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Maternal & Neonatal Nursing Care Plans



Annamma Jacob

2nd Edition

JAYPEE

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CHAPTER 2

Genetic Counseling

DESCRIPTION

Genetic counseling is a communication process that deals with human problems associated with the occurrence or risk of a genetic disorder in a family. The process can be prospective (counseling delivered to a client/couple of reproductive age before conception or before birth of an affected child), or it can be retrospective (counseling delivered after the birth of an affected child). In many cases, however, the need for genetic counseling first becomes apparent during the first trimester.

Counseling also involves genetic screening, whereby a high-risk, such as consanguineous couples or general population is analyzed to detect the presence of disease, and case finding of couples at potential risk based on medical/family histories.

Client Assessment Database

- Hypertension.
- Bleeding (insulin-dependent).
- Weight gain may be inappropriate for gestational age (smaller gain may negatively affect fetus).
- Maternal insulin-dependent diabetes.
- Presence of eating disorders (e.g. anorexia nervosa, bulimia, or obesity).
- Infection (e.g. sexually transmitted disease (STD), pelvic inflammatory disease).
- Presence of seizure disease, degree/method of control.
- Significant exposure to radiation, toxic chemicals, or infectious teratogens (e.g. rubella, toxoplasmosis, cytomegalovirus, HIV virus/AIDS, and other STDs).
- Potential infections (e.g. meningitis, encephalitis).
- Postnatal nutritional or stimulatory deprivation.
- Breech presentation especially with anencephaly.
- History of two or more first trimester abortions, fetal demise, or previous child with chromosomal abnormality.
- Identifiable genetically transmitted disorder.
- Use of ovulation stimulants, such as clomiphene or menotropins.
- Interfamily marriages (consanguinity).
- Guilt/blame toward self and/or partner who carries defective gene.
- Positive family history/pedigree of known genetic or inherited disorders (e.g. sickle cell anemia, hemophilia, cystic fibrosis, phenylketonuria, craniospinal defects, renal malformations, Huntington's chorea).
- Familial disorders (cancer, heart disease, diabetes, allergies), congenital abnormalities (Down syndrome, mental retardation, neural tube defects), or inborn errors of metabolism (Tay-Sachs disease, maple syrup urine disease).
- Drug usage [alcohol, over-the-counter (OTC), prescribed or street drugs, anticonvulsant medication].

Diagnostic Studies

- Chromosomal analysis: Using buccal smear/serum.
- Amniocentesis (at 14–16 weeks): Determines sex chromatin (rules out sex-linked disorders), biochemical studies (rules out inborn errors of metabolism): Elevation of alpha-fetoprotein (AFP) levels reflects neural tube defects (NTD).
- Ultrasonography (using real time) preceding or during amniocentesis: Rules out missed abortion, visualizes term pregnancies, and identifies gross abnormalities/problems.
- Serum AFP levels (16–18 weeks): Identifies NTD; if abnormally high/low, test is repeated.
- Amniography/fetography: Identifies gross problems such as major soft-tissue abnormalities, esophageal/duodenal atresia, and bone abnormalities (may be potentially teratogenic as special X-ray techniques are used).
- Chorionic villi sampling (CVS) at 8 weeks: May replace amniocentesis as a genetic diagnostic tool.

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Nursing Priorities

1. Assist client/couple/family to recognize and understand specific situation.
2. Facilitate therapeutic use of informational resources.
3. Provide ongoing emotional support.

Discharge Goals

1. Copes effectively with situation.
2. Completes counseling process.
3. Understands information specific to individual situation.

Nursing Diagnosis

Anxiety (Specify Level)

Definition

Vague, uneasy feeling of discomfort or dread accompanied by an autonomic response; the source of it is often nonspecific or unknown to the individual.

Possible Etiological Factors / Related Factors

- Presence of specific risk factors (e.g. history of genetic problem, exposure to teratogens).
- Situational crisis.
- Threat to self-concept (perceived/actual).
- Conscious or unconscious conflict about essential values (beliefs) and goals of life.

Nursing Assessment

- Manifestations of anxiety.
- History of genetic disorders or risk factors client is aware of.
- Expressed concerns and fears.
- Family and social support available.

Defining Characteristics

- Increased tension.
- Apprehension.
- Uncertainty.
- Feelings of inadequacy.
- Concern regarding changes in life events.
- Insomnia.

Expected Outcomes

Client will:

- Acknowledge awareness of feelings of anxiety.
- Verbalize realistic concerns related to process of genetic counseling.
- Appear relaxed and report that anxiety is reduced to manageable level.
- Identify and use resources or support systems effectively.

Nursing Interventions

Interventions	*Rationales*
Assess nature, source, and manifestations of anxiety.	Identifies specific areas of concern and determines direction for and possible interventions.
Provide information about specific genetic disorder, risks involved with reproduction and available prenatal diagnostic measures.	May relieve anxiety associated with the unknown and assist family to cope with stress, make decisions, and adapt positively to choices.
Promote ongoing sharing of concerns and feelings.	Provide opportunity for discussion of test results on fetus and assist with interpretation of information, especially following abortion. Listen to expressions of concerns and feelings about situation.

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Interventions	*Rationales*
Review procedure and what to expect in terms of discomfort, if fetus is affected and couple elects to terminate pregnancy.	Client/couple may be extremely anxious and guilt-ridden during an uncomfortable procedure. Information can reduce anxiety.
Visit couple after procedure. Provide anticipatory guidance in terms of physical and psychological changes.	Follow-up visit by the nurse may relieve anxiety/depression in couple following abortion for genetic indications.
Provides opportunity for client/couple to begin resolution of situation. Level of anxiety is usually higher in the couple who have given birth to a child with a chromosomal disorder.	Helps to confirm the diagnosis, reduces anxiety associated with uncertainty of whether fetus was really affected and whether couple made the right choice. When concerns and feelings are expressed/listened to, client needs can be identified more readily.
*Refer to further counseling as indicated, e.g. psychiatric referral.	Anxiety may not be resolved sufficiently necessitating additional professional assistance.
*Assist couple in identifying community agencies to aid in care of their newborn in the event that they elect to continue the pregnancy after fetus is found affected or when diagnosis is made after delivery.	Helps to reduce anxieties regarding how the couple will meet their baby's special needs.
** Denotes collaborative interventions*	

Evaluations for Expected Outcomes

Client/couple:
- Appears relaxed and reports that anxiety is reduced to manageable level.
- Identifies and uses resources/support systems effectively.

Nursing Diagnosis

Knowledge Deficit Regarding Purpose and Process of Genetic Counseling

Definition

Absence or deficiency of cognitive information related to genetic counseling.

Possible Etiological Factors / Related Factors

- Lack of awareness of the purpose of genetic counseling, its applicability to their situation, and components of the decision-making process.
- Lack of awareness of available options and available methods of prenatal diagnosis.

Nursing Assessment

- Family history of genetic problems.
- Obstetric history, such as miscarriages, fetal demise, hypertension, bleeding, infection, exposure to radiation, environmental pollutants, etc.
- Available support systems.

Defining Characteristics

- Verbalization of the problem.
- Statement of misconceptions.
- Request for information.

Expected Outcomes

Client/couple will:
- Describe the disorder and identify available options.
- Discuss purpose and findings of specific prenatal detection methods.
- Estimate risks associated with reproduction.

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Nursing Interventions

Interventions	*Rationales*
Discuss the purposes/goals of genetic counseling.	Serves to educate parents about the risks in their situation and ultimately to help them make educated/informed decisions. Misconceptions may interfere with the decision-making process.
Obtain pertinent family history. Create pedigree chart from information and include both parents in interview.	Provides accurate picture of the affected person in relation to other family members and serves to identify other persons similarly affected. Ensures that both parents are provided the same information.
Identify risk factors in obstetric history, such as 3 or more miscarriages, fetal demise, hypertension, bleeding, infection, radiation and environmental pollutants (e.g. lead, formaldehydes) drug use, birth trauma, or identifiable genetically transmitted disorder.	These factors may produce a phenocopy, an imitation of a genetic disorder.
Distinguish between environmental causes of mental retardation and those with a genetic component (e.g. Down syndrome)	Environmental causes include birth trauma, chemical teratogens, infectious teratogens (e.g. rubella, toxoplasmosis, cytomegalovirus), postnatal infections (e.g. meningitis, encephalitis), and postnatal nutritional/sensory deprivation.
Carry out ongoing family assessment for available support systems. Note signs of inappropriate coping/maladaptation. Offer information, as family is ready to hear it.	Negative coping may interfere with cognitive functioning and hinder educational process. Some families are not ready to listen immediately after diagnosis is made and may not listen effectively the first time information is presented.
Arrange for follow-up visits to counseling service.	Provides opportunity to reinforce information when family is ready to hear it.
Provide information about individually appropriate diagnostic procedures/screenings, such as amniocentesis, serum testing for AFP, and ultrasonography, as well as newer methods such as CVS, fetoscopy, percutaneous umbilical blood sampling (PUBS) or cordocentesis, computed tomography scanning, and magnetic resonance imaging.	Prenatal diagnosis permits preparation for or offers an alternative to having children with genetic disorders.
Permit couple to arrive at own decision regarding their participation in or refusal of prenatal testing procedures.	Allows couple to make informed choices based on understanding of long-term problems associated with the disorder in question at their own pace. May assist in determining whether the fetus at risk for genetic disorder is actually affected.
Review anticipated follow-up, especially if client is currently pregnant.	May include regular assessment of fetal heart tones, such as obtaining a 20-minute tracing on electronic monitoring.
Encourage participation with support groups and groups concerned with specific genetic disorders as indicated.	Helps client identify with others who have dealt with similar genetic disorders. Provides role model and information about identified disorder.
Obtain referrals with specialty clinics and agencies that deal with specific genetic disorders (e.g. cystic fibrosis, hemophilia).	May be needed to answer questions/concerns that are presented in genetic counseling, especially if genetic component is involved.
Obtain informed consent for procedures or tests. Review risks associated with procedure.	Client needs to be aware of any risks involved to herself or the fetus, especially if the procedure is invasive such as amniocentesis; chorionic villus sampling (CVS) or percutaneous umbilical blood sampling (PUBS).

Evaluations for Expected Outcomes

Client:

- Describes the disorder and identifies available options.
- Discusses the purpose and findings of prenatal detection methods.
- Estimates risks associated with reproduction.

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Nursing Diagnosis

Risk for Altered Family Process

Definition

At increased risk for change in family relationships and/or functioning.

Possible Etiological Factors / Risk Factors

- Situational crisis.
- Individual/family vulnerability.
- Difficulty reaching agreement regarding decision.

Nursing Assessment

- Relationship of couple to one another.
- Strength of family bonds.
- Perception of the disorder.
- Attitudes and religious beliefs.

Defining Characteristics

None, as this is a risk diagnosis.

Expected Outcomes

Client/couple will:

- Discuss available options.
- Participate in decision-making process.
- Verbalize comfort with the decision taken.

Nursing Interventions

Interventions	*Rationales*
Determine couple's relationship to each other, strength of family bonds and support systems.	Establishes baseline. The threat of a hereditary disorder often results in family strife as well as marital disharmony, and family disintegration, such as separation and divorce.
Assess couple's perception of disorder as well as their attitudes and religious beliefs.	The way family members respond depends on the nature of the condition and on their perception of the burden (total amount of distress created by the diagnosis/birth of an affected baby). Reactions may be more negative in dominant, X-linked disorders where only one member of the couple is to "blame" or is identified as the carrier.
Provide information about risk estimates for disorder. Assess meaning of "carrier" status to client/couple, especially if couple desires male child and diagnosis is X-linked.	Allows couple to deal with the reality of the situation. For instance, if both parents are carriers of an autosomal recessive disorder for deleterious gene, the risk is 1 in 4 or 25%, that an offspring will be affected. The nature of the condition influences the family's responses. If female is a carrier for X-linked disorder, such as hemophilia, male fetuses have a 50% chance of being affected.
Provide opportunity to verbalize concerns. Be truthful and straightforward about genetic transmission.	Helps resolve feelings of guilt by exploring random nature of genetic transmission. Knowledge that everyone has 6–10 defective genes and that it is just chance that spouses have the same recessive deleterious gene may be reassuring/limit "blame".
Review all options available to the client/couple, such as adoption, postponing childbearing, artificial insemination by donor sperm or surrogate parenting. If client is carrying an affected child, options include terminating the pregnancy, treating the fetus in utero (if possible) and carrying the pregnancy to term and keeping the neonate.	Promotes communication between client and partner. Provides opportunity to begin decision-making process based on factual information.
Assist in problem solving and exploring alternative solutions, weighing impact on family members.	Helps to arrive at a decision, bringing family out of crisis, and facilitates growth of individual members.
Provide emotional support individually and to the couple as a unit.	Sometimes, it is helpful to talk to members of the couple individually as well as together to assess individual perceptions of situation. Separate meetings are particularly critical if the couples are not in agreement about possible solutions.
* Refer to social worker, counselor or support group (if available).	May be necessary to help solve the problem and prevent family disintegration. Situations may arise whereby the couple are unable to agree on a decision when aware of risks/potential outcomes.
* Refer to specialists after birth of an affected child	Assists in management of short- and long-term medical/psychologic problems.
* Denotes collaborative interventions	

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Evaluations for Expected Outcomes

- Client/couple discusses available options and participates in decision-making process.
- Client/couple verbalizes comfort with final decision.

Nursing Diagnosis

Spiritual Distress

Definition

Distress of the human spirit in which the individual experiences disruption in the life principle that pervades a person's entire being and that integrates and transcends one's biological and psychosocial nature.

Possible Etiological Factors / Related Factors

- Intense inner conflict about the outcome.
- Normal grieving for the loss of a perfect baby.
- Anger that is often directed at God.
- Religious beliefs and normal convictions.

Nursing Assessment

- Concerns and religious beliefs.
- Value systems.
- Vulnerability of client/couple.
- Emotional state.

Defining Characteristics

- Verbalization of inner conflict about beliefs.
- Questioning of the moral and ethical implications of therapeutic choices.
- Questioning the meaning of the event.
- Questioning regarding condition as punishment.
- Development of anger, hostility and crying.

Expected Outcomes

Client/couple will:

- Make informed choice regarding childbearing and/or childrearing within religious/moral framework.
- Verbalize acceptance/resolution of stress if prenatal diagnosis is positive.

Nursing Interventions

Interventions	*Rationales*
Assist in identifying and discussing concerns and analyzing options based on religious beliefs and value system.	Helps couple to recognize their dilemma. The decision regarding the pregnancy outcome and participation in prenatal diagnosis is ultimately the right and responsibility of the parents, and is dependent on ethical, religious, and moral convictions.
Analyze client's/couple's emotional state. Assess for normal or abnormal state of grieving.	Grieving is anticipated, but excessive denial may interfere with progression/resolution of grief work.
* Refer to pastor/priest/poojari or counselor as appropriate, especially if client/couple has religious affiliations that might affect decisions regarding conditions, such as Down syndrome or Tay-Sachs disease.	May assist in or be necessary for resolution of ethical issues. In certain religions, such as Catholicism, and orthodox Judaism, abortion is viewed as an attack on the sanctity of life, with the mother's endangerment being the only permissible reason, in which case permission from a higher church authority is required.
* Denotes collaborative interventions	

Evaluations for Expected Outcomes

- Client makes informed choice regarding childbearing/childrearing within their religious and moral framework.
- Client verbalizes acceptance and resolution of stress.

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Nursing Diagnosis

Low Self-esteem, Situational

Definition

Development of a negative perception of self-worth in response to genetic disorder of fetus/newborn.

Possible Etiological Factors / Related Factors

Perceived failure of a life event.

Nursing Assessment

- Perceptions of self.
- Expressions of feelings.
- Manifestations of grief.

Defining Characteristics

- Denial.
- Self-negating verbalization.
- Feelings of guilt, powerlessness and anger.
- Expressions of the unfairness of the situation.
- Projection of blame/responsibility for problem.

Expected Outcomes

Client/couple will:

- Verbalize lessened sense of guilt and restored feelings of self-worth.
- Set realistic goals.
- Participate actively in conflict resolution process.

Nursing Interventions

Interventions	*Rationales*
Determine self-perception of client/couple. Identify and reinforce individual/mutual strengths.	Provides opportunity for nurse to learn about client's/couple's expectations and perceptions of genetic disorder. Helps put feelings and opinions into perspective. When the diagnosis of genetic inheritance is involved, and one or both parents are found to be carriers of the defect, guilt, powerlessness, anger, and feelings of unfairness are normal reactions and to be anticipated.
Provide or reinforce information regarding risk estimate, discuss with client and couple together and separately. Reaffirm inability to control outcome of conception; affirm ability to control decision to reproduce, adopt, or remain childless.	Helps reduce guilt feelings, promotes recognition by couple that some control over outcome is possible.
Arrange for follow-up visit in clinic or office to assess status after abortion.	Feelings of loss, depression, and negativism often occur following discharge from the hospital, resulting in alteration in ego functioning.
* Assess for recurring unresolved conflicts.	Professional help may be required to resolve low self-esteem.
* Refer for counseling or psychiatric care as needed.	May be necessary to assist in working through the grief process in a healthy manner so that a positive sense of self emerges. Provides opportunity to establish or re-establish family integrity.
* Denotes collaborative interventions	

Evaluations for Expected Outcomes

- Client/couple verbalizes lessened sense of guilt and restored feelings of self-worth.
- Client/couple sets realistic goals and participates actively in conflict resolution process.

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Nursing Diagnosis

Risk for Injury, Related to Complications of Testing Procedures

Definition

At increased risk for injury as a result of the environmental conditions in testing procedures.

Possible Etiological Factors / Risk Factors

- Invasive procedure.
- Tissue hypoxia/changes in circulation.
- Teratogenic effects of radiation.

Nursing Assessment

- Specific tests planned/performed.
- Manifestations of complications.

Defining Characteristics

None, as this is a risk diagnosis.

Expected Outcomes

Client/couple will:

- Identify individual risks and ways of minimizing them.
- Be free of preventable complications.

Nursing Interventions

Interventions	*Rationales*
Identify the necessity of the specific test(s) and associated risks.	Testing involving radiation, CVS, amniocentesis and so forth carries with it some degree of risk (e.g. spontaneous abortion/preterm labor, bleeding, infection, rupture of membranes, direct fetal injury or loss. Information helps client/couple make informed decision.
Review indications for genetic counseling.	Some indications for the procedure are the previous birth of a child with a chromosomal disorder or known metabolic defects, any woman identified as a carrier of a sex-linked trait, any woman aged 35 or older, paternal age of 45 or older, family history of NTD, or both parents carrying an autosomal recessive disease or a chromosomal abnormality.
Review signs and symptoms of complications, such as premature labor, infection, and abruptio placentae.	Client needs to recognize these as being important to report to the healthcare provider.
* Assist with obtaining blood or tissue, placental or chromosomal biopsy.	In the event of a stillbirth or neonatal death or birth of an affected baby, nature of disorder is determined. This information is helpful in counseling couple regarding future pregnancies.
* Assist with procedures, such as amniocentesis, taking necessary measures (e.g. monitor blood pressure, pulse, respiration, and fetal heart rate). Assist fetal/uterine activity, use external fetal monitor for 20–30 minutes after procedure.	Testing procedures carry risks to the pregnancy/fetus. Careful monitoring may reduce untoward effects.
Position mother on left side.	Increases placental circulation, dilating blood vessels and reducing risk of supine hypotension and uterine irritability.
* Administer medications as indicated. Antibiotics.	Given prophylactically for 10 days after invasive procedures to prevent amnionitis.
Rh immune globulin (Rh Ig) after amniocentesis and CVS.	Protects Rh-negative client from sensitization. **Note:** CVS is contraindicated in an Rh-sensitized woman unless the purpose of the test is to detect an Rh-positive fetus.

** Denotes collaborative interventions*

Evaluations for Expected Outcomes

- Client/couple identifies individual risks and ways of minimizing them.
- Client/couple remains free of preventable complications.

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- Babies following assisted births and experiencing neonatal health problems
- Neonates who are preterm, post-term and with growth deviations.



Annamma Jacob MSc(N) completed her Degree in Nursing in 1966 and Masters in Nursing in 1976 from Christian Medical College and Hospital (CMCH), Vellore, Tamil Nadu, India. She has worked as Nurse Educator, Clinical Practitioner, Nurse Manager and Nursing College Principal in India, Middle East and United States of America. Other books authored are:

- *A Comprehensive Textbook of Midwifery and Gynecological Nursing*
- *Manual of Midwifery and Gynecological Nursing*
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