.), we have not completed our ethical obligations by merely informing her that the tests exist.

All tests can cause harm as well as benefits for the patient. Women need to know what the balance of the benefit/harm ratio is for her in the particular setting of her current pregnancy. For instance, if she is pregnant for the first time, is less than 30 years of age, and has a very healthy family history and personal health history, her chances of having a pregnancy with a complication including a significant birth defect are very small. In this setting, it may cause her more harm (false positives) by having a screening triple test rather than declining it.

It is important for the woman to know what the nature of the test is and who will benefit most from having the test performed. In the example above, she would have more of a chance of having a false positive result. This would then entail either obtaining a level 2 or 3 ultrasound or an amniocentesis depending upon the exact results of the triple test. Both situations would minimally cause significant emotional stress for the woman and her family and if she had an amnio, she would be exposed to other risks related to the amnio procedure, including a 1:100 to

1:200 chance of losing the pregnancy.

Given this context, it is important to remember that screening tests are designed to identify those at highest risk for a disease or condition. As physicians or clinicians, we are obligated to have a conversation with our low-risk patients regarding the nature of the screening test and the likelihood of risks and harms vs. benefits for her and her fetus. On balance, for most low-risk pregnant women, the risk of the test is the false positive result. This will then "obligate" most physicians and women to proceed with further testing which has even higher risks and harms associated with them.

Secondly, we as physicians and clinicians need to hear why the woman wants to have the test performed. Her story, her personal understandings need to be heard by the professional providing her care. Some women have had many friends whose pregnancies were complicated by genetic illnesses; for their own peace of mind, they want to have the screening test(s) performed. Other women want to decline the test(s) because if they knew that they had a fetus with a significant illness ahead of time, it would cause such emotional distress that they would have emotional difficulties throughout the remainder of the pregnancy. They would opt to not know now and would rather "deal with the situation at the time of birth." Still others would want to have the test(s) performed because if a significant genetic illness were diagnosed, they would opt for abortion.

In summary, it is my ethical obligation to have a conversation with my patients who are pregnant regarding appropriate screening tests. On my part, I need to listen carefully and respectfully to the request of the woman and her reasons for the request or denial for the testing. It is my obligation to be sure that she has the information she needs to make an informed decision. Part of this discussion must include information regarding the consequences of beginning the screening program.

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Center News and Announcements

At the 2000 American Anthropological Association meetings in San Francisco, **Libby Bogdan-Lovis** presented "The Evolution of Evidence-based Medicine: A Critical Analysis."

Howard Brody will be taking a six-month sabbatical between January and June, 2001, when he will be serving as a visiting professor at the Center for Ideas and Society, University of California-Riverside.

Judith Andre spoke on "Ethical Issues in the Use of Placebos," at Grand Rounds, Providence Hospital, Southfield, MI.

Tom Tomlinson has been appointed to the Patient Safety Subcommittee of the Michigan Hospital Association. The subcommittee is evaluating proposals for reducing adverse events in Michigan hospitals.

Len Fleck presented "Genetics, Reproduction, and Public Policy: Emerging Ethical and Political Challenges," at Washtenaw Community College in Ypsilanti, MI, November 2000.

Libby Bogdan-Lovis presented "Evidence-based Medicine, Childbirth, and Morality" to the College of Human Medicine Medical Scholars, as part of their fall thematic program.

Howard Brody served as a visiting professor and consultant to the National Center for Bioethics in Research and Health Care, Tuskegee University, Alabama, January 22-26, 2001.

Clayton Thomason spoke on "Spiritual Needs and Pastoral Care of Physicians in Training: Workshop for Pastoral Care Providers and Medical Faculty," at Kettering Hospital, Dayton, OH, December 16, 2000.