

# Genetic Counseling

Genetic counseling is a communication process that deals with human problems associated with the occurrence or risk of a genetic disorder in a family. Counseling involves genetic screening, whereby a high-risk or general population is analyzed to detect the presence of disease, and case finding for couples at potential risk based on medical/family histories. The process can be prospective (counseling delivered to a client/couple of reproductive age before conception or before the birth of an affected child), or it can be retrospective/postnatal (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.

(Refer also to care plan [CP]: The High-Risk Pregnancy, as appropriate.)

## CLIENT ASSESSMENT DATA BASE

### Circulation

Hypertension  
Bleeding

### Ego/Integrity

May express feelings of inadequacy

### Food/Fluid

Weight gain may be inappropriate for gestational stage (smaller gain may negatively affect fetus).  
Maternal insulin-dependent diabetes.  
Presence of eating disorders (e.g., anorexia nervosa, bulimia, or obesity).

### Safety

Infection (e.g., STDs, pelvic inflammatory disease)  
Presence of seizure disorder, degree/method of control  
Significant exposure to radiation, toxic chemicals, or infectious teratogens (e.g., rubella, toxoplasmosis, cytomegalovirus [CMV], HIV/AIDS), postnatal infections (e.g., meningitis, encephalitis); postnatal nutritional/stimulatory deprivation  
Breech presentation (especially with anencephaly)

### Sexuality

History of two or more first-trimester abortions, fetal demise, or previous child with chromosomal abnormality  
Birth trauma or identifiable genetically transmitted disorder  
Use of ovulation stimulant such as clomiphene (Clomid) or menotropins (Pergonal)

### Social Interaction

Interfamily marriage/sexual activity (consanguinity)  
Guilt/blame toward self and/or partner who carries defective gene

### Teaching/Learning

Positive family history/pedigree of known genetic or inherited disorders (e.g., sickle cell, cystic fibrosis, hemophilia, phenylketonuria, craniospinal defects, renal malformations, thalassemia, Huntington's chorea) familial disorders (cancer, heart disease, diabetes, allergies), congenital abnormalities (Down syndrome, mental retardation, neural tube defects), or inborn metabolic disorder (e.g., maple syrup urine disease, Tay-Sachs disease)  
Ethnic background at risk for specific disorder (e.g., black African, Mediterranean, Ashkenazi Jewish)  
Maternal age >35 years  
Drug usage (alcohol; over-the-counter [OTC], prescribed, or street drugs; anticonvulsant medication)

## DIAGNOSTIC STUDIES

**Chromosomal Analysis:** Using buccal smear/serum.

### Prenatal Diagnosis:

**Amniocentesis (at 14–16 wk):** Determines sex chromatin (rules out sex-linked disorders), karyotype (rules out chromosomal disorders), biochemical studies (rules out inborn errors of metabolism), elevation of AFP levels reflecting neural tube defects (NTDs).

**Ultrasonography (using real time) Preceding/During Amniocentesis:** Rules out missed abortion, visualizes term pregnancies, and helps to identify genetic/congenital problems. (Current research awaiting validation suggests a scan using a vaginal probe can be done as early as 10 weeks' gestation to screen for Down syndrome by looking for an accumulation of fluid in the neck of the fetus.)

**Serum AFP Levels (16–18 wk):** Identifies NTDs; if abnormality high/low, test is repeated.

**Triple-Screen (MSAFP<sub>3</sub>):** Maternal serum testing at about 16 weeks' gestation measures AFP, unconjugated estriol, and human chorionic gonadatropin (HCG) to detect problems such as open spinal or ventral wall defects, anencephaly, Down syndrome, and trisomy 18.

**Amniography/Fetography:** Identifies gross problems, such as major soft-tissue abnormalities, esophageal/duodenal atresia, and bone abnormalities, using special x-ray technique (may be potentially teratogenic).

**Fetoscopy (considered experimental, of limited availability):** Permits direct visual examination of the fetus and sampling of fetal blood/tissue samples to diagnose hereditary blood disorders (e.g., sickle cell anemia, hemophilia).

**Chorionic Villi Sampling (CVS) (at 8–10 wk):** May replace amniocentesis as a genetic diagnostic tool because it is performed earlier, and results are reported in 7–10 days, which is particularly beneficial if a first-trimester termination is being considered.

## 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

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**NURSING DIAGNOSIS:****Anxiety [specify level]****May Be Related To:**

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

**Possibly Evidenced By:**

Increased tension, apprehension, uncertainty, feelings of inadequacy, or expressed concern regarding changes in life events, insomnia

**DESIRED OUTCOMES/EVALUATION CRITERIA—CLIENT/COUPLE WILL:**

Acknowledge awareness of feelings of anxiety.

Verbalize realistic concerns related to process of genetic counseling/prenatal diagnosis.

Appear relaxed and report that anxiety is reduced to a manageable level.

Identify and use resources/support systems effectively.

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**ACTIONS/INTERVENTIONS****RATIONALE**

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**Independent**

Assess nature, source, and manifestations of anxiety.

Identifies specific areas of concern and determines direction for and possible options/interventions.

Provide information about specific genetic disorder, risks involved in reproduction, and available prenatal diagnostic measures/options. (Refer to nursing diagnosis [ND]: Knowledge deficit [Learning Need].)

May relieve anxiety associated with the unknown and assist family to cope with stress, make decisions, and adapt positively to choices. Note: A large number of clients at risk of producing a child with a genetic abnormality do not receive prospective counseling/diagnostic services before conception because of ineffective case finding/lack of awareness and often enter counseling, during the first trimester or, retrospectively, after the birth of an affected child. New genetic research at the gene level will have future implications for diagnosis, carrier status, or prenatal detection of genetic disease. Some of the techniques used include restriction endonuclease, DNA probes, polymerase chain reaction (PCR), Southern blot, restriction fragment length polymorphisms (RFLPs).

Promote ongoing sharing of concerns/feelings.

Opportunity for client/couple to begin resolution of situation. Note: Level of anxiety is usually higher in the couple who have already given birth to a child with a chromosomal disorder.

Review procedure and what to expect in terms of discomfort if fetus is affected and couple elects to terminate pregnancy and so on. (Refer to CP: Elective Termination.)

Client/couple may be extremely anxious, guilt-ridden during uncomfortable procedure; information can enhance coping, reduce anxiety.

Visit couple after procedure. Provide anticipatory guidance in terms of physical/psychological changes.

Provide opportunity for discussion of test results on fetus and assist with interpretation of information, especially following abortion.

Listen to expressions of concern/feelings about situation.

### **Collaborative**

Refer for further counseling (e.g., psychiatric, group).

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 to be affected, or when diagnosis is made after delivery. (Refer to CP: The Parents of a Child with Special Needs.)

After abortion for genetic indications, follow-up visit by the primary nurse may help to reduce couple's anxiety/depression.

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.

Anxiety may not be resolved sufficiently, necessitating additional professional assistance.

Helps to reduce anxieties regarding how the couple will meet their baby's special needs.

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#### **NURSING DIAGNOSIS:**

#### **May Be Related To:**

#### **Possibly Evidenced By:**

#### **DESIRED OUTCOMES/EVALUATION CRITERIA—CLIENT/COUPLE WILL:**

#### **Knowledge deficit [Learning Need], regarding purpose/process of genetic counseling**

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

Verbalization of the problem, statement of misconceptions, request for information

Describe the disorder in question.

Identify available options.

Discuss, in own words, purpose and findings of specific prenatal detection methods.

Estimate risks associated with reproduction.

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### **ACTIONS/INTERVENTIONS**

#### **Independent**

Discuss the purpose/goals of genetic counseling.

Obtain pertinent family history; create pedigree chart from information. Include both parents in interview.

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### **RATIONALE**

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.

Provides accurate picture of the proband (afflicted person/index case) in relation to other family members and helps identify other persons similarly affected. Ensures that both parents are provided with the same information.

Identify risk factors in obstetric history, such as maternal age, three or more miscarriages, fetal demise, hypertension, bleeding, infection, significant exposure to radiation, low-level exposure to environmental pollutants (e.g., lead, formaldehyde, anesthetic gases), drug use (OTC, prescribed, street), birth trauma, or identifiable genetically transmitted disorder.

Distinguish between environmental causes of mental retardation and those with a genetic component (e.g., Down syndrome).

Define specific genetic disorder in question. Provide prognosis/estimate of risks and probable consequences. Clarify/interpret data; avoid making recommendations or decisions for the client/couple.

Carry out ongoing family assessment for available support systems. Note signs of inappropriate coping/maladaptation. Offer information when family is ready to hear it.

Arrange for follow-up, telephone calls, visits to home/counseling service.

Provide information about readily available/individually appropriate diagnostic procedures and screenings, such as amniocentesis, serum testing for AFP, and ultrasonography, as well as CVS, fetoscopy, amniography, percutaneous umbilical blood sampling (PUBS) or cordocentesis, computed tomography (CT) scanning, and magnetic resonance imaging (MRI).

Permit couple to arrive at own decision regarding their participation in, or refusal of, prenatal testing procedures.

Review anticipated follow-up evaluative procedures, especially if client is currently pregnant.

Discuss time element associated with receiving test results.

These factors may produce a phenocopy, an imitation of a genetic disorder.

Environmental causes include birth trauma, chemical teratogens, infectious teratogens (e.g., rubella, toxoplasmosis, cytomegalovirus), postnatal infections (e.g., meningitis, encephalitis), and postnatal nutritional/stimulatory deprivation.

Determines the probability of risk and recurrence of risk, allows client/couple to make informed choices incorporating values/goals/personal circumstances, free of biases or recommendations from the nurse.

Negative coping may interfere with cognitive functioning and hinder educational process. Some families are not ready to listen immediately after a diagnosis is made and many do not listen effectively the first time information is presented.

Provides opportunity to reinforce information when family is "ready" to hear it.

Prenatal diagnosis permits preparation for, or offers an alternative to, having children with genetic disorders. Allows couple to make informed choices at their own pace, based on understanding of long-term problems associated with the disorder in question.

Although testing may assist in determining whether the fetus at risk for a genetic disorder is actually affected, the client/couple has the right to refuse prenatal evaluation. Their decision needs to be accepted and respected.

May include regular assessment of fetal heart tones, obtaining 20-min tracing on electronic monitoring, and so forth.

Prior knowledge of length of time required to obtain results can lessen client's anxiety. Test results may take up to 4 wk to analyze. Although CVS may be limited in its availability, it has advantages over amniocentesis because it can be performed earlier in the pregnancy (first versus second trimester), and results are available quickly (in 1–2 wk versus 2–4 wk). PUBS, or cordocentesis, provides a rapid fetal karyotype.

Encourage participation in community programs, i.e., education/support groups and groups concerned with specific genetic disorders as indicated (Tay-Sachs disease, Down syndrome, and so forth).

Obtain/coordinate referrals with specialty clinics or adoptive agencies that deal with specific genetic disorders (e.g., cystic fibrosis, hemophilia).

Obtain informed consent/surgical permit for procedures/tests. Review risks associated with procedure.

Discuss future options, such as preimplantation diagnosis when both partners are identified as carriers of a defective gene.

Helps client identify with others who have dealt with genetic disorders. Provides role models and information about the identified disorder.

May be needed to provide appropriate care/support that is needed in genetic counseling, especially if genetic component is involved.

Client needs to be aware of any risks involved to herself or the fetus, especially if the procedure is invasive, such as amniocentesis, CVS, fetoscopy, or PUBS.

Several eggs can be fertilized, the embryos evaluated for genetic abnormalities, and then only a healthy embryo is implanted in the uterus.

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**NURSING DIAGNOSIS:**

**Risk Factors May Include:**

**Possibly Evidenced by:**

**DESIRED OUTCOMES/EVALUATION CRITERIA—CLIENT/COUPLE WILL:**

Family Processes, risk for altered

Situational crisis, individual/family vulnerability, difficulty reaching agreement regarding decision

[Not applicable; presence of signs/symptoms establishes an *actual* diagnosis]

Discuss available options.

Participate in the decision-making process.

Verbalize comfort with final decision.

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**ACTIONS/INTERVENTIONS**

**RATIONALE**

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**Independent**

Determine couple's relationship to one another, strength of intrafamily bonds, and support systems.

Establishes baseline. The threat of a hereditary disorder often results in intrafamily strife, as well as marital disharmony and family disintegration (separation and divorce).

Assess couple's perception of disorder as well as their attitudes/religious beliefs. (Refer to ND: Spiritual Distress.)

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 child). Reactions may be more negative in dominant, X-linked disorders in which 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.

Provide opportunity to verbalize concerns. Be truthful and straightforward about genetic transmission.

Review all options available to the client/couple, e.g., adoption, postponing childbearing, artificial insemination by donor sperm, electing to reproduce using prenatal diagnosis, or surrogate parenting. If client is carrying an affected child, options include terminating the pregnancy; refusing aggressive intervention (e.g., not stopping premature labor, no CPR or intensive care); or treating the fetus in utero (if possible), carrying the pregnancy to term and keeping the neonate, or placing the child for adoption/foster care or institutionalization. (Refer to CP: The Parents of a Child with Special Needs.)

Assist in problem solving and exploring alternative solutions, weighing impact on family members.

Provide emotional support individually and to the couple as a unit.

## **Collaborative**

Refer to community support groups, counseling, or social services.

Refer to early intervention programs, community groups, or specialists after birth of an affected child.

Allows couple to deal with the reality of the situation. For instance, if both parents are carriers of an autosomal recessive disorder for a 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 the female is a carrier for an X-linked disorder such as hemophilia, male fetuses have a 50% risk of being affected.

Helps absolve 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.”

Promotes communication between client and partner. Provides opportunity to begin decision-making process based on factual information. Note: Choices may be limited by cultural/religious beliefs (e.g., Catholic, Christian Science, Jehovah’s Witnesses and Islamic religions restrict or prohibit abortion).

Helps to arrive at a decision, bringing family out of crisis, and facilitates growth of individual members and family as a unit.

Sometimes it is helpful to talk to members of the couple independently as well as together to assess individual perceptions of situation. Separate meetings are particularly critical if the couple are not in agreement about possible solutions.

May be necessary to help problem-solve and prevent family disintegration. Situations may arise whereby the couple are unable to agree on a decision when aware of risks/potential outcomes.

Assists in management of short- and long-term medical/psychological problems.

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**NURSING DIAGNOSIS:****May Be Related To:****Possibly Evidenced By:****DESIRED OUTCOMES/EVALUATION  
CRITERIA—CLIENT/COUPLE WILL:**

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**Spiritual Distress (distress of the human spirit)**

Intense inner conflict about the outcome, normal grieving for the loss of a perfect child, anger that is often directed at God, religious beliefs/moral convictions

Verbalization of inner conflict about beliefs, questioning of the moral and ethical implications of therapeutic choices, questioning the meaning of this event, regarding condition as punishment; the development of anger, hostility, and crying

Make informed choices regarding childbearing/childrearing within their religious/moral framework.

Verbalize acceptance/resolution of stress if prenatal diagnosis is positive.

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**ACTIONS/INTERVENTIONS**

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**RATIONALE****Independent**

Assist in identifying/discussing concerns and analyzing options based on religious beliefs and value system.

Analyze client's/couple's emotional state. Assess for normal or abnormal states of grieving.

Helps couple to recognize their dilemma. The decision regarding the pregnancy outcome and participation in prenatal diagnosis is ultimately the right/responsibility of the prospective parents, with informed decision making dependent on ethical/religious/moral convictions.

Grieving is anticipated, but excessive denial may interfere with progression/resolution of grief work.

**Collaborative**

Refer to pastor/rabbi/priest or other spiritual counselor as appropriate, especially if client/couple has religious affiliations that might affect decisions regarding conditions such as Tay-Sachs disease or Down syndrome.

May assist in, or be necessary for, resolution of ethical issues. In many religions, such as Catholicism, Orthodox Judaism, and Islam, abortion is viewed as an attack on the sanctity of life, with the mother's endangerment being the only generally permissible reason in some religions, in which case permission from a higher church authority is required.

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**NURSING DIAGNOSIS:****May Be Related To:****Possibly Evidenced By:**

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**Self Esteem disturbance**

Perceived failure at a life event

Denial, self-negating verbalization; feelings of guilt, powerlessness, anger; expressions of the unfairness of the situation, projection of blame/responsibility for problem

**DESIRED OUTCOMES/EVALUATION  
CRITERIA—CLIENT/COUPLE WILL:**

Verbalize lessened sense of guilt and restored feelings of self-worth.

Set realistic goals.

Participate actively in conflict resolution process.

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**ACTIONS/INTERVENTIONS**

**RATIONALE**

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**Independent**

Determine self-perception of client/couple.  
Identify and reinforce individual/mutual strengths.

Provides opportunity for nurse to learn about client's/couple's expectations/perceptions of genetic disorder. Helps put feelings/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/universal reactions to be anticipated.

Provide or reinforce information regarding risk estimate, to 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. Note: Some cultures believe in external locus of control and client/couple may resign themselves to the belief that they have no control over the events that are occurring.

Arrange for follow-up visit in clinic or office to assess status after abortion.

After discharge from the healthcare setting, feelings of loss/depression/negativism often occur, resulting in alteration of ego functioning.

Assess for recurring unresolved conflicts.

More intensive interventions support may be required to resolve low self-esteem.

**Collaborative**

Refer for counseling/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.

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**NURSING DIAGNOSIS:**

**Injury, risk for complications of testing procedures**

**Risk Factors May Include:**

Invasive procedure, tissue hypoxia/changes in circulation, teratogenic effects of radiation

**Possibly Evidenced By:**

[Not applicable; presence of signs/symptoms establishes an *actual* diagnosis]

**DESIRED OUTCOMES/EVALUATION  
CRITERIA—CLIENT/COUPLE WILL:**

Identify individual risks and ways of minimizing them.

Be free of preventable complications.

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## **ACTIONS/INTERVENTIONS**

## **RATIONALE**

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### **Independent**

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, i.e., spontaneous abortion/preterm labor, bleeding, infection, rupture of membranes, direct fetal injury/loss. Information helps the client/couple make informed decisions.

Review indications for genetic testing.

Some indications for this procedure are previous birth of a child with a chromosomal disorder or known metabolic defects, identification of client as a carrier of a sex-linked trait; maternal age of 35 or older/paternal age of 45 or older; family history of NTD; both parents carrying an autosomal recessive disease or a chromosomal abnormality; history of environmental teratogenic exposures, e.g., street drugs, dioxin/agent orange.

Review signs/symptoms of complications such as premature labor, infection, and abruptio placentae.

Client needs to recognize these as being important to report to healthcare provider to obtain timely evaluation/intervention.

### **Collaborative**

Assist with obtaining blood or tissue, placental or chromosomal analysis/biopsy.

Determines nature of disorder in the event of a stillbirth/neonatal death/birth of an affected child, which 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). Assess fetal/uterine activity; use external fetal monitor for 20–30 min after procedure.

Testing procedures carry risks to the pregnancy/fetus. Careful monitoring may reduce untoward effects.

Position mother on left or right side.

Increases placental circulation, dilating blood vessels and reducing risk of supine hypotension and uterine irritability.

Administer medications as indicated:  
Rh immune globulin (RhIg) 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.

Antibiotics.

Given prophylactically after fetoscopy to prevent amnionitis.

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**NURSING DIAGNOSIS:****Risk Factors May Include:****Possibly Evidenced By:****DESIRED OUTCOMES/EVALUATION  
CRITERIA—CLIENT COUPLE WILL:****Health Maintenance, risk for altered**

Ineffective coping methods, unresolved grief, disabling distress, lack of material resources.

[Not applicable; presence of signs/symptoms establishes an *actual* diagnosis]

Assume responsibility for health care needs.

Identify factors contributing to condition/situation.

Verbalize ability to cope adequately with the existing situation.

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**ACTIONS/INTERVENTIONS****RATIONALE**

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**Independent**

Assess level of dependence/independence and ability to make decisions to meet own needs.

Determine needs (e.g., communication skills, knowledge of resources, motivation).

Initiate/participate in case finding/referral of individuals with real or potential genetic concerns.

Encourage participation in group activities, such as diagnosis-related support groups, March of Dimes, or Easter Seals.

May need more help to gain information necessary for problem resolution.

Helps determine extent and type of interventions necessary.

Helps reduce the number of children born with genetic defects.

Can assist client/family in dealing with own situation.