Prenatal diagnosis of chromosomal abnormalities, neural tube defects, and specific genetic disorders is becoming increasingly available to pregnant women in this country. As the Human Genome Project yields more information about the genes responsible for illnesses, medical conditions that develop later in life, and basic human differences, and as medical science develops safer and simpler techniques for obtaining genetic material, the spectrum of conditions for which testing will be available will expand dramatically. Decisions about the appropriate use of these new tests and technologies will be challenging. This will especially be true when decisions about genetic testing intertwine with decisions made by women about reproduction, as occurs in the use of prenatal genetic testing.

In the general practice of medicine, it is important to consider not only therapeutic results, but also the nonclinical implications of what is done to or for patients. This is particularly imperative when one considers prenatal genetic screening and testing and its application to broad populations of women. A few basic questions need to be asked: Do we know whether prenatal genetic testing is a good thing for women, children, or society? Does prenatal genetic testing go beyond merely providing more information about a woman's pregnancy to actually improving her health? The term health is used broadly in this context, as
described by *Dorland's Illustrated Medical Dictionary* (1988) as a "state of optimal physical, mental, and social well-being, not merely as the absence of disease and infirmity."

While it is clear that prenatal genetic testing is very useful for identifying a particular chromosomal or genetic abnormality in a fetus, and that it can offer meaningful choices to families at risk for specific genetic disorders, less evidence exists to indicate whether the widespread application of testing to pregnant women will succeed in achieving the broader goal of improving the health and well-being of obstetric patients. Indeed, several goals can be postulated for a broadly applied program of prenatal genetic testing.

**Is Prenatal Diagnosis Intended to Benefit Society?**

The notion that individual women should be offered or encouraged to undergo prenatal genetic testing in order to spare society the expense of coping with diseased or disabled offspring has been put forward (Shaw, 1984). Viewed in terms of medical economics, the burden of genetic diseases on society is significant. "Genetic disorders account for about 20 percent of pediatric hospital admissions and for an even higher percentage of long term admissions" (Simpson, 1986). Widespread application of prenatal genetic testing, if accompanied by treatment or by termination of pregnancy, would decrease the social burden of genetic disease, at least in economic terms. Currently, however, most diagnosable conditions are not treatable prenatally. Furthermore, access to abortion, even for diagnosed genetic conditions, may be limited, either legally, economically, or by inadequate availability of abortion services in certain areas of the country.

Even if prenatal diagnostic testing could decrease the social burden of genetic disease by preventing the birth of affected individuals, it is not clear that our society is ready to promote prenatal genetic testing in conjunction with abortion primarily for this end. In statistical terms, the selective abortion of fetuses affected with genetically determined conditions would in fact increase the prevalence of abnormal genes in the population if fetuses found to be carriers of recessive conditions were spared
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(Boss, 1990). Furthermore, it is unlikely that decreasing the rate of genetic disease would address the ills of society as effectively as other sorts of social measures such as improved access to health care or education. Even more important, the limitations to individual reproductive freedom required by a program of mandatory screening and termination of pregnancy would be intolerable.

Does Prenatal Genetic Testing Provide a Benefit to the Child?

Benefit to the expected child can be achieved in some situations. If the diagnosed condition is one treatable before birth, the future child should benefit. For example, there has been some success in treating alpha thalassemia and combined immunodeficiency syndromes by fetal bone marrow transplantation. If the condition diagnosed will impact obstetric or prenatal management, the future child may benefit. For example, if hemophilia is diagnosed in a fetus, scalp sampling and the use of a vacuum extractor to assist in delivery can be avoided for fear of injuring the child. However, when no treatment exists and pregnancy termination is the only alternative to doing nothing, there may be no benefit to the child. Whether being spared a life with disease, pain, or disability is a benefit to the child is a difficult ethical question on which a spectrum of articulate opinions exists (Botkin, 1988; Elias & Annas, 1987; Liu, 1987).

Is Prenatal Genetic Testing Intended to Benefit Women?

When one studies the impact of a medical intervention on the health or the quality of life of patients, the approach is typically to assess the relative burdens and benefits of the intervention in question. Another consideration is whether and in what ways prenatal genetic testing is intended to benefit the parents (or more specifically, the mother) of the future child. Prenatal diagnosis may in some ways enhance the quality of life for women, both during and after pregnancy. It may be useful in allowing a woman to have the choice of whether to bear a child
with a genetic or other congenital disorder. Further, it may pro-
vide the pregnant woman with needed information about the
medical condition of her fetus. However, there may also be risks
involved such as an increased anxiety, leading to a reduction in
her quality of life. The practice of prenatal diagnosis can be ex-
pected to impact women in a number of ways, and it is not
absolutely apparent that all of these impacts are positive. In fact,
what is construed as a benefit for one woman may on the con-
trary be burdensome for another. Experiences will vary among
women depending upon their individual backgrounds, experi-
ences, and values.

Although the published data on the scientific aspects of pre-
natal diagnosis are extensive, significantly less data are available
to demonstrate whether prenatal testing is truly beneficial or
harmful to women’s health in a broad sense. The majority of
the data currently available relate to the prenatal diagnosis of
chromosomal abnormalities and neural tube defects. Much less
work has been done thus far on the impact of prenatal testing
for specific genetic disorders. Even so, some interesting trends
are apparent, and some preliminary questions about the less
tangible effects of widespread prenatal genetic testing can be
posed.

Is Prenatal Genetic Testing Reassuring?

One possible benefit of prenatal genetic testing might be to re-
assure a pregnant woman that the child she expects will be nor-
mal. This benefit has been widely promoted by advocates of
testing. Consider, however, what role the availability of testing
itself may have had in creating or sustaining the need for testing
and reassurance. Are healthy women, entering the natural stage
of pregnancy, being labeled “at risk” as a result of the develop-
ment of tests for rare but potentially disabling genetic disorders?

There is evidence that prenatal testing, at least chromosomal
and alpha-fetoprotein (AFP) testing, is reassuring to most
women (Finley, Varner, Vinson, & Finley, 1977; Dixson et al.,
1981; Robinson et al., 1988). There are, however, also indi-
cations that testing can raise anxiety prior to the procedure,
reducing it if the results are normal (Beeson & Golbus, 1979).
The aspect of testing found to be most distressing has been the need to wait for results (Finley et al., 1977; Dixson et al., 1981; Evers-Kiebooms, Swerts, & Van den Berghe, 1988). One study found that a group of women who received an abnormally low result on AFP screening were noted to be significantly more worried about their baby's health three weeks after testing (after normal follow-up results were available) than were women who received normal results initially. Those differences were not appreciable at later points in the pregnancy (Marteau et al., 1992). According to other studies, it is not clear whether women who have undergone prenatal testing are left with less anxiety than those untested, once normal results are received (Tabor & Jonsson, 1987; Phipps & Zinn, 1986).

Prenatal diagnosis use may, in the end, raise more concerns about the pregnancy than it can answer. In deciding whether to have testing done, a woman must face the possibility of an untoward outcome when, in fact, her chances of having a normal infant are very high. Dixson followed women who received genetic counseling and then either underwent amniocentesis or declined. The rate of continuing concern about possible congenital abnormalities was just over 20% in both the group who received normal amniocentesis results and the group who declined testing (1981). Evers-Kiebooms (1988) found that one third of women tested felt reassured only after their child had been born. Although some women will experience residual anxiety, others will show more positive responses than had they not received reassurance from the test results.

The issue of reassurance can be examined in the converse. Can the information that is obtained through prenatal diagnosis be falsely reassuring? Even if the specific diagnosis being sought, such as for muscular dystrophy, is ruled out, there remains a two to three percent chance that the child will be born with some other kind of congenital disorder or genetic disease. Will women who undergo prenatal genetic testing be more or less prepared for other congenital problems that may arise? With the availability of testing, are women more inclined to focus on what may go wrong in a pregnancy rather than on the fact that things usually go right? While prenatal genetic testing may be valuable and reassuring for that group of women in...
whom significant risk for a specific abnormality exists, more data is needed before concluding that testing is on the whole reassuring to women.

**Does Prenatal Genetic Testing Enhance the Autonomy of Pregnant Women?**

Eric Cassell (1977), a physician and medical ethicist, has stated that "the primary function of medicine is to preserve, to repair, and to restore the patient's autonomy." While prenatal genetic testing does provide options to women at risk for bearing children with genetic diseases, it is not clear from existing data about reproductive choice that patient autonomy is actually enhanced through testing.

There is evidence that testing is, for some women, associated with a sense that control over important reproductive decisions is, to a degree, being usurped by family, by society, and by the medical profession. Berit Sjogren (1988) used a combination of questionnaires and interviews to assess whether women undergoing amniocentesis or chorionic villi sampling (CVS) made autonomous decisions regarding their testing. Most of the women surveyed (75%) found it difficult to refrain from prenatal diagnosis when it was offered. Seventy-eight percent felt it would be more difficult to give birth to a disabled child if they had not accepted prenatal diagnosis. Many stated that while they felt "free from external pressure," they still felt an "obligation" to have testing performed. Among women of a certain age, willingness to undergo prenatal diagnosis seems to be construed as a sign of responsible parenting. Is a woman acting irresponsibly in the eyes of others if prenatal diagnosis is foregone? Might a woman sense less sympathy and support if testing is foregone or if a pregnancy is continued in the face of adverse results? If perceptions such as these are common among pregnant women, the availability of prenatal diagnosis may indeed limit autonomy rather than enhance it.

To the extent that prenatal genetic testing actually provides women with the choice of whether to bear children with specific abnormalities, it does enhance their autonomy. If a specific disorder is diagnosable, it can be ruled out in a fetus. However, the woman may then be left with unrealistic ideas as to the
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chance of a different sort of disorder existing in her child. Will her ability to make responsible choices be limited by unrealistic expectations? How well will she be able to adjust if her child is affected with a disorder for which testing was not done? These questions remain unanswered.

The availability of prenatal genetic testing has brought the interests and influence of family, friends, and society into an individual woman's pregnancy to an increasing degree. Wertz (1992) found, in asking couples who had children affected with cystic fibrosis (CF) about prenatal testing for that condition, that "respondents' perceptions of their siblings' approval of abortion for CF was the best predictor of use of prenatal diagnosis." The opinions of the spouse and members of the extended family were also influential. Prenatal genetic testing appears to have opened the door to third-party involvement in the most private of decisions being made by women during pregnancy. Whether a woman chooses one form of childbirth preparation over another is an important decision but not fundamentally a moral one. Whether she has prenatal genetic testing and, further, what she decides to do with the information she receives may be one of the most serious moral decisions she will ever make. Involvement by parties such as employers and insurance companies in an individual woman's decisions about prenatal genetic testing will restrict reproductive choice in an unprecedented way (Gates, 1990).

In one illustrative case, a pregnant woman whose living child was affected with cystic fibrosis sought prenatal testing for the disease. When testing revealed that the expected child would also be affected with CF, the woman and her husband faced an agonizing decision. They ultimately decided not to abort the fetus. Their insurance company, an HMO, had agreed to extend special coverage for this woman's prenatal testing. It decided, however, that it would not provide medical coverage for an affected child, a child who would be born with a "preexisting condition" on account of the prenatal diagnostic procedure. As one journalist wrote, "The insurance company's message was clear: The parents could either abort the defective baby or struggle alone with the financial burden of a sick child" (Thompson, 1989). Although the HMO ultimately capitulated and agreed to cover the child's treatment, their initial policy
serves as a sign of the new limits on maternal choice, which may evolve as the capacity for prenatal diagnosis expands. In the setting of widely available prenatal genetic testing, reproductive choices may become as much social choices as personal ones. It will be important to assess the impact of such testing on women's sense of their ability to retain control over the choices made about their pregnancies.

Is the Impact of Prenatal Genetic Testing Influenced by Issues Concerning Access to Family Planning Services?

At the same time that state legislatures around the country are planning and establishing programs to enable women through genetic screening and prenatal diagnosis to avoid conceiving or bearing affected children, a trend toward restricting a woman's right to decide to abort an affected fetus is evident. It seems likely, in view of recent Supreme Court decisions, that individual state legislatures will have increasing power to define the circumstances under which abortion will be legally available.1 If access to abortion is significantly restricted, one really must ask just what kind of choices prenatal genetic testing offers. How much benefit is knowledge in the absence of meaningful options?

Although many are currently optimistic that abortion will remain a legal option in this country, women are still faced with significant limitations on the accessibility of abortion services both in financial terms and in terms of the number and geographical distribution of facilities offering abortion procedures. Women facing the termination of a desired pregnancy based on an abnormal result from prenatal testing may have even more difficulty because they must locate facilities that offer termination of second-trimester pregnancies.

Are the Choices Offered by Prenatal Genetic Testing Burdens in Themselves?

It is likely that the decisions required by the availability of prenatal genetic testing may be perceived as burdensome by some women. As Rayna Rapp (1987) has framed the question,
"Does amniocentesis offer women a 'window of control', or an anxiety-provoking responsibility?" Certainly, freedom and responsibility go hand in hand. Many women are able to make an informed decision to accept the burdens associated with testing in order to obtain the knowledge they seek. If testing is offered routinely and on a widespread basis, however, an informed and thoughtful decision may not be encouraged, and the burdens of testing may be both unexpected and troublesome.

If the test results indicate an abnormality, the woman must decide how to proceed—what to do with the information she has received. In Sjogren's study in Stockholm (1988), half the women surveyed stated that when they received their test result they had not yet reached a decision about what to do in the case of a fetal abnormality. Dixson (1981) found that one-third of the women surveyed remained uncertain at the time results were received. Adding to the complexity of the issue, at times the abnormality diagnosed prenatally, whether by chromosomal analysis, ultrasound, or specific genetic testing, is one for which the implications and prognosis are unclear. Thus, if a woman chooses to continue her pregnancy, anxiety about the outcome may be heightened; and if she elects termination of the pregnancy, she may wonder whether her decision was indeed the "right" one. In a group of patients surveyed by Drugan (1990), 93% of patients whose fetuses were diagnosed with a disorder bearing a severe prognosis elected to terminate their pregnancy. Twenty-seven percent of those with a questionable prognosis made this decision.

What Is the Impact of Prenatal Genetic Testing on a Woman's Relationships with Family and with Her Future Child?

The decisions a woman is required to make regarding prenatal genetic testing can be very stressful ones for her and for her partner. For many couples, this stress comes at a time when their relationship is already being redefined by the expectation of a child. The woman's partner may want prenatal diagnosis more than she does, and may be less anxious about it (Evans et al., 1988; Keenan, Basso, Goldkrand, & Butler, 1991), potentially leading the woman to experience a sense that "he'll blame
me if the child has a diagnosable abnormality that we don't find out about,” or “he’ll blame me if we have an amniocentesis complication.” If the fetus is affected by a genetic disorder, the woman and her partner may also feel different about the prospect of abortion. Those differences may impact a couple’s relationship far into the future, regardless of the decision they ultimately make. The disagreement may furthermore affect their relationship with the child, if the pregnancy is continued. The integrity of these relationships is clearly important to women’s overall health. Little work has been done in terms of delineating the effects of decisions about prenatal diagnosis on a woman’s family relationships. It will be important to clarify such ramifications before widespread testing is initiated.

Taken as a whole, investigation of amniocentesis’s impact suggests that anticipation of the procedure and its results may lead to disruption of an otherwise normal adjustment to pregnancy. Studies have revealed that women undergoing amniocentesis may experience a “suspension of commitment to pregnancy” while awaiting test results (Beeson & Golbus, 1979; Spencer & Cox, 1988). Beeson and Golbus (1979) observed that this was reflected both in social spheres, such as in not telling others about the pregnancy, and in personal domains, such as in avoiding thinking about the pregnancy. Rothman (1986) noted a greater frequency of women not feeling movement until after the 18th week among those undergoing amniocentesis. In contrast, Dixson (1981) could demonstrate no significant differences between amniocentesis and nonamniocentesis groups in the timing of selecting names or in willingness to talk about the pregnancy. Furthermore, Phipps and Zinn (1986) found that amniocentesis patients in a United States sample showed a greater increase in fetal attachment over the course of pregnancy than did nontested controls. Caccia (1991) demonstrated that maternal-fetal attachment increased significantly for amniocentesis and CVS patients once normal results were received. No nontested controls were included. It has also been noted that attachment may be enhanced by viewing the fetus on ultrasound, a technology used in conjunction with amniocentesis (Fletcher, 1983). If suspension of commitment does occur for some women during the early months of pregnancy and if it
involves decreased compliance with suggested regimens such as good nutrition and abstaining from alcohol use, long-term adverse effects might result. Further investigation could clarify which women are at risk for this type of response to prenatal testing and could lead to meaningful interventions.

Jeffrey Botkin (1990) states that "a fundamental aspect of parenting is the recognition of the unique and independent nature of our children's personalities and lives. Try as we might, they rarely fit the molds that we design for them. A knowledge that our intrinsic personal characteristics were the intentional product of our parents' designs would have a profound influence on the parent-child relationship." Abby Lippman (1991) points out that "prenatal diagnosis does approach children as consumer objects subject to quality control." Those observations will be important to consider as the use of prenatal genetic testing expands. It will be important to clarify our understanding of women's expectations in relation to their children, as they depart from the illusion that they actually have some control over the "quality" of the infants they bear.

**What Influence Does the Health Care Provider Have over the Effects of Prenatal Genetic Testing on Women?**

The physician who counsels a woman about testing or about her response to abnormal results will have an important impact on her decision. Physicians may express, explicitly or implicitly, certain expectations about a woman's response to an adverse diagnosis. A woman, valuing her relationship with her physician and being in a relatively less powerful position in the physician-patient relationship, may hesitate to go against her physician's recommendations. If prenatal genetic testing becomes widespread, it will be essential that obstetricians become skilled in counseling women as they confront stressful decisions. A balance must be developed between supporting a woman through a difficult decision and limiting her ability to make her own choice. Holmes-Seidel (1987) reported an increased tendency for women to terminate a pregnancy affected by a sex chromosome abnormality when counseled by a general
obstetrician rather than a geneticist. It is not clear how expert general obstetricians currently are at counseling women about genetic problems, nor is it clear that a balanced view about the disorders is presented by providers who have little or no personal experience with individuals who have disabilities.

When prenatal diagnostic testing is provided for a woman who is carrying a fetus at increased risk for a particular genetic condition, pretest and posttest genetic counseling are essential. The importance of the nonclinical implications of test results, and the acknowledgment that an individual woman's personal values are of primary importance are two considerations that underlie the concept of nondirective genetic counseling as practiced by most geneticists and genetics counselors. Currently, good pretest counseling consists of the provision of accurate information about the testing techniques involved and about the conditions being studied (Hsia & Hirschhorn, 1979). This information needs to be offered by an individual who has not only a solid knowledge of the science of genetics but also a familiarity with the particular condition being discussed. Once the information has been provided, the couple then goes on to make a decision about whether to have testing done and about which technique they would prefer. Such standards for genetic counseling have been developed with good reason: to ensure the benefits of genetic testing and, more important, to protect the individuals being tested from harm.

To date, a counselor has been able to focus on one or two conditions in counseling. A couple may, for example, want CVS to rule out chromosomal aneuploidy in addition, perhaps, to testing for a particular condition for which their fetus is at increased risk, such as muscular dystrophy. In the future, what will be offered may be a battery of tests for a spectrum of conditions of variable severity and for which an individual fetus or couple may not be at increased risk. The counseling challenge will be magnified. With broader application of prenatal genetic testing, the capacity of the current system for genetic counseling will be overwhelmed. A recent survey reveals that as many as 39% of the genetics counseling job openings in this country this year will probably not be filled because the number of
individuals graduating from training programs is inadequate (Meyer, Edwards, Young, & Brooks, 1992). As a result, more and more of the burden of patient education and genetic counseling will fall to general obstetric providers.

It is worth considering how the genetic counseling model might be translated into a busy general obstetrics and gynecology practice where newly pregnant women and women planning a pregnancy are seen. The provider, whether obstetrician-gynecologist, midwife, or nurse-practitioner, is typically faced with a large volume of patients in order to satisfy patient demand and meet overhead expenses. Time constraints are a problem. Most important, the patients have many physical and psychological needs to be met. Providing information about tests for genetic conditions for which patients may not be at increased risk will probably not be a top priority in most situations, except perhaps when a woman comes specifically for fertility consultation, preconception counseling, or for an initial patient visit.

There is evidence that women's health care providers generally are not as skilled as trained genetic counselors, either in genetic knowledge or in the technique of counseling. For example, a survey of residents in obstetrics and gynecology in the Philadelphia area indicates that while most residents did have fairly good knowledge of teratology, those individuals scored, on average, between 54 and 65% correct on questions about clinical genetics and genetic testing (Kershner, Hammond, & Donnenfeld, 1992). If these results are representative, it is apparent that obstetrician-gynecologists will need to broaden their understanding of genetics before they will be able to inform patients adequately about the testing options and results available to them. One must also acknowledge that providers are under pressure to learn more about other important areas such as hormonal replacement in menopause, the evaluation and treatment of cervical intraepithelial lesions, and to improve their skill in the use of new technologies such as Norplant placement and endoscopic surgery. All of these areas are important to women's health, as much if not more so than is prenatal genetic testing.
Possible Approaches

In deciding which tests to start offering on a widespread basis and when, it will be important to keep in mind that testing should be performed with the intent of doing more than merely identifying randomly selected conditions in a limited number of women. Testing should be aimed at improving the health of women in the broadest sense. There is both a scientific and a moral obligation to demonstrate that prenatal genetic testing achieves this end effectively. Not only do studies on prenatal genetic diagnosis need to answer the scientific and practical questions (can the diagnosis be made accurately? can appropriate follow-up for results be provided?), they should also assess whether testing is beneficial to those who are meant to benefit, and whether testing results in significant harm to any parties.

Recent reports published about triple marker screening, for example, focus on the test's ability to identify fetuses with the targeted condition (Phillips et al., 1992; Haddow et al., 1992). As the use of this kind of screening is investigated further, and before such screening is offered on a widespread basis, attention needs to be devoted to assessing its impact on women in terms of their psychological adaptation to pregnancy, their sense of control over the outcome of their pregnancy, and their relationship with family. The impact of counseling on a woman's reactions to testing should also be studied further, including finding ways of enhancing a woman's sense of autonomy in making decisions about prenatal diagnosis, and the impact of prenatal "quality control" on a woman's relationship with her infant and child. Models like those used for quality of life assessment in clinical trials on disorders such as cancer and arthritis can be applied to trials of prenatal genetic diagnosis (Feeny & Torrance, 1989).

Providers of health care to women, whether physicians, midwives, or nurse-practitioners, will need to be educated more thoroughly about the principles of genetics, the nature of the conditions for which widespread testing is available, and the alternatives open to women who learn that their fetus is affected with a genetic condition. It is particularly important that those providers become skilled at counseling women in a manner
which enhances their understanding of the tests being offered while enabling them to make thoughtful and uncoerced decisions about the use of this new technology.

**Conclusion**

In summary, two points should be recognized. First, there is not yet clear evidence of the impact that prenatal genetic testing will have on pregnant women and on their overall health. It is not yet even clear whether the testing that is already in place, AFP screening and chromosomal analysis for women aged 35 and older, is beneficial to women's health. Second, it is likely, at least for the foreseeable future, that the counseling available in physicians' offices will not meet the standard set for genetic counseling, adopted in order to ensure the benefits of genetic testing and to protect individuals from harm.

The next decades will bring a continued expansion in genetic technology and in the use of genetic testing in many areas of health care, including obstetrics. Certainly, most of the questions posed here cannot be answered until a large number of women actually go through the processes of considering testing and of the testing itself. One of the challenges faced is to offer testing in its initial stages in an investigative manner that both protects the women involved and allows important questions to be addressed. Ideally, new tests would be offered only in those settings where adequate counseling, follow-up, and safeguards are in place, and where a conscientious effort is made to answer difficult questions about the impact of testing on patients and their families.

Currently, concerns about the risk to a fetus of diagnostic methods such as amniocentesis and chorionic villi sampling are considered an understandable reason for a woman's decision to decline prenatal genetic testing. Those concerns also underlie policies limiting testing to older women or women with a specific genetic risk. Soon, however, it will be possible to perform genetic testing on fetal cells obtained by withdrawing blood from a pregnant woman's arm, eliminating the most obvious physical risk factor. The claim may then be made that there is no good reason not to have testing done. Widespread testing
could potentially be initiated—and for all pregnant women. It is imperative both that solid evidence exists indicating that prenatal genetic testing is truly beneficial to the majority of women, and that effective mechanisms for reducing the burdens involved in testing are available before the routine testing of every pregnant woman becomes standard medical practice.

NOTES


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