

Part I

PLANNING FOR  
PREGNANCY



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# Chapter 1

## Planning for Pregnancy



SOMETHING AS NATURAL as being able to have a baby cannot be taken for granted. Planning ahead for childbirth is important. The body cannot be expected to perform on demand without preparation. In the same way that an athlete strengthens her body to run a grueling marathon with a plan of gradual physical training, a woman can ready her body so that pregnancy will be as comfortable and healthy as possible. Women are starting to recognize that planning ahead and preserving fertility are important. Older women facing a first pregnancy often ask us about the possible effects of a long-ago abortion or an infection. Concern and even fear are common.

Women are having children later in life. The number of births in general is going up, rising 3 percent in the year 2000. Fertility rates have increased by 1 to 2 percent, and there has been an increase in the number of twin pregnancies, reflecting the new reproductive technologies. There has been a rise in the birthrate among thirty- to forty-five-year-old women—women who were once thought to be beyond childbearing years. Births to women in their forties to early fifties were up again in the year 2000. The reason for this is twofold: Women are marrying later and waiting to have children until their educations are completed and their careers established; in addition, 37 million of the postwar babies, the so-called baby boomers, are now older women, whose sheer numbers add clout to any changes that they as a group decide upon. Also, affluence affords many older women access to the advances of reproductive technologies, giving them an opportunity to bear children later in life. In the past, this was not possible.

In these times of women having children later, it has become even more important for you to take care of yourself at an early age. Active participation in your health and well-being is important to maintain fertility later in your life. In our complex society, many factors may affect your future childbearing. They include environmental

hazards such as X rays; chemical pollutants in air, water, and food; smoking; and exposure to prescription and nonprescription drugs. Exposure to various illnesses such as rubella and toxoplasmosis can harm a developing fetus. By being informed, you can avoid many potential threats and minimize the impact of possible dangers.

The information in this chapter is intended to give you the best possible chance of having a healthy baby. We will discuss such diverse important areas as genetics, medical history, work environment, medications, eating, and exercises. Planning ahead for a healthy child will help you protect your fertility until the time is right for childbearing and will help you prepare your body and mind once you are ready for pregnancy. Planning ahead will help you get pregnant and help get your baby off to a healthy head start. Years ago not many women thought of having a baby after age thirty-five, but twenty years ago women didn't run twenty-six-mile marathons either. With the right knowledge, the right game plan, and the right care and training, both marathons and healthy pregnancies have become a reality for more women today than ever before.

## PRECONCEPTIONAL CARE

Before you become pregnant or stop using birth control, you can begin to maintain a healthy lifestyle, which will help you have a healthier baby and a healthier pregnancy. This includes eating right, exercising, and avoiding alcohol, cigarettes, and exposure to harmful drugs and chemicals. Early medical care and a checkup are also important. All this is part of preconceptional, or prepregnancy, care, ensuring your good health before you become pregnant. Many women do not know they are pregnant until five, six, or even eight weeks after they have conceived. Those early weeks may be some of the most important for the baby because during that time the organs form. Certain substances, such as alcohol, tobacco, chemicals, and some medications, can interfere with that growth. Likewise, medical conditions such as diabetes or high blood pressure should be under special supervision before a woman becomes pregnant.

You should discuss your plans for pregnancy with your doctor. This provides a chance for you to get advice on any questions or concerns you might have. A doctor may also offer suggestions based on your special needs. Women with diagnosed medical problems such as diabetes and anemia have an even greater need for accurate medical advice prior to pregnancy. Find out exactly what the risks are to you and your unborn child, what you can do to reduce them, and what choices you face for risks that can't be entirely predicted or controlled.

## Components of Preconceptional Care

The main components of preconceptional care are the same as those of prenatal care: assessing risk factors, promoting good health, and obtaining any necessary medical and psychosocial treatment. The anchor of all other preconceptional care activities is a comprehensive assessment of the risk you and your family carry for a poor pregnancy. You and your doctor can conduct such a comprehensive risk assessment during a special preconceptional visit or as part of a visit for other purposes. It consists of a complete history, a physical exam, and some laboratory tests. You and your physician will both review your health status, identifying any risks for a poor pregnancy outcome, and produce a plan to reduce those risks, revising it later if necessary.

In addition, you should undertake activities designed to promote good health in general. Table 1.1 lists the factors the Expert Panel on the Content of Prenatal Care identified as being important in evaluating preconceptional health. Table 1.2 lists the laboratory tests that may be valuable.

Your doctor evaluates preconceptional care and recommends tests based on their potential to improve not only your health but that of your future child as well. Some tests are of little value to you before pregnancy but are important after conception. (They will be discussed in chapter 7.) Others are valuable during the early first trimester of pregnancy, but since many women do not have prenatal care until the second trimester, those should be performed at the preconception visit. For example, such risks as susceptibility to rubella should be identified prior to pregnancy. Your physician can guide you as to which laboratory tests are important for you.

### Infectious Risks

When an infection occurs, for the most part, pregnant women have a normal immune system. While some infectious conditions may have only minor or no effects on a pregnant woman, the effects on the fetus can be devastating. Conversely, potentially serious infections may have little or no effect on the fetus.

### *HIV/AIDS*

Frequency of HIV—human immunodeficiency virus—infection among women varies greatly among different population groups. Heterosexual women represent the fastest-growing group of newly diagnosed HIV-positive cases. Although recent studies have shown a decline in the incidence of HIV in high-risk populations, such as those who use IV drugs, identification of HIV-positive pregnant women has become

TABLE 1.1  
*Preconceptional Risk Assessment:  
 History and Physical Examination*

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<i>Medical Factors</i>	<i>Psychosocial Factors</i>
Sociodemographic data	Smoking
Menstrual history	Alcohol
Past obstetric history	Drugs
Contraceptive history	Social support
Sexual history	Stress levels
Medical/surgical history	Physical abuse
Infection history	Mental illness status
Family and genetic history	Pregnancy readiness
Nutrition	Exposure to teratogens (drugs producing fetal abnormalities)
<i>Physical Examination</i>	Housing, finances, and so on
General physical examination	Extremes of physical work, exercise, and other activities
Blood pressure/pulse	
Height	
Weight	
Height/weight profile	
Pelvic examination and gynecologic examination	
Breast examination	

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SOURCE: *Caring for Our Future: The Content of Prenatal Care*, a report of the Public Health Service Expert Panel on the Content of Prenatal Care (Department of Health and Human Services, 1989).

extremely important. In 2001 the U.S. Public Health Service updated the 1995 guidelines for routine AIDS counseling and voluntary testing of all pregnant women. It is now customary at your first prenatal visit for your obstetrician to request a test for the AIDS virus. The reason for this is twofold. First, HIV-positive women are at risk for many different infections, which could put their lives and the lives of their unborn children at risk, and there are now treatments that can minimize the risk of developing such infections. Second, medicines, such as zidovudine (ZDV), have been developed that can fight the AIDS virus. ZDV can be administered to women before and during labor and can be given to babies after birth to prevent children from developing this disease. In recent studies, HIV-positive women were treated with ZDV before delivery, and the babies were given ZDV after birth. As a result, the chance of a baby developing AIDS was reduced to less than 3 percent. Having a C-section, instead of a vaginal delivery, may also reduce the risk of transmitting the virus to the baby. Given this

TABLE 1.2  
*Preconceptional Risk Assessment:  
 Laboratory Tests*

Hemoglobin or hematocrit	Hepatitis B
Rh factor	Toxoplasmosis
Rubella titer	CMV (Cytomegalovirus)
Urine: protein, sugar	Herpes simplex
Pap smear	Varicella
Tuberculosis screen	AIDS (HIV) test
Gonococcal culture	Genetic tests: Tay-Sachs, sickle cell,
Chlamydia culture	parental karyotype
Syphilis test	Illicit-drug screen

SOURCE: *Caring for Our Future: The Content of Prenatal Care*, a report of the Public Health Service Expert Panel on the Content of Prenatal Care (Department of Health and Human Services, 1989).

new therapy, pregnancies may be safer in this expanding population of patients with AIDS.

### ***Hepatitis B (HB) and Hepatitis C (HC)***

About 0.3 percent of all adults in the United States are chronic hepatitis B carriers. Approximately 300,000 new cases of HB occur annually. Acute HB occurs in 1 to 2 per 1,000 pregnancies, and chronic HB occurs in 5 to 15 of every 1,000 pregnancies. An estimated 18,500 births occur among these women annually. Without the HB vaccine, approximately 4,300 newborns would acquire HB infection from these women each year. Unless treated, about 1 percent of infected infants will develop a fatal infection, and 85 percent to 90 percent will become chronic HB carriers.

To prevent newborn HB infection, doctors must first recognize the mother's infection. Then they can immunize and administer HB immune globulin promptly after delivery. Screening during pregnancy can identify positive cases.

Women at substantial risk of having or acquiring the HB virus include those who have had sexual contact with HB-infected partners, users of illicit injectable drugs, prostitutes, institutionalized women, those with tattoos, and certain immigrant groups, including Southeast Asians. Those with an ongoing risk of acquiring HB infection should have a preventive vaccination.

HB screening is now legally required of all pregnant women in many states. It makes sense to obtain a screening prior to pregnancy.

Hepatitis C (HC) appears to affect as much as 0.6 percent of the pregnant population, and the risk factors are similar to those for HB. Unlike HB, there is no vaccine to prevent infection and no treatment to offer pregnant women to reduce the risk of transmission to the baby. Pregnant women with HC should not breast-feed because there is a 2 to 3 percent risk that the virus can be transmitted this way. Approximately 7 to 8 percent of pregnant women with HC will produce offspring with HC infection.

### *Rubella (German Measles)*

A pregnant woman infected with German measles, particularly during the first sixteen weeks, risks spontaneous abortion, stillbirth, or a baby with congenital rubella syndrome. The incidence of congenital rubella has declined by more than 99 percent since 1969, the year the rubella vaccine was licensed.

All women should be tested for rubella immunity. If you had rubella as a child, you are not necessarily immune as an adult, so routine susceptibility testing should be conducted. Most studies carried out during the early 1980s found that 10 to 20 percent of women of childbearing age had no immunity to rubella. Women considered susceptible to rubella should receive the vaccination prior to conception and then avoid pregnancy for three months. Should conception occur soon after vaccination, however, there is not a great risk of infection from the vaccination.

### *Toxoplasmosis*

Toxoplasmosis is an infection caused by a one-cell parasite named *toxoplasma gondii*. It can be found in the feces of infected cats and in raw or uncooked meats. Foods that are in contact with these contaminated meats can also be infected. It is not found in raw fish, so fear not, sushi lovers!

About one-third of adult women in the United States have antibodies to toxoplasmosis; the remainder may be at risk for a primary infection during pregnancy, which can result in fetal infection. If a pregnant woman is infected during the first or the second trimester, chances are greater that the fetus will be severely affected than if the woman is infected during the third trimester. Although earlier exposure in the pregnancy has more severe consequences, the actual risk of fetal infection is much less in the earlier trimesters.

If preconceptional testing shows that you lack immunity, you should be advised about the proper cooking of meat and during preg-

nancy avoid close contact with cats, cat litter, or soil that may contain cat feces.

Acute infection in an adult is often subtle. Symptoms, when present, generally are nonspecific and include fatigue, swollen glands, and fever.

If symptoms develop during pregnancy, prior testing can help your physician identify whether an acute infection is due to toxoplasmosis. If repeated testing during pregnancy shows the presence of toxoplasmosis antibodies in a woman who previously had negative antibodies, then acute infection is indicated. In the absence of such prior information, the interpretation of tests obtained during pregnancy may be confusing. Labs in the United States can now pinpoint the time of exposure to the parasite quite accurately, so that women can be adequately advised. Screening of those women who own or have regular contact with cats, especially outdoor cats, is very important. Probably all women considering pregnancy should be screened.

### *Fifth Disease*

Fifth disease is usually a childhood illness caused by a virus named parvovirus B19. Children who get infected have a rather mild illness, characterized by very red cheeks (slapped cheeks), and they completely recover. Adults who get exposed to this virus often have no symptoms at all. Most adults already are immune to this virus (over 60 percent) and therefore need not worry. When outbreaks of this virus occur (usually in the spring), around 50 percent of people who are exposed and not immune will catch this virus. Typically, the virus can be transmitted five to ten days before the appearance of the rash. If pregnant women were to seroconvert (become infected with parvovirus B19), up to one-third of them would infect the fetus, according to some studies. If fetal infection occurs, there is a small risk of fetal loss (2 to 9 percent), and it may cause a condition known as hydrops fetalis. Long-term development appears to be normal in the fetuses with congenital parvovirus when the fetus does survive. Your doctor has blood tests available that can check your immunity to this virus, should you be exposed, and can test to see if you have become infected.

### *Varicella Zoster Virus (Chicken Pox)*

Most women of childbearing age are immune to chicken pox. Infection during pregnancy is quite rare (0.4 to 0.7 per 1,000 women). If a non-immune pregnant woman develops chicken pox within the first half of

her pregnancy, there is a small risk of major birth defects (around 2 percent). However, if a pregnant woman develops chicken pox from five days before delivery to two days after delivery, there is a very high risk of neonatal death due to neonatal infection. Therefore, women who are not immune to chicken pox should strongly consider receiving the vaccination prior to pregnancy. (This is a live virus vaccine and cannot be given to pregnant women.) If a pregnant woman is not immune, she should avoid contact with those who may be infectious.

### *Herpes Simplex*

There are two types of herpes infections, type 1 and type 2. Although both types can occur along the birth canal, type 2 is more commonly found there. About 45 million adult Americans have been infected with genital herpes, and 1,500 to 2,000 newborns contract a herpes infection each year. Newborns who develop a herpes infection usually get infected while traveling through an infected birth canal. The newborns can get a localized infection of the skin, the eyes, and the mouth; an infection of the central nervous system; or an infection throughout the entire body. With a localized infection, there is no risk to the baby, but if the nervous system is infected, 15 percent of babies can die. If there is a generalized infection, 57 percent can die. The biggest risk to the baby occurs when the mother develops her very first outbreak (primary herpes) at the time of delivery. Some studies show that the baby can then get infected 50 percent of the time. For recurrent infections, the risk is substantially less, at around 0 to 3 percent. Because the risk to the baby is so high, it is generally recommended that if a woman has a clear infection at the time of labor, whether primary or a recurrence, she should have a cesarean section so that the baby avoids contact with the birth canal. Many doctors are now using antiviral agents, such as acyclovir, during the later weeks of pregnancy to reduce herpes recurrences and decrease the risk of needing a cesarean section.

Although uncommon, first-time outbreaks early in the pregnancy may lead to an increase in miscarriages and birth defects.

### *Other Infectious Diseases*

Screening for syphilis, gonorrhea, and chlamydia should be performed as part of general preventive health care. These infections may affect your ability to conceive and may increase the risk for ectopic pregnancy. The routine testing for cytomegalovirus, varicella, and herpes simplex has not yet been recommended by the Expert Panel on the Content of Prenatal Care, but your own physician may want to test for these.

## Maternal Medical Disease

Advances in the effectiveness of medical treatment and the greater number of pregnancies in women in their late thirties have resulted in more women with various types of chronic diseases deciding to conceive. It is reported that 15 percent of women may have medical problems at their first prenatal visit. In women over forty years old, 25 percent are often found to have medical conditions that predate the pregnancy.

If you have a preexisting chronic medical condition, preconceptional care should include an assessment of your disease status and an evaluation of the likelihood that pregnancy will affect your health and the medical condition will affect your pregnancy. Your therapy may have to be modified to optimize the pregnancy outcome. For women with certain conditions, this may include advice regarding the timing or the avoidance of pregnancy.

Modifying and identifying a drug treatment before pregnancy can be very important. Harmful drugs should be eliminated whenever possible; some medications may be replaced with alternatives that are safer in pregnancy; and the goals of therapy may be reevaluated in relationship to a pregnancy. These issues must be considered before the pregnancy.

It is impossible to list all the medical conditions of importance to a pregnancy, but some of the more common ones are:

- High blood pressure
- Chronic kidney disease
- AIDS
- Diabetes
- Alcoholism
- Thyroid disease
- Anemia
- Tuberculosis
- Cardiac diseases
- Chronic infections
- Liver disease
- Breast cancer history

*Cardiovascular diseases*, including chronic hypertension, are the most common chronic conditions among women of childbearing age. Many women have heart murmurs that have not been evaluated. A medical or a cardiological workup in such conditions may be helpful in counseling women regarding the potential impact of a pregnancy

upon their health. For some women, corrective surgery may even be advisable before conception. Women who are found to have a congenital cardiac defect may also benefit from genetic counseling.

Depending on the age group, 0.5 to 1.5 percent of women who become pregnant have preexisting *diabetes mellitus*. The children of these women are twice as likely as those of healthy women to have congenital defects. Most of the common congenital malformations in infants of diabetic mothers arise before the seventh week of gestation, so steps to reduce the rate of malformations must be taken before most women seek prenatal care or even recognize they are pregnant. Rigorous control of sugar levels at conception and during the first trimester may reduce such defects.

Special care and counseling diabetics who are planning pregnancy have resulted in significantly lower rates of congenital malformation and of affected newborns. Chronic diabetic women who receive adequate education, counseling, and preparation prior to conception are more likely to participate in controlling their diabetes and achieve a more successful outcome.

### *Other Medical Conditions*

A woman may have a history of a successfully treated medical disease that nonetheless can seriously jeopardize her pregnancy. Special testing and care prior to conception may identify her need to arrange for high-risk care during pregnancy. These diseases include thyroid disease, a history of immune thrombocytopenia, lupus erythematosus, kidney disease, and many others.

Other medical conditions may be screened for and treated as a part of routine health maintenance. For instance, identification and treatment of iron deficiency anemia can be done prior to pregnancy. Screening for tuberculosis may be of benefit in some high-risk groups. A Pap smear and treatment of chronic gynecologic infections are all important health maintenance measures, which will benefit a future pregnancy. The diagnosis of fibroid tumors of the uterus, ovarian cysts, or endometriosis may have potential reproductive effects.

## EXERCISE BEFORE PREGNANCY

A strong, fit, healthy body is an essential element in a happy, secure, and psychologically healthy pregnancy. The time to get your fitness level up and your eating habits in line is well before you start trying to

get pregnant. A strong body adapts to the physical challenge of pregnancy more easily. A well-nourished body has the nutrient reserves for the developing fetus to draw on: calcium for building bones, iron to manufacture red blood cells, and so on. Once you are pregnant, some of these basic rules will change. Later in the book we will discuss diet and exercise guidelines to follow during pregnancy, but your goal prior to pregnancy should be to get or stay in shape for your own health and the health of your future child.

There are many components of physical fitness, each important in its own right. The three most important are body composition, that is, how much of the body is fat and how much is lean muscle, bone, and water; flexibility, or the degree to which muscles and joints let us bend, stretch, and twist; and cardiovascular or aerobic efficiency, which measures the body's ability to extract oxygen from the air we breathe and speed it via the blood to every cell in the body. Although each aspect of fitness is important to health, you should balance your exercise program to meet your own particular goals.

## Getting in Shape

Exercise is appropriate for every woman at every age. It is particularly important for you when contemplating pregnancy. Being overweight at any age takes a health and a psychological toll, but for a woman planning a new pregnancy, it poses additional problems. For one thing, obesity increases the risk of toxemia and diabetes. For another, dieting is not wise during pregnancy since it would deprive the fetus of necessary nutrients. The best plan, then, is to get to a normal weight before pregnancy.

It is difficult to lose weight and keep it off by diet alone. The body seems to sense when you are dieting and lowers its metabolic rate. In other words, when you suddenly start to eat less, the body prepares to wait out a possible famine by slowing down in order to conserve fuel. If, instead of cutting calories to tip the fuel and energy balance in favor of weight loss, you increase the body's need for calories through exercise, the metabolic rate will remain stable and even increase. Therefore, the combination of reducing food intake and increasing activity will produce slow and steady weight loss. The best types of exercise to add to your routine for weight loss are those that expend large amounts of calories. They include cycling, jogging, skiing, swimming, and aerobic dancing—activities that keep a large percentage of the body's muscles working over an extended period of time.

It is difficult to be patient when setting off on a weight-loss plan, but it is important to start up slowly. Joining a health club or an

exercise class is an excellent way to get through the critical early stage of an exercise plan. During the first six months, as many as 50 percent of women lose their resolve and drop out. It takes a long time to lose 20 pounds, perhaps six months or even a year, but keep in mind that it is only one year to shed weight that may have accumulated over a period of many years.

### **Flexibility**

Joints and muscles can lose flexibility with age and lack of use, leaving the body stiff and injury prone. Proper stretching can correct and enhance your mobility. Increase flexibility by moving each joint gently, without bouncing, and then stretching it a bit beyond the current limit. This tension should not become painful. Stretching to increase flexibility in any joint can loosen stiff muscles. Tightness in the back, the trunk, and the back of the thigh muscles is associated with back problems, which often worsen during pregnancy due to the added weight of the fetus. To test your flexibility in this area, sit on the floor and slowly reach for your toes, keeping your feet flexed so that the toes point to the ceiling. If there is a gap of more than 2 to 3 inches between your fingertips and your toes, your back muscles need to be stretched. Use this test as an exercise, holding the reach for ten to twenty seconds until you feel the tightness in your back and hamstrings ease.

### **Aerobic Capacity**

The rate at which you can keep steadily running, walking, or bicycling depends on your aerobic fitness. Aerobic fitness is the body's ability to take in, transport, and use oxygen. If exercise is too intense, your body can't supply the oxygen fast enough. It makes up the difference with less efficient anaerobic (without oxygen) metabolism. This produces a substance called lactic acid in the muscles. A large amount of this acid interferes with your muscles' smooth and efficient functioning and may cause cramping pain. The idea is to reach a balance and push the body only to the point at which lactic acid can be whisked away in the blood as quickly as it is produced. Then you can continue to exercise for long periods of time.

Of all the measures of fitness, aerobic capacity is probably the most important. Unlike flexibility or muscle strength itself, the ability to endure an exercise—that is, to jog 2 miles or bike 10—is linked to some of the most important qualities of overall good health. Aerobic exercise can change your cardiovascular system and decrease your risk

of heart disease. Regular exercise may cause healthier patterns in your lipoproteins, which are the carriers of fat and cholesterol in the blood. Levels of low-density lipoproteins (LDL) tend to be lower in active people. Lowering LDL has been shown to reduce the risk of heart attacks.

High-density lipoproteins (HDL) protect against coronary artery disease. They seem to remove cholesterol from the tissue, including the inner walls of the blood vessels. Cholesterol left on blood vessels can form plaques that narrow vessels and eventually block the blood supply to the heart. This may cause a heart attack or, if the blood vessel goes to the brain, a stroke. Very active women, such as runners, have been shown to have much higher HDL levels.

Aerobic workouts must be reasonably vigorous, equivalent, say, to at least 10 miles of running per week for at least six months, to have an effect on your cardiovascular system. With aerobic fitness, your blood pressure tends to go down; your heart learns to do more work more efficiently, and your heartbeat slows down. One way of getting to know more about your fitness is to measure your resting pulse rate before you get out of bed. Eighty beats per minute or higher is common for unfit women, whereas sixty beats or lower may occur for a highly trained athlete.

How can you tell if you are aerobically fit right now? One test is to measure how long it takes you to walk or run a mile and a half on level ground. For a young woman, eleven minutes is excellent, fourteen minutes is average, and seventeen minutes is poor. To improve your score, you must train your body to improve its ability to use oxygen.

The “sing-talk method,” although not very precise, can also be used to measure how hard you are working out. If you cannot talk without gasping for breath while exercising, you have probably exceeded the target zone. If you can sing while exercising, you are probably not pushing hard enough. In order to achieve a training effect, an aerobic workout should last twenty to thirty minutes and be done at least three times a week. Starting out slowly will minimize the risk of injury as your body adapts to the increased activity.

The intensity, the duration, and the frequency of aerobic exercise should be increased gradually, one component at a time. A day off between workouts is sensible, to minimize stress on the joints and the ligaments. By gradually increasing your capacity, you may be able to exercise daily. Varying your type of workout will relieve parts of the body from continual stress and will create a welcome change of pace for your mind as well. Exercise should feel pleasantly tiring. If it is just pleasant, you are probably not working hard enough. If it is only

tiring, you are working too hard. An hour after finishing your workout you should feel good and rested but not exhausted.

A well-rounded exercise program should aim at improving strength and flexibility, as well as aerobic capacity. Muscle building, calisthenics, or weight lifting can be added to strengthen parts of the body neglected by your choice of aerobic exercise. This may include the abdominal and the upper-body muscles. You might alternate days of aerobic workouts with days of strength training. Whether done alone or as part of a longer workout, the aerobic segment should begin with a warm-up and be followed by a cool down.

There is an additional advantage to regular exercise, besides enhancing cardiovascular health and making weight control easier. Getting in shape can improve your psychological well-being. Regular exercisers feel better about themselves in general, and many report finding relief from tension, anxiety, and depression. These good feelings of mental and physical health often encourage us to improve other habits as well—eating better, sleeping more regularly, and cutting down on smoking and on alcohol and drug use. The goal of complete fitness is to strengthen your muscles and increase your endurance so that your body, on command, can walk to the store, play three sets of tennis, or carry a baby with grace and energy to spare.

Pregnancy is quite a bit like a marathon. It is physically demanding, takes a long time to complete, and requires that you have a last burst of energy in reserve for getting over the finish line. Fortunately, you don't have to be in marathon form for pregnancy, but being in shape and having strength, stamina, and a feel for your body's responses to physical demands can be a big advantage.

## NUTRITION BEFORE PREGNANCY

All of an unborn baby's nutrients come from you, the mother. How well nourished you are at the time of conception seems to play a role in the health of your developing fetus. Studies have shown that if you are well nourished and have a balanced diet when you conceive, you are more likely to give birth to a healthy baby.

A balanced diet is basic for good health at all times in your life, but especially before and during pregnancy. Eating wisely means choosing your meals from four basic food groups. In general, you will be well nourished if you eat plenty of fresh fruit, vegetables, and whole grain products, as well as two servings a day of meat, fish, poultry, or other protein food. Calcium, found mostly in milk, cheese, and other dairy products, is also important during pregnancy.

Women who are underweight and subsequently gain little weight during pregnancy are at high risk for fetal and neonatal morbidity and mortality. One recent study found that more than half the infants born to women with a prepregnancy weight of 80 percent or less than standard weight and who gained less than 23 pounds during pregnancy were of low birth weight. At the other extreme, marked obesity is associated with gestational diabetes, hypertension, large infant size, and resultant prolonged and difficult labor. A study done in 2002 demonstrated that women who were obese prior to pregnancy were more likely to gain excessive weight during pregnancy and were, on average, 18 pounds heavier than their prepregnancy weight 6 months after delivery. The optimal weight gain for very obese women during pregnancy is not definitely known. Thus, for both significantly underweight and very obese women, preconception nutritional intervention is highly desirable.

A proper medical assessment of your nutritional status should include an individual and a family history, height and weight measurement, and a discussion of your optimal weight. The history will elicit special diets, food allergies you may have, your habits and attitudes, and your knowledge of proper nutrition. Lactose intolerance may be identified and the adequacy of your dietary calcium intake assessed; lactose enzyme or calcium supplementation may be prescribed if indicated. Women on vegetarian or other special diets should receive nutrition counseling to assure the adequacy of their diet prior to and during pregnancy. Women with preexisting medical conditions, including diabetes, anemia, liver disease, and gastrointestinal disease, may require special diets. Fad diets or unusual dietary practices should be identified and discussed.

A nutritionist may be of help in evaluating and counseling you on the adequacy of your diet for pregnancy. If bulimia, anorexia nervosa, or other emotional problems affecting nutrition are identified, psychiatric counseling as well is important, particularly for women contemplating pregnancy.

Some women may benefit from special nutrition counseling due to their life situation. These include adolescents contemplating repeat pregnancy, women with a low income or a limited food budget, or those without budgeting or cooking skills.

Neural tube defects (NTDs) such as spina bifida may be prevented by early vitamin supplementation, especially with preparations containing folic acid. NTDs are more common among women of lower socioeconomic status and those with dietary histories demonstrating vitamin deficiencies. One recent study demonstrated a rate of NTDs of 3.5 per 1,000 among infants of women who never used multivitamins

before or after conception. In contrast, the rate for those who used multivitamins containing folic acid during the first six weeks of pregnancy was 0.9 per 1,000. Note that the neural tube is formed soon after conception, so it is very important to begin taking folic acid while you are trying to conceive. By the time you discover that you're pregnant, the neural tube has mostly formed and it is too late. To date, many studies have proved that folic acid can reduce the risk of this birth defect. How much folic acid should be taken? The average prenatal vitamin has 0.4 mg of folic acid. According to a large study, 0.4 mg of folate will reduce NTDs by 36 percent, whereas 5 mg will reduce the risk by 85 percent. Therefore, either through diet or supplementation, a woman should try to get at least this amount. For those who used multivitamins without folic acid during the first six weeks of pregnancy and those who began using multivitamins with folic acid at seven weeks of pregnancy, the prevalence of NTDs was similar to that among nonusers.

### Energy Balance

If you want to gain weight, you have to take in more calories and burn up fewer. If you want to lose weight, you have to take in fewer calories and burn up more. This is easy to say but difficult to translate into everyday practice. Since we eat to satisfy much more than our body's simple need for nutrients, cutting down on eating is in itself no simple task. It is also difficult to determine exactly how many calories your body needs. There is a certain baseline requirement just in being alive. Added to this basic rate is your caloric need to support movement, whether walking up a flight of stairs or running a marathon. But even that doesn't describe the complexity of the calorie in–calorie out balance.

Table 1.3 shows how many calories walking or running uses up, but these numbers are just approximations. Your exact need may vary, depending on many factors: the shape you are in, for instance; what your body's proportion of fat is; how old you are; or how hot it is outside. The only way to know what will work for your body right now is to cut back a bit on calories and see what happens.

What you eat is also critical to your health and well-being. Try to eat a little bit less to lose weight; a mouthful here and there can make an astonishing difference over a month. Of course, if your diet is too high in fats or simple carbohydrates, you should try to bring them into healthier balance as well. As far as weight loss goes, it's the total number of calories that counts. A healthy weight loss is usually no greater than 2 pounds a week. More than that and you are probably

TABLE 1.3  
*Comparative Table of Calories In/Calories Out*

FOOD	CALORIES	Minutes of Activity Required to Burn Up Calories		
		IN WALKING	IN BICYCLING	IN RUNNING
Apple	100	19	12	5
Beer (1 glass)	115	22	14	6
Raw carrot	42	8	5	2
Cottage cheese (1 tbsp.)	27	5	3	1
Ice cream (1/6 pint)	193	37	24	10
Pizza (1 slice)	180	35	22	9
Hamburger	350	67	42	18
Orange	68	13	8	4

SOURCE: Modified from *Complications of Pregnancy*, ed. S. H. Cherry, M.D., and I. Merkatz, M.D. Baltimore, Md.: Williams & Wilkins Co., 1991.

disturbing the body's water balance and excreting water, rather than burning fat. You won't be able to sustain that rate of loss anyway. If you were to eat 500 fewer calories per day and increase the amount of exercise to burn another 500 calories, you would be reducing your total calories by 1,000 a day, or 7,000 a week. That is exactly 2 pounds' worth of calories per week.

As soon as you start to think about getting pregnant, you should plan to first get your body in shape. If you are not sure what a healthy weight is, ask your doctor. Then you can work backward from your planned conception time, following the 2-pounds-a-week rule. Give yourself some leeway so the process can be as relaxed, gradual, and enjoyable as possible. By the time you are ready to get pregnant, you may just be in better shape than ever before.

## HAZARDS TO PREGNANCY

Certain hazards to a healthy pregnancy can crop up and affect reproduction. The damage can be done long before you get pregnant. Environmental hazards can damage your eggs, for example; render men sterile; or cause them to produce mutant sperm incapable of fertilization. Such hazards can crop up in what we normally think of as the environment—air, water, and soil—or from things like drugs and X rays that become part of our environment when we get sick. They may arise from things you choose to do, such as smoking, consuming

alcohol, drinking excessive amounts of coffee, or using various recreational drugs. They may also come from things you are not aware of or have no direct control over, such as toxic substances in food, water, or air; pesticides; food additives; industrial pollution; and contaminants.

When exposure occurs during pregnancy, the harmful effects vary, depending on the stage of fetal development. Different kinds of damage may occur during the preimplantation period, during the third to twelfth weeks, and after twelve weeks of gestation. Sometimes there may be long-term disabilities in our offspring, such as development of behavioral disorders or cancer, that do not become apparent until later in life. Knowledge of these potential hazards is important in preparing for pregnancy, for some of them will affect your body prior to pregnancy, while others may be hazardous during the early weeks when you may not even know yet that you are pregnant.

### **Medications That Endanger Pregnancy**

Between 1957 and 1962, thousands of people took a drug called thalidomide as a tranquilizer. Since it was available without a prescription, people assumed it was a safe drug. Inevitably, it was taken by some pregnant women, many of whom did not even know they were pregnant at the time. The problem was that this drug, if taken between the twentieth and twenty-fifth days after conception—only about two to three weeks after a missed period—interfered with the development of the fetal limbs. As soon as the association was confirmed, thalidomide was taken off the market, but not soon enough to undo the damage it had caused. The thalidomide tragedy focused much-needed attention on the effects of drugs on pregnant women and their fetuses. It served as a stimulus to intensify drug safety studies and underscored the extreme caution that must be used in prescribing any drugs during pregnancy or taking any drugs while trying to conceive.

Many studies have shown that most women take one or more drugs during their pregnancies. A Centers for Disease Control report from 1987 questioned 492 pregnant women in New York and found that 90 percent took drugs, either over-the-counter or prescription. Average drug consumption was about four medications, and this probably underreports actual maternal use.

The drugs pregnant women reported taking were from many different classes, including analgesics, antibiotics, and tranquilizers. More recently, drugs with known teratogenic effects contain very specific warning labels in both graphic and text form. Unfortunately, in

2001 researchers at the CDC found that of the drugs that harbored teratogenic effects and therefore contained warning labels, only 21 percent of people surveyed had interpreted these labels as meaning that the drugs should not be taken while attempting conception and that pregnant women should not take the medications. If you are trying to conceive, review closely your drug history with your doctor.

There is an increasing general awareness that recreational, prescribed, and over-the-counter medications may contribute to fetal damage. It has been estimated that 1 to 2 percent of congenital anomalies today are caused by drugs, environmental chemicals, or both.

Table 1.4 lists some commonly encountered drugs administered to pregnant women and their relative safety. Consult your physician before you take any medication while trying to conceive or while pregnant.

### **Toxins in the Workplace and the Environment**

The number of chemical and physical agents that pose a potential threat to some aspect of reproduction is enormous. Over 100,000 chemicals are now used in industry, of which only a small portion have been tested for possible effects on men's and women's reproductive capacity or a direct effect on a developing fetus. While most chemicals have been tested for their potential to cause cancer, the standards for reproductive safety in occupational exposure lag far behind. Table 1.5 lists some of the factors that may be hazardous to reproduction.

It is a good idea to identify hazards both you and your partner may be exposed to and to inform your doctor. Consider the environment of your home, your job, and any other places you frequent. To evaluate possible reproductive hazards, your doctor must know the type and the duration of the exposure and the stage of your pregnancy at the time of exposure, along with your history of adverse outcome in previous pregnancies.

Accurately estimating the stage of pregnancy at the time of exposure is essential because there are critical periods when the fetus is most susceptible and the potential harm can be the highest. Also, different organs have varying susceptibility to hazardous agents at different times during pregnancy. It is only during the second to the eighth week after conception (the embryonic period) that most structural defects occur.

When assessing the workplace, consider all potential means of exposure, including inhalation, skin absorption, and ingestion. Pay attention to routine exposures, as well as peak exposures, accidents, and spills.

TABLE 1.4  
*Drugs in Pregnancy*

DRUG	SAFETY
<i>Antibiotics</i>	
Penicillins	Safe
Tetracycline	Unsafe
Cephalozimines	Safe
Erythromycin (Base)	Safe
(Estolate)	Unsafe
Sulfonamides (first and second trimesters)	Safe
(third trimester)	Unsafe
Clindamycin	Safe
Metronidazole	Controversial
Miconazole (topical agents for fungus)	Safe
Acyclovir (for herpes)	Okay for indicated cases
Isotretinoin (Accutane)	Very unsafe
Progesterone (natural)	Safe
(synthetic)	Not approved
<i>Anticoagulants</i>	
Heparin	Safe
Warfarin	Unsafe
Anticonvulsants for epilepsy	None proven totally safe— some used with monitoring
Psychotropic drugs	None safe—some used carefully, if necessary, under doctor's observation
Lithium	Unsafe
Prozac	Acceptable when indicated
Zoloft	Acceptable when indicated
Hormones	Unsafe for the most part— check for specifics
Aspirin (third trimester)	Unsafe
Baby aspirin	May be therapeutic in certain pregnancies
Ibuprofen	Unsafe but can be used temporarily to treat certain conditions
Acetaminophen (Tylenol)	Safe

Since many employees may be exposed to wide varieties of potentially hazardous substances, it is extremely difficult to demonstrate that a specific substance is a reproductive hazard. In such cases, groups of occupational hazards, rather than a specific agent, are evaluated.

TABLE 1.5  
*Hazardous Occupational  
 and Environmental Exposures*

EXPOSURE	POSSIBLE OUTCOME
Anesthetic gases in the operating room	Spontaneous abortion, prematurity, fetal malformation
Lead in paint, glass, pottery, glazing	Spontaneous abortion, low birth weight, abnormal sperm production
Mercury	Cerebral palsy, mental retardation, fetal malformation
Beryllium and selenium	Birth defects
Polychlorinated biphenyls (PCBs) from industrial use, fish contaminants	Low birth weight, intrauterine growth retardation
Herbicides with dioxin	Spontaneous abortion, intrauterine growth retardation
Pesticides	Decreased sperm production
Benzine and other solvents in dry cleaning and other industries	Increased menstrual flow, anemia, bleeding disturbances
Carbon disulfide in the textile industry	Medical disorders of various types, decreased fertility, excessive fetal loss from miscarriage, stillbirth
Chemicals at toxic waste dumps	Spontaneous abortion, intrauterine growth retardation
Arsenic in pesticides, industrial waste, and copper methinemissius	Small infants, fetal malformation
Air pollutants such as carbon monoxide	Increased miscarriages, abnormalities
Cadmium and nickel in the electroplating industry and in cigarette smoke	Fetal malformation, small infants
Hyperthermia (increased body temperature in saunas and hot tubs)	Fetal malformation
Radiation (X-ray technicians, physicians)	Fetal malformation, male infertility, future cancer in offspring
Smoking	Increased miscarriage, prematurity, low birth weight, neonatal death
Formaldehyde and other sterilization media	Increased miscarriage
Vinyl chloride in plastics	Fetal malformation

## High-Risk Behaviors

### *Smoking*

From 1965 to 1990, there was a 40 percent decline in the overall prevalence of smoking; subsequently, there has been no change. Approximately one-third of women who become pregnant are smokers, but about 20 percent will quit by their first prenatal visit. Five percent of infant deaths and 14 percent of all premature deliveries in the United States have been attributed to maternal smoking. Still, 15 to 29 percent of pregnant women smoke throughout pregnancy. Smoking has been associated with increased early fetal loss, ectopic pregnancy, ectopic pregnancy, preeclampsia, placental abnormalities, intrauterine growth retardation, and congenital anomalies. Maternal smoking during pregnancy has been estimated to account for 12.8 percent of deaths from sudden infant death syndrome. A 2002 study found that smoking increased pregnancy-related health-care costs over \$360 million per year in the United States. A 1985 study showed that approximately one-third of women who smoked were unaware that smoking increased the rate of miscarriage, contributed to stillbirth, caused premature birth, and increased the risk of low birth weight. While up to 20 percent of women will quit smoking after becoming pregnant, the majority relapses during or immediately following pregnancy.

All women who smoke and who are contemplating pregnancy should be offered special counseling or programs designed to help them stop. Since most people try to quit several times before they are successful, preconceptional intervention may be particularly worthwhile. Some women may be motivated by learning about the impact of smoking during pregnancy. Heavy smokers, and those who have tried to stop but failed, may benefit from nicotine gum or a patch, as well as from a referral to a stop-smoking program. Nicotine gum and patches are considered during pregnancy only when counseling has failed. Recently, the FDA has approved Zyban as a drug to curb the craving for smoking. Zyban is actually an antidepressant, also known as Wellbutrin, and its potential effects in pregnancy are unknown.

### *Alcohol Consumption*

About 3 percent of women nationally are estimated to take three or more drinks per week during pregnancy, although much higher rates have been reported in specific populations. While many women reduce their consumption of alcohol once they learn they are pregnant, those least likely to be moderate to heavy drinkers.

The adverse effects of alcohol on the fetus may begin early in pregnancy, often before a woman realizes she is pregnant. A safe level of alcohol consumption during pregnancy has not been established. Women who consume two to four drinks per week during the first trimester are at double the risk of spontaneous abortion. At a level of three drinks a day, the average IQ of offspring in a middle-class white population has been found to be decreased by five points, and the rate of offspring with IQs less than 85 tripled. The working executive who has a drink at lunch, a second while preparing dinner, and a third during dinner is incurring significant risk if she is pregnant.

Fetal alcohol syndrome was described as recently as 1973. It clearly showed a group of newborns' problems as being due to the ingestion of alcohol by pregnant women. We now know that more than 50 percent of infants born to alcoholic women have features of this syndrome—namely, growth retardation, brain and spinal cord abnormalities, and characteristic facial changes. Since the initial report of fetal alcohol syndrome, numerous studies have appeared in the medical journals. Today the two main concerns are: What is the minimal dose of alcohol that will produce adverse effects? When during pregnancy is the fetus most vulnerable? Recent data indicate that the prevalence of this syndrome in the 1980s and 1990s was between 0.5 and 2 cases per 1,000 births.

Severe fetal alcohol syndrome occurs only in children whose mothers were heavy drinkers during pregnancy. While a decrease in alcohol consumption by these heavy drinkers before pregnancy results in less fetal problems than those observed in women who continued to drink heavily, adverse effects of alcohol as measured by psychological testing have still been seen in infants whose mothers quit drinking even early in their pregnancy. Some studies have shown significant drops in birth weight and a greater frequency of malformations in children of alcohol users. The stillbirth rate among women consuming three drinks a day was found to be two and a half times that of women drinking less than this amount. Spontaneous miscarriages are more than twice as common in pregnant women drinking moderately twice a week or more.

All of this poses a dilemma to the obstetrician and to women who are pregnant or contemplating pregnancy: How much drinking, if any, is really safe? It would seem simple to just advise complete abstinence, but such a recommendation is often not followed, especially by women who have, in fact, consumed alcohol in the past and produced normal and even exceptional children. A recommendation for complete abstinence might also cause considerable guilt and anxiety

among women who drank before they knew they were pregnant. Perhaps a total alcohol ban might lead women to substitute other drugs, which could be even more risky. Despite these realities, the medical community's official response has been that women should beware of any alcohol at all and even the small amounts of alcohol that are present in some foods and drugs. Some doctors have taken a more moderate approach, advising women of the hazards of excessive drinking and discussing this matter with each pregnant woman on an individual basis. This seems like the logical strategy, since it is not yet possible to form rigid guidelines concerning a safe quantity of alcohol during pregnancy.

### *Caffeine Use*

Caffeine has been shown to produce abnormalities in the offspring of laboratory animals. Some studies in humans have also suggested that excessive use may be bad for the fetus. One study, in which 30 ounces of coffee were consumed a day, showed a high incidence of miscarriage and prematurity among pregnant women. However, a recent study of coffee consumption involving 12,000 women did not reveal any relationship between prematurity or malformations and coffee consumption. Some studies have looked at an association between caffeine consumption and poor fetal growth. A 2002 study found that women who used caffeine and smoked substantially increased their risk for having a small baby, compared to that of women who only smoked. Our advice to patients attempting pregnancy or already pregnant is to cut coffee down to one cup per day; drink weak tea; cut out medications with caffeine, such as Empirin and Anacin; and watch out for excess cola consumption.

### *Recreational Drug Use and Substance Abuse*

The use of cocaine, heroin, and other illegal substances may lead to spontaneous abortion, intrauterine growth retardation, low birth weight, congenital defects, and fetal or neonatal death. An estimated 10 to 15 percent of women use cocaine, heroin, methadone, amphetamines, PCP, marijuana, or any combination of these, during pregnancy. Those with serious addiction problems are unlikely to seek preconceptional care. However, most women using these substances do so only occasionally.

A careful history to identify occasional use of illegal substances is part of a preconceptional evaluation. Users often do not consider occasional recreational use to be a problem, nor may they be aware of its dangers during early pregnancy. Use of cocaine during the first

trimester may be associated with subsequent problems or with congenital defects, even if use does not continue later in pregnancy.

Occasional drug users should be encouraged to abstain completely, especially if they are not actively preventing pregnancy. Women who regularly use cocaine or other illicit substances should maintain effective birth control until their substance abuse has been properly treated.

In summary, all mood-altering drugs have adverse effects on the fetus and the newborn baby, although to varying degrees. It is best to refrain from all such substances during pregnancy.

## GENETIC COUNSELING

Advances in the science of medical genetics have dramatically increased our understanding of genetic disorders. As researchers uncovered the principles behind the inheritance of genetic abnormalities, they recognized that a small number of diseases followed specific patterns as these were passed from generation to generation, and that the likelihood of their occurrence with any particular couple could be determined. In the last decade, there has been an astonishing increase in the amount of information gleaned from genes and even more so in the number of ways this information can be applied. For example, some genetic defects can be corrected after the baby is born. Some chromosome abnormalities and metabolic disorders can be accurately detected in utero with little risk to mother or fetus.

Approximately 3 percent of newborns are affected by genetic defects. A careful history can identify genetic risk. If you have a specific indication, such as a previous affected pregnancy, a family history of genetic disease, advanced maternal age, toxic exposures in the environment or at work, or a specific ethnic background, you may obtain genetic counseling and screening. Common disorders for which genetic screening is recommended include Tay-Sachs in Ashkenazic Jews, beta-thalassemia in Greeks and Italians, alpha-thalassemia in Southeast Asians and Filipinos, and sickle-cell anemia in Africans and Hispanics. In addition, if either you or your partner is affected by a genetic disease or has an affected relative, you should receive genetic counseling and possibly genetic testing.

The number of individuals and couples seeking genetic services increases each year. The number of centers offering such counseling is also growing. There are now more than 600 hospital-based genetic programs in the United States, with associated laboratory services to do studies. Some major medical centers have developed satellite clinics

that reach out into surrounding communities to provide education and improved services. Your local medical school and the March of Dimes program in New York City are good places to call for advice on finding a genetic-counseling program.

The progress of medical genetics has come during a time when attitudes toward abortion and population control are evolving, but ethical, social, religious, and economic issues remain to be resolved. The choices a couple faces are extremely difficult ones. Partners should not hesitate to lean on anyone available—especially each other—for emotional support. And they should dig up every scrap of information they can so that whatever their decision, they feel it is right for them at that time and is based on medical knowledge and careful, honest self-questioning. Then they will know they've done the best they could under the circumstances.

### Basic Genetic Principles

Genetic disorders fall into three basic groups: chromosome, single-gene (or Mendelian), and multifactorial.

#### *Chromosome Disorders*

These disorders are classified as genetic because they involve genes. They are not necessarily genetic in the way most people think of the word. They can be one-time mutations that spontaneously occur at the time of early cell division and are not inherited or passed on to offspring in succeeding generations. However, most chromosomal disorders *are* inherited.

The cells of all living organisms contain a specific amount of genetic material in their nuclei, which generally stays the same throughout the organism's lifetime. This genetic material is arranged in distinct units called chromosomes. Chromosomes are made up of special proteins called bases. These bases are arranged in a special order to make up genes. Each chromosome is believed to contain hundreds, if not thousands, of genes, and each gene is responsible for a particular characteristic of the organism. Through advances in genomic technologies, the order or the sequence of these bases has nearly been unraveled. Around 3.1 billion bases have been sequenced, and there are around 30,000 genes. In humans, every cell contains forty-six chromosomes arranged in twenty-three pairs—every cell, that is, *except* the reproductive cells. The ova of a woman and the sperm of a man come in halves, each with twenty-three unpaired chromosomes, which will make a new, complete, unique combination of genes when they come together to form a new human being.

Of the twenty-three chromosome pairs in all nonreproductive cells, twenty-two pairs are called *autosomes*. The last pair, the sex—or X and Y—chromosomes determine whether the person is male or female. A normal human female is designated 46XX because she has the correct total chromosome number of forty-six, or twenty-three pairs, one of which is XX, the combination of two X chromosomes that makes her female. A normal male is designated 46XY—he, too, has a total chromosome number of forty-six, that is, twenty-two autosomal pairs plus one pair made up of the single X and single Y chromosome combination that makes him a male.

Chromosome analysis can be done on the white cells in the blood, on cells in the bone marrow, on skin cells, on cells shed from a fetus into the amniotic fluid, or on a small sample of the placenta. The cells are grown in tissue culture so that some can be caught at the precise time in the division process when the individual chromosomes can be seen—in a normal, nondividing cell they're invisible. The chromosomes are counted and arranged in pairs according to the size, the shape, and the dark-and-light patterns they display. Each pair is then carefully numbered according to a uniform identification system.

In 1959 an extra number 21 chromosome was discovered in white blood cells grown from individuals with Down's syndrome. These people are said to have trisomy-21, another name for a type of Down's, which simply means they have three number 21 chromosomes instead of the expected two. This was an amazing genetic breakthrough: It was the first time a specific, visible chromosome abnormality was linked to specific physical defects in a human being. Since then, many other human chromosome abnormalities have been documented—some, like trisomy-21, associated with a structural alteration of the autosomal chromosomes' genetic material, others associated with a change in the sex chromosomes.

It was to be expected that these initial discoveries were made in people with obvious problems—abnormal sex characteristics, physical malformations, mental retardation, or diseases such as leukemia. But it was extremely surprising that, eventually, laboratory studies on cells from people who appeared absolutely normal sometimes showed extreme chromosome deviations. Genetics is clearly an area in which we have much left to learn.

Chromosomal surveys done on infants show that approximately 1 in 200 has a major chromosome variation, which may or may not have a negative impact on the baby's life. The most frequently seen chromosome disorder is trisomy-21, or Down's syndrome, which occurs in approximately 1 in 600 live births.

### *Single-Gene, or Mendelian, Disorders*

Mendelian disorders occur when one member of a pair of genes is abnormal. More than 2,000 genetic variations of this type have been discovered by charting inheritance patterns, although how often most of them occur is unknown. These disorders follow the nonclassic patterns first described by Gregor Mendel in 1865. Remember that genes containing specific biochemical information occur in pairs, which are located at specific places in the chromosomes. Normal individuals receive one gene (half of each gene pair) from each of their parents. Mendelian single-gene disorders are classified according to the way they are inherited: autosomal dominant, autosomal recessive, or X-linked recessive.

*Autosomal dominant* means the problem, whatever it is, will show up when just one abnormal gene is present. That single gene will “dominate” the other normal one. These problems are thought to arise through spontaneous mutations, which can then be passed directly from one generation to the next. These are different from the one-time mutations discussed previously. It is estimated that a person with a known autosomal dominant condition has a fifty-fifty chance of transmitting the mutant gene to his or her child. There are over 1,200 autosomal dominant disorders known today, and new ones are being discovered all the time. While these disorders are generally not familiar names, one example is neurofibromatosis—the Elephant Man’s disease.

Of course, not only genetic *problems* are inherited in the autosomal dominant manner. Brown eyes are dominant over blue: One blue-eyed gene from the mother and one brown-eyed gene from the father produces a brown-eyed child.

*Autosomal recessive* conditions occur when both members of a particular gene pair are abnormal. A person is said to be a carrier if he or she has only *one* abnormal gene: The single gene isn’t strong enough alone (as the dominant genes are) to make the carrier have a disorder, but the gene can be passed on to a child. If two carriers of the same abnormal recessive gene have children, there is a 25 percent (one in four) chance that their offspring will receive a double dose of the harmful gene and actually have symptoms of the disorder. Statistically, a carrier couple has *the same* 25 percent chance with each pregnancy of giving birth to a child with a recessive disorder: The chances don’t go up or down, depending on whether one child does or doesn’t have the disease. But the couple also has a 50 percent (two in four) chance of having a carrier child and a 25 percent (one in four) chance of having a child with no abnormal genes at all.

Recessive genes are also involved in the inheritance of normal traits. Remember, blue eyes are recessive. If two brown-eyed parents both have a gene for blue eyes, each child has a one-in-four chance of having blue eyes.

There are over 900 known autosomal recessive disorders, including many disorders of metabolism, such as Tay-Sachs disease. Babies born with Tay-Sachs (but *not* carriers) lack a specific enzyme, which results in the buildup of a fatty substance in the cells of the brain and the spinal cord. Symptoms—neurological problems—first appear in infants at about six months; death is inevitable by about four to six years. There is no treatment for this disorder. It occurs primarily in Jews of Eastern European origin, with a frequency of approximately 1 in 3,000 live births. Sensitive, reliable genetic tests can determine whether prospective parents are carriers, and amniocentesis can determine the genetic status of a fetus of carrier parents.

Phenylketonuria (PKU) is another metabolic problem passed by autosomal recessive inheritance. Here, too, an enzyme is lacking that is necessary for normal central nervous system function. In most of the United States, inexpensive, simple, and accurate tests for PKU are required by law for newborns. If affected babies are put on a special diet from birth, a substantial amount of otherwise inevitable mental retardation can be prevented. However, there is no carrier test available for PKU.

X-linked recessive disorders are problems that occur primarily in men but are passed along by women. This occurs because, as was explained earlier, men have one X and one Y sex-determining chromosome, while women have two Xs. So if a woman inherits a single X gene for one of these diseases, she won't *have* the disease—since in recessive disorders the single healthy X will dominate—but she will be a carrier, capable of passing on that one abnormal X to a child. Men, on the other hand, have only a single X gene; if they inherit a faulty X, they will have the disorder. There are approximately 150 X-linked recessive disorders, including hemophilia and Duchenne's muscular dystrophy.

### *Multifactorial Disorders*

These are due to the interaction of many gene pairs with one another and with environmental factors (X rays, drugs, and so forth). Multifactorial disorders result in congenital malformations such as a cleft palate, cleft lip, and defects of the nervous system. The frequency of these disorders is unknown, but the risk of recurrence in subsequent children is low.

## Genetic Counseling

Genetic counseling is a very detailed, complex, and time-consuming process. The first thing the genetic counselor does is determine the reason a couple or a person is seeking counseling. Then a complete history, or “pedigree,” is obtained, including a review of all past and present medical problems and information about age, nationality, habits, diet, hobbies, education, and vocation. Exposures to various infections and environmental hazards, such as X rays and chemicals, are investigated. If you have had a child with a genetic problem, the counselor may review all information about that child’s conception, abortions or stillbirths, and your methods of contraception. The counselor will study all available records, including birth, medical, and autopsy reports, and sometimes even family records and photographs if available. A physical examination will sometimes be done, and consultations with neurologists and ophthalmologists may be set up. Biochemical and chromosome tests may also be run.

The genetic counselor then assembles and reviews the information and makes a diagnosis. If there is a problem, the counselor must first decide if it is genetic or environmental. If it is genetic, the type of inheritance is figured out. The diagnosis and the implications will then be explained in an interview. Mode of inheritance, recurrence risk, and, finally, all possible options are fully explained and considered.

## Carriers of Genetic Disorders

The ability to detect carriers of genetic disorders can sometimes seem like a double-edged sword. Screening programs, available to test populations at risk for such disorders as sickle-cell anemia and Tay-Sachs disease, provide valuable genetic information. Genetic disorders—and carriers of them—are no longer viewed as a threat to society’s well-being, but rather as an individual or a family problem. Being labeled a carrier is an understandably upsetting experience—so much so, in fact, that some people may avoid genetic studies and all the benefits that can follow good counseling, because they fear they simply can’t deal with the knowledge.

Carrier detection may have an enormous impact on mate selection and childbearing decisions. For example, people who find out before marriage that they are carriers of a recessive disorder may, after counseling, decide to restrict their choice of mates in order to prevent the birth of abnormal children. Deciding when to tell this to a potential mate is an extremely difficult task; this is just one small example of the kind of pressure this knowledge can bring. Carrier detection

*after* marriage brings its own set of problems, which may lead to discord and even divorce. Parents of a child with a genetic disorder are usually devastated, frightened, and full of grief and guilt. Studies of mothers of hemophiliac children suggest that their severe guilt may come from their perception of themselves as being genetically responsible for their baby's condition.

Much more needs to be learned about the effects of disclosing the carrier state, so that doctors, friends, potential and actual mates, and the carriers themselves can better cope with this extraordinarily difficult situation.

Ashkenazi Jewish patients are now requested to test for a myriad of genetic diseases before attempting pregnancy. Examples include testing for Tay-Sachs disease, Canavan's disease, cystic fibrosis, Gaucher's disease, Nieman-Picks disease, Bloom's syndrome, Fanconi's anemia, mucopolidosis, and familial dysautonomia.

### Genetic Prenatal Testing

The use of amniocentesis and the new technique of chorionic villus sampling are prime examples of the way genetic advances in sterile laboratories can benefit women in real-life situations. These techniques (explained in detail in chapter 7) remove fetal cells so that their genetic makeup can be determined. Such prenatal diagnosis usually provides reassuring news. When these tests do reveal a genetic problem in a fetus, couples have the option of terminating the pregnancy.

Unfortunately, while prenatal testing can rule out a number of problems, it can't guarantee a healthy baby. Diagnosis is still not possible for many congenital abnormalities or Mendelian single-gene disorders. For example, prenatal testing can determine the sex of a fetus but *not* whether the fetus actually has an X-linked disorder such as hemophilia. Interestingly, a new technique has been developed to actually test the egg before fertilization for evidence of the abnormal gene. In vitro fertilization (IVF) is used to fertilize a normal egg and then implant it into the mother. This could assure the parents that the baby doesn't have the disease.

All women should have knowledge about prenatal tests and the kinds of information they can and cannot provide. Great advances have been made in current genetic counseling, fetal diagnosis, and screening for carriers of problematic genetic traits. Sometimes, however, a couple caught in the middle of a terribly difficult situation may feel that what is known simply isn't enough. That is true, in many ways, and will continue to be true for the foreseeable future. But sometimes

a little bit of knowledge can make a big difference, and in many cases, genetic innovations have reduced human suffering.

## THE PREPREGNANCY CHECKLIST

The following list summarizes all the things a health-conscious couple should take care of before trying to become pregnant.

- Get a checkup and discuss with your physician possible effects of existing medical conditions on pregnancy.
- Discuss possible laboratory tests to screen for toxoplasmosis, syphilis, and HIV.
- Check on your immunization to rubella and hepatitis.
- Stop smoking. Ideally, give your body a period of adjustment—say, six months. Short of that, throw away your last pack of cigarettes with your last packet of contraceptive pills or when you tuck away your diaphragm or have your IUD removed.
- Cut down on alcohol. The official word from the surgeon general is that no alcohol should be consumed during pregnancy. Since that period includes the critical time before you'll even know for sure that you are pregnant, you should stop drinking from the time you start trying to conceive. If that seems unreasonable, ask your doctor to suggest more workable guidelines.
- Maximize nutrition. If you have weight to lose, try to drop it. If you have a few pounds to gain, start eating. Remember, these things take time—a two-pound weight change per week in either direction is a safe, healthy maximum. But that's not all: Even normal-weight women often have remarkably irregular eating habits. It's time to change this, too. A body built on a strong nutritional bedrock is likely to have a healthier, easier time supporting pregnancy.
- Get fit. Pregnancy is a physical challenge, and like any other physical challenge it will be faced most comfortably with a strong, fit body. Since it's not a good idea to tackle a fitness plan once you are pregnant, the time for physical conditioning is before conception.
- Have a dental checkup. Although almost any dental work can be done during pregnancy if necessary, why worry about the possibility? Also, your dentist may want to see

you more frequently during pregnancy because of gum changes brought about by pregnancy hormones, so set up a schedule now.

- Have a gynecologic checkup three to four months before conception. It should include:
  - A breast, a pelvic, and a general physical examination
  - A Pap smear
  - Tests for sexually transmitted diseases
  - Blood tests as indicated, such as hemoglobin, blood sugar, and liver tests
  - A rubella test and, if necessary, immunization
  - A toxoplasmosis (rare infection from raw meat, fish, or cats) test if you have or have had a cat
- Have a genetic screening if there's any question of family diseases on either partner's side.
- Avoid drugs, medications (unless prescribed by your physician), X rays, and environmental hazards while trying to conceive.
- Stop birth control. The pill should be stopped three months before you try for a pregnancy. An IUD can be removed a month or two ahead of time. Barrier methods should be used during the intervening months, then stopped with your period when you want to start trying to conceive.
- Record when your last menstrual period occurred to accurately date the onset of pregnancy.

