

Preconceptional Counseling: Introduction

- ♣ In the early part of the 20th century, women with medical problems were often unable to conceive or were advised not to.
- ♣ Discoveries such as insulin and the development of effective antihypertensive medications subsequently made it possible for many of these women to contemplate pregnancy.
- ♣ Obstetrical care of women with medical problems during this time dealt almost exclusively with protecting maternal health, as little was known about pathological influences on fetal development.
- ♣ In the 1960s, research began to focus on the pathophysiology of pregnancy and perinatal outcome.
- ♣ As a result, prenatal care was gradually extended to include fetal concerns, and interest in perinatal research increased dramatically.
- ♣ The etiologies of many maternal and fetal conditions were determined, and research also clarified the genetic origins of many diseases.
- ♣ At the same time, effective contraception was developed, allowing women to postpone pregnancy and limit family size while striving to optimize perinatal outcome. The focus of **obstetrical care thus changed once again, from treating maternal and fetal diseases to predicting and preventing them.**
- ♣ "Preconceptional screening and counseling offer an opportunity to identify and mitigate maternal risk factors before pregnancy begins."
- ♣ For example, the two leading causes of death in the first year of life—**birth defects** and disorders caused by **preterm birth**—can both be significantly reduced or eliminated by the preconceptional initiation of specific preventive strategies.
- ♣ Morbidity caused by a variety of factors, including uncontrolled maternal disease, environmental exposures, and nutritional deficiencies, also can be prevented by preconceptional care.
- ♣ Furthermore, as discussed in Chapter 38, the intrauterine fetal environment has a tremendous impact on the health and well-being of the adult that fetus will become—the **Barker hypothesis**
- ♣ Thus, optimizing pregnancy conditions and outcomes has long-term health impacts that are only beginning to be apparent.
- ♣ "The preconceptional visit may be the single most important health care visit when viewed in the context of its effect on pregnancy."

Definition

- ♣ Preconceptional counseling is **preventive medicine for obstetrics**.
- ♣ Factors that could potentially affect perinatal outcome are identified, and the woman is advised of her risks. Whenever possible, a strategy is provided to reduce or eliminate the pathological influences revealed by her family, medical, or obstetrical history, or by specific testing.

Benefits of Preconceptional Counseling

- ♣ Randomized trials of the efficacy of preconceptional counseling are scarce, partly because withholding such counseling for research purposes would in many cases be considered unethical.
- ♣ In addition, because maternal and perinatal outcomes are dependent on the interaction of a variety of maternal, fetal, and environmental factors, it is often difficult to ascribe pregnancy outcome to a specific intervention.
- ♣ Nevertheless, what follows are several prospective and case-control trials that clearly demonstrate that preconceptional counseling improves pregnancy outcome.

Unplanned Pregnancy

- ♣ To be effective, counseling about potential pregnancy risks and strategies to prevent them must be provided before conception.
- ♣ By the time most women realize they are **pregnant—1 to 2 weeks** after the first missed period—the fetal spinal cord has already formed and the heart is beating.
- ♣ Many prevention strategies, for example, folic acid to prevent neural-tube defects, are ineffective if initiated at this time.
- ♣ The Centers for Disease Control and Prevention estimate that up to half of all pregnancies are unplanned, and there is evidence that these may be at greatest risk.
- ♣ An important measure of the **effectiveness of preconceptional counseling** is, therefore, its **influence in reducing the number of unintended pregnancies**.

Chronic Medical Disorders

1. Diabetes Mellitus

- ♣ Because maternal and fetal pathology associated with hyperglycemia is well known,
- ♣ Diabetes is the prototype of a condition for which preconceptional counseling is beneficial.
- ♣ Many of the complications can be avoided if conception occurs when glucose control is optimal.
- ♣ Such control requires either that glucose levels be chronically well regulated—always a goal, but difficult to achieve—or that the woman make necessary changes before attempting conception.
- ♣ Preconceptional counseling can educate her about risks and provide a program designed to reduce them. The utility of preconceptional counseling in preventing diabetes-related complications at all stages of pregnancy has been confirmed.
- ♣ Importantly, all studies showed that counseling is associated with significantly fewer malformations.
- ♣ Dunne and co-workers (1999) reviewed the impact of preconceptional counseling on other diabetes-related neonatal morbidity. Women who received counseling sought prenatal care earlier, had lower hemoglobin A_{1c} levels, **and were less likely to smoke during pregnancy**. Their outcomes were compared with those of a cohort of women who did not receive such counseling. Of the women who received counseling, none were delivered before 30 weeks compared with 17 percent in the uncounseled group.
- ♣ In addition, the counseled women had fewer macrosomic infants—25 versus 40 percent; they had no growth-restricted infants compared with 8.5 percent; they had no neonatal deaths compared with 6 percent; and their infants had 50 percent fewer admissions to the intensive care nursery—17 versus 34 percent.
- ♣ **Preconceptional counseling also reduces obstetrical complications and health care costs in diabetic women.** These improved outcomes were associated with savings of \$34,000 in direct medical costs per patient who received counseling.

2. Epilepsy

- ♣ Women with epilepsy are **two to three times more likely** to have infants with structural anomalies than unaffected women.
- ♣ Some reports indicate **that epilepsy itself increases** the incidence of congenital anomalies, independent of the **effects of antiseizure medication**.
- ♣ Preconceptional counseling usually includes recommendations to switch to monotherapy with the least teratogenic medication.
- ♣ Epileptic women also are advised to take supplemental folic acid.

3. Other Chronic Diseases

- ♣ Women with hypertension, renal disease, thyroid disease, asthma, and heart disease had significantly better outcomes than previous pregnancies.
- ♣ Indeed, 80 percent of those counseled gave birth to a normal infant compared with only 40 percent in the previous uncounseled gestation.

Genetic Diseases

- ♣ Birth defects are currently the leading cause of infant mortality and account for **20 percent of all infant deaths**.
- ♣ These defects can be avoided with three types of prevention strategies.
- ♣ The preferred strategy is **primary prevention**—avoidance of causal factors—which is becoming possible for more congenital diseases as their etiologies are discovered.
- ♣ **Secondary prevention**—identifying and terminating affected pregnancies—is an alternative strategy for single-gene disorders and other defects that cannot be prevented.
- ♣ Surgical correction of structural defects is one type of **tertiary prevention**, but it is not possible for most genetic disorders.
- ♣ The benefits of preconceptional counseling usually are measured by comparing the incidence of new cases before and after the initiation of a counseling program.
- ♣ Some examples of congenital conditions that clearly benefit from counseling include **neural-tube defects, phenylketonuria, Tay-Sachs disease, and the thalassemias**.

A. Neural-Tube Defects (NTDs)

- ♣ The incidence of these defects is 1 to 2 per 1000 live births, and they are second only to cardiac anomalies as the most frequent structural fetal malformation.
- ♣ Some NTDs are associated with a specific mutation in the methylene tetrahydrofolate reductase gene (677C to T), the adverse effects of which appear to be largely overcome by periconceptional folic acid supplementation
- ♣ The Medical Research Council on Vitamin Study Research Group (1991) conducted a randomized double-blind study of preconceptional folic acid therapy at 33 centers in seven European countries. Women with a previous affected child who took supplemental folic acid before conception and throughout the first trimester reduced their NTD recurrence risk **by 72 percent**. Perhaps more importantly, because 90 to 95 percent of NTDs occur in families with no prior history, (1992) subsequently showed that supplementation reduced the a priori risk of a *first* NTD occurrence.

B. Phenylketonuria (PKU)

- ♣ This disorder is an inborn error of phenylalanine metabolism.
- ♣ It is an example of a disease in which the **fetus is not at risk to inherit the disease but may be damaged by the effects of maternal genetic disease.**
- ♣ For such conditions, preconceptional counseling regarding strategies to improve the intrauterine environment constitutes primary prevention and may significantly reduce fetal morbidity.
- ♣ Individuals with PKU who eat an unrestricted diet have abnormally high blood phenylalanine levels. This amino acid readily crosses the placenta and can damage **developing fetal organs; especially neural and cardiac tissues.**
- ♣ With preconceptional counseling and adherence to a phenylalanine-restricted diet before pregnancy, the incidence of fetal malformations is dramatically reduced.

C. Tay- Sachs Disease

- ♣ This disease is a severe, autosomal-recessive neurodegenerative disorder that leads to death in childhood.
- ♣ The effectiveness of preconceptional counseling in reducing genetic disease has been most clearly demonstrated in Tay-Sachs disease.

D. Thalassemias

- ♣ These disorders of globin-chain synthesis are the most common single-gene disorders worldwide.
- ♣ Approximately 200 million people carry a gene for one of these hemoglobinopathies.
- ♣ Hundreds of mutations have been identified that cause several important thalassemia syndromes.
- ♣ Some of these could be avoided by both primary and secondary prevention.
- ♣ In endemic areas such as Mediterranean countries, counseling and other prevention strategies have reduced the incidence of new cases by at least 80 percent.

Preconceptional Counselors

- ♣ Practitioners providing routine health maintenance for reproductive-aged women have the best opportunity to provide preventive counseling.
- ♣ Gynecologists, internists, family practitioners, and pediatricians can do so at the annual examination.
- ♣ The occasion of a negative pregnancy test is a good time for counseling.
- ♣ Basic preconceptional advice regarding diet, alcohol use, smoking, illicit drug use, vitamin intake, exercise, and other behaviors can be provided by the primary care provider, including the obstetrician-gynecologist.
- ♣ Medical records should be obtained and reviewed. Counselors should be knowledgeable about relevant medical diseases, prior surgery, reproductive disorders, or genetic conditions, and must be able to interpret data and recommendations provided by other specialists.
- ♣ The practitioner who is uncomfortable providing counseling should refer the woman or couple to a counselor with special expertise.

Preconceptional Counseling Visit

Personal and Family History

- ♣ Counseling begins with a thorough review of the medical, obstetrical, social, and family histories.
- ♣ Useful information is more likely to be obtained by asking specific questions about each aspect of the history and about each member of the family than by asking general, open-ended questions.
- ♣ **The interview may take 30 minutes to an hour.**
- ♣ Some important information can be obtained by questionnaire, ideally at a routine prepregnancy visit.
- ♣ Commercially prepared questionnaires are available that address medical and surgical history; reproductive history, including outcomes of each prior pregnancy; medication use and drug allergies; family history of medical or genetic diseases and reproductive abnormalities; racial or ethnic origin; social risk factors such as alcohol, illicit drugs, smoking, high-risk sexual behavior, and spousal abuse; environmental risk factors such as exposure to pesticides or other chemicals; and home environment and stress inducers. Answers should be reviewed with the patient to ensure appropriate follow-up, including obtaining relevant medical records or consultant notes.

Medical History

- ✿ Preconceptional counseling should address all risk factors pertinent to both mother and fetus.
- ✿ General questions to be answered include how pregnancy will affect maternal health, and how a high-risk condition will affect the fetus.
- ✿ Almost any medical, obstetrical, or genetic condition warrants some consideration prior to pregnancy.
- ✿ These conditions should be discussed in terms of general maternal and fetal risks, and suggestions for prepregnancy evaluation should be offered.
- ✿ Finally, advice for improving outcome is provided. More detailed information on specific diseases such as diabetes, hypertension, collagen vascular disorders, and others are found in the relevant chapters.

Genetic Diseases

- ✿ Women whose ethnic background, race, or personal or family history places them at increased risk to have a fetus with a genetic disease should receive appropriate counseling.
- ✿ Women who have a genetic disease usually require additional counseling about their own risks by someone knowledgeable about genetics.
- ✿ This is because genetic conditions are often associated with unique medical problems that may be adversely affected by pregnancy or that can adversely affect pregnancy outcome.
- ✿ They also may benefit from consultation with other specialists, for example, anesthesiologists, cardiologists, or surgeons. A variety of genetic resources can be accessed for detailed information about many inherited disorders.

Reproductive History

- ✿ This component includes questions regarding infertility; abnormal pregnancy outcomes, including miscarriage, ectopic pregnancy, recurrent pregnancy loss, and preterm delivery; and complications such as preeclampsia or placental abruption.
- ✿ The reproductive history of first-degree relatives also may be helpful.
- ✿ For example, if a patient with recurrent pregnancy loss has other family members with the same history, **her risk of carrying a familial translocation or another chromosomal rearrangement is increased.**
- ✿ A history suggesting an incompetent cervix or a uterine anomaly should prompt an appropriate evaluation. The need for assisted reproductive technologies should be noted and associated risks discussed.

- ♣ The latter include a significantly increased risk of multiple gestation. Counseling regarding intracytoplasmic sperm injection and cryopreservation of embryos also should be provided.
- ♣ Risk factors for recurrent preterm delivery, preeclampsia, placental abruption, and repeat cesarean delivery are summarized in discussions of these disorders later in this text.

Social History

Maternal Age

- ♣ The mother's age can have an impact on pregnancy at both ends of the reproductive spectrum.

Teenage Pregnancy

- ♣ According to the American College of Obstetricians and Gynecologists (2003), the pregnancies of women between the ages of 15 and 19 years **accounted for about 11** percent of all births in 2001.
- ♣ Teenagers are more likely to be anemic, and they are at increased risk to have growth-restricted infants, preterm labor, and higher infant mortality.
- ♣ The incidence of sexually transmitted diseases—common in adolescents—is even higher during pregnancy.
- ♣ Because most teenage pregnancies are unplanned, teens rarely seek preconceptional counseling.
- ♣ Teenagers usually are still growing and developing, and thus have greater caloric requirements than older women.
- ♣ The normal or underweight teenager should be advised to increase caloric intake by 400 kcal/day.
- ♣ The obese **teenager may not need additional calories.**
- ♣ Nonjudgmental questioning may elicit a history of substance abuse. Instructions regarding the identification or prevention of common pregnancy complications should be given.

Pregnancy after Age 35

- ♣ Currently, **about 10 percent of pregnancies** occur in women in this age group.
- ♣ The older woman is more likely to request preconceptional counseling, either because she has postponed pregnancy and now wishes to optimize her outcome, or because she plans to undergo infertility treatment.

- ♣ In the past, the indelicate term *elderly gravida* was used to arbitrarily define women over 35. **Although the term is now passé, certain age-related adverse pregnancy outcomes do begin to increase at this age.**
- ♣ Some studies indicate that after age 35, **women are at increased risk for obstetrical complications as well as perinatal morbidity and mortality.**
- ♣ There is no doubt that the older woman who has a chronic illness or who is in poor physical condition has increased risks. For the physically fit woman without medical problems, however, the risks are much lower than previously reported.
- ♣ Maternal mortality is higher in women age 35 and older, but improved medical care may ameliorate this risk.
- ♣ Buehler and colleagues (1986) reviewed maternal deaths in the United States from 1974 through 1982. From 1974 through 1978, **older women had a fivefold increased relative risk of maternal death compared with that of younger women.** By 1982, however, the mortality rates for older women had decreased by 50 percent. They concluded that this was probably due to improvements in health care.
- ♣ Maternal age-related fetal risks primarily stem from
 1. Iatrogenic preterm delivery required for maternal complications such as hypertension and diabetes,
 2. Spontaneous preterm delivery,
 3. Fetal growth disorders related to chronic maternal disease or multiple gestation,
 4. Fetal aneuploidy, and
 5. Pregnancies resulting from use of assisted reproductive technology.
- ♣ Most researchers have found that fetal aneuploidy is the only **congenital abnormality related to maternal age.**
- ♣ Older paternal age is associated with an increased incidence of genetic diseases caused by new autosomal-dominant mutations, but the incidence is still low.
- ♣ Accordingly, whether targeted ultrasound examinations should be performed solely for **advanced maternal or paternal age is controversial.**
- ♣ Although the incidence of dizygotic twinning increases with maternal age, the most important cause of multifetal gestations in older women currently is conception with the use of **assisted reproductive technology and ovulation induction.**
- ♣ According to the Centers for Disease Control and Prevention (2002), **0.7 percent of all 3.9 million births in the United States in 1998 were the result of these techniques.**
- ♣ More than half of this percentage were multiple infant births, which account for much of the morbidity from preterm delivery and neurological sequelae

Recreational Drugs and Smoking

- ♣ Alcohol-related mental retardation is currently the **only mental retardation syndrome amenable to primary prevention.**
- ♣ The first step in preventing this and other types of drug-related fetal damage is the woman's honest assessment of her usage.
- ♣ Questions regarding usage should be asked in a **nonjudgmental manner.**
- ♣ The alcoholic patient can be identified by asking the well-studied TACE questions, which correlate with DSM-IV criteria for lifetime alcoholism diagnoses.
- ♣ **TACE** is a series of four questions concerning **tolerance to alcohol**, **being annoyed** by comments about their drinking, attempts to **cut down**, and a history of drinking early in the morning—the **eye opener.**
- ♣ According to the Centers for Disease Control and Prevention (2003), almost one fourth of women who are **between the ages of 18 and 44 years smoke cigarettes.**
- ♣ Smoking affects fetal growth in a dose-dependent manner. **It increases the risk of preterm labor, fetal growth restriction, and low birthweight.**
- ♣ It has also been associated with an increased incidence of **attention-deficit/hyperactivity disorder** and behavioral and learning problems typically identified by school.
- ♣ Smoking also increases the risk of pregnancy complications related to vascular damage, such as uteroplacental insufficiency and placental abruption.
- ♣ After counseling, the woman should be provided with a prepregnancy program to reduce or eliminate smoking.

Environmental Exposures

- ♣ Everyone is exposed to environmental substances, but fortunately only a few agents have an impact on pregnancy outcome.
- ♣ Exposures to infectious organisms and chemicals impart the greatest risk.
- ♣ Examples of high-risk exposures include
 1. **Pregnant nurses** exposed to cytomegalovirus or respiratory syncytial virus;
 2. Day-care workers exposed to **parvovirus and rubella;**
 3. Industrial workers exposed to chemicals such as heavy metals or organic solvents; and
 4. Women living in rural areas exposed to potentially harmful chemicals through **pesticide use or contaminated well water.**
- ♣ Methyl mercury is a recently recognized environmental contaminant that is especially important because all pregnant women are potentially at risk.

- ♣ It is now well established that certain kinds of large fish are contaminated.
- ♣ Mercury is a neurotoxin that readily crosses the placenta and has adverse effects on fetal neurological development.
- ♣ Accordingly, the US Food and Drug Administration (2004) has recommended that pregnant women not eat shark, swordfish, king mackerel, or tilefish and that they consume no more **than 12 ounces of other kinds of shellfish or other fish per week.**
- ♣ Albacore or "white" tuna has more mercury than other canned tuna.

Lifestyle and Work Habits

Diet

- ♣ Pica for ice, laundry starch, clay, dirt, or other nonfood items should be discouraged.
- ♣ In some cases, it may represent an unusual physiological response to iron deficiency.
- ♣ Many vegetarian diets are protein deficient but can be corrected by increasing egg and cheese consumption.
- ♣ Obesity is associated with a number of maternal complications such as hypertension, preeclampsia, gestational diabetes, labor abnormalities, postterm pregnancy, cesarean delivery, and operative complications.
- ♣ It is also associated with adverse fetal outcomes, **including spina bifida and ventral wall defects, late fetal death, and preterm delivery.**
- ♣ In addition to nutritional deficiencies, anorexia and bulimia increase the risk of associated maternal problems such as electrolyte disturbances, cardiac arrhythmias, and gastrointestinal pathology..

Exercise

- ♣ There are no data to suggest that exercise is deleterious during pregnancy; thus, conditioned **pregnant women usually can continue to exercise** throughout gestation.
- ♣ As pregnancy progresses, balance problems and joint relaxation may predispose to orthopedic injury.
- ♣ The woman should be advised to **not exercise to exhaustion, avoid supine positions, avoid activities requiring good balance**, augment heat dissipation and fluid replacement, and avoid **extreme weather conditions.**

Domestic Abuse

- ♣ Pregnancy can exacerbate interpersonal problems and is a time of increased risk from an abusive partner.
- ♣ **One in six women is abused during pregnancy.**
- ♣ The interviewer should inquire about risk factors for domestic violence, and should offer intervention as appropriate.
- ♣ Abuse is more likely in women whose partners abuse alcohol or drugs, are recently unemployed, **have a poor education or low income**, or have a history of arrest.

Family History

- ♣ The most thorough method for obtaining a family history is to construct a pedigree using symbols.
- ♣ The health and reproductive status of each "blood relative" should be individually reviewed for medical illnesses, mental retardation, birth defects, infertility, and pregnancy loss.
- ♣ Certain racial, ethnic, or religious backgrounds may indicate increased risk for specific **recessive disorders**.
- ♣ Although most women can provide some information regarding their history, their understanding may be limited.
- ♣ For example, several studies have shown that pregnant women often fail to report a birth defect in the family or report it incorrectly.
- ♣ It is thus important to verify the type of reported defect or genetic disease by reviewing pertinent medical records or by contacting affected relatives for additional information.

Immunizations

- ♣ Preconceptional counseling includes assessment of immunity to rubella and hepatitis B.
- ♣ Depending on health status, travel plans, and time of year, other immunizations may be in order.
- ♣ Vaccines consist of either :
 1. Toxoids (e.g., tetanus);
 2. Killed bacteria or viruses (e.g., influenza, pneumococcus, hepatitis B, meningococcus, and rabies); or
 3. Attenuated live viruses (e.g., **varicella-zoster, measles, mumps, polio, rubella, chickenpox, yellow fever**).

- ♣ Immunization during pregnancy with toxoids or killed bacteria or viruses has not been associated with adverse fetal outcomes.
- ♣ *Alternatively, live-virus vaccines **are not recommended** during pregnancy and ideally should be given at least 1 month before attempts to conceive.*
- ♣ Women inadvertently given measles, mumps, rubella, or varicella vaccines during pregnancy, however, are not necessarily advised to seek pregnancy termination.
- ♣ Most reports indicate that immunization to any of these agents poses only a theoretical risk to the fetus.
- ♣ Lastly, immunization to smallpox, anthrax, and other bioterrorist diseases should be discussed.

Screening Tests

- ♣ Certain laboratory tests may be helpful in assessing the risk of complications during pregnancy.
- ♣ These include basic tests that are usually **performed during prenatal care**.
- ♣ For example, **rubella, varicella, and hepatitis B** immune status should be determined so that vaccination can be carried out as part of preconceptional care.
- ♣ A **complete blood count** with mean red blood cell volume will exclude most serious inherited anemias.
- ♣ **Hemoglobin electrophoresis** is performed in individuals at increased risk, such as African-Americans for sickle syndromes and
- ♣ Women of Mediterranean or Asian origin for **thalassemias**.
- ♣ Women with Jewish ancestry are candidates for carrier testing for **Tay-Sachs and Canavan** disease, whereas Caucasians of northern European descent may elect screening for **cystic fibrosis**.
- ♣ Partners of women discovered to be carriers of autosomal-recessive diseases should be tested to determine the risk to future offspring.
- ♣ Couples discovered to be at significant increased risk of having an affected child can then contemplate their **reproductive options before undertaking a pregnancy**.
- ♣ More specific tests may assist the evaluation of women with certain chronic medical diseases. Examples of some, but certainly not all, chronic diseases that may be assessed by prenatal testing **include kidney disease, cardiovascular diseases, and diabetes**.
- ♣ In the first example, the outcome of pregnancies complicated by chronic renal disease can be predicted to some extent by the serum creatinine level.
- ♣ In the second example, the likelihood that a woman with cyanotic heart disease will have a successful pregnancy outcome can be predicted by a number of factors.

- ♣ The third example is the use of hemoglobin A_{1c} measurement to assess diabetic control during the preceding 6 weeks.
- ♣ This in turn can be used to compute risks for major anomalies. These figures may be useful in gauging patient compliance and motivation.
- ♣ Although these data are from women with severe diabetes, the incidence of fetal anomalies in women who have gestational **diabetes associated with fasting hyperglycemia is increased fourfold compared with nondiabetic control women.**

Electromagnetic Energy

- ♣ There is no evidence in humans or animals that exposure to various electromagnetic fields such as high-voltage power lines, electric blankets, microwave ovens, and cellular phones causes adverse fetal effects

Relationship of Chronic Renal Insufficiency with Pregnancy Outcome (in Percent)			
	Serum Creatinine (mg/dL)		
Outcome	<i>Mild <1.5</i>	<i>Moderate 1.5–3.0</i>	<i>Severe ≥3.0</i>
Preterm birth	13	50	100
Perinatal death	5	17	33
Fetal growth restriction	10	20	100
Abortion	11	21	25
Surviving infants	84	62	50