MRS.KAVITHA

ASSO.PROF

ICON

GENETICS

Meaning

The term genetics was introduced by Bateson in 1906. It was derived from Greek word gene'- "to become" or to grow into'. Therefore, Genetics is the science of coming into being

Definition

Genetics is that branch of biological sciences which deals with the transmission of characteristics from parent to offspring"

Prenatal development begins at the cellular level through a process referred to as fusion when the male germ cell, the sperm, unites with the female germ cell, the ovum to create a new cell referred to as a zygote. The zygote contains the base genetic prototype from which all the remaining cells will develop.

The developing cellular process is divided into two processes:

1. Meiosis - cellular divisions occurring prior to fertilization. This process provides for random combinations of genetic materials from each parent through production of haploid germ cells. Haploid germ cells contain half the number of chromosomes found in other body cells.

2. Mitosis - cellular divisions occurring after fertilization that create the ongoing growth and development of the organism. This process creates the diploid cells containing a complete chromosome number. The diploid cells are

the result of a combination of one maternal X chromosome with one paternal X or Y chromosome to create either an a) XX female or b) XY male.

TERATOGENESIS

Teratogenesis refers to the production of defects in the fetus. A Teratogenic agent is responsible for producing such a defect. The term teratogen usually is cited in the context of causing anatomical defects in an embryo that was previously differentiating normally. The time during embryogenesis when the fetus is exposed to a potential Teratogens is crucia. The capability of formation of congenital anomalies in fetus is known as teratogenicity

Teratogens include radiation, chemicals (drugs), Stressors, malnutrition and infectious agents. This chapter reviews principles of teratology and discusses drug use in pregnancy.

Harmful substances such as drugs or radiation that invade the womb and result in birth defects are called Teratogens. Teratogens are especially damaging in the embryonic stage because it is a critical period in prenatal development. Later, during the fetal stage, the environment provided by the mother affects the baby's size, behavior, intelligence and health, rather than the formation of organs and limbs.

DRUGS AND BIRTH DEFECTS

The clinical consequences of drug teratogens must be placed in the overall context of developmental defects in humans. Defects may be from genetic, environmental, or unknown causes. Approximately 25% are known to be genetic in origin (e.g., Mendelian, chromosomal). Approximately 65% of

defects are of ostensibly unknown etiology but probably reflect combinations of genetic and environmental factors.(polygenic/multifactorial).

The risk for malformation after exposure to a drug must be compared with the background rate, which for major malformations in the general population usually is cited as 2-3%. A major malformation is defined as one that is incompatible with survival, such as anencephaly; or one requiring major surgery for correction.

VARIABLES AFFECTING TERATOGENESIS

Specificity of Agent

Some agents are more teratogenic than others. Less obvious is the axiom that an agent may be teratogenic in only certain species. For example, thalidomide produces phocomelia in primates but not in rodents. Within a given species, however, a given teratogen may affect many organ systems. Some organ systems are preferentially affected, but the pattern of anomalies also reflects the organ systems differentiating at the time the agent was administered. For example, administering thalidomide between days 35 and 37 causes ear malformations administering the agent between days 41 and 44 causes amelia or phocomelia

Dosage

Although high doses of a proven teratogen usually are more deleterious than low doses, this is not always true. At any given time, an embryo can respond to a teratogen in one of three ways: (1) at a low dose, there is no effect: 2) at an intermediate dose, a pattern of organ-specific malformations can result; and (3) at a high dose, the embryo may be killed, causing the organ-specific teratogenic action to go unrecognized.

Timing

The effect of a teratogen on the developing organism depends on what period in the pregnancy (in development) the child is exposed to the teratogen. some teratogens cause damage only during specific days of weeks in early pregnancy other teratogens are harmful at any time during the pregnancy--for example, for behavioral teratogens. There is no safe period---the brain and nervous system can be harmed throughout the pregnancy.

Genotype

The genotype of the mother and the fetus influences the efficacy of a teratogen. For example, genotype determines the prevalence of cleft palate in inbred strains of mice whose mothers are administered cortisol during pregnancy.

Drug Interactions

Simultaneous administration of two(Drugs) teratogens may produce a different effect from that existing when the two are administered separately

Other Factors

Variability in teratogenic response sometimes is associated with other environmental or morphologic factors: maternal or fetal weight, in utero position of the fetus, proximity to other affected litter mates, uterine vasculature, and diet.

GENETIC COUNSELLING

INTRODUCTION:

Rapid expansion in the identification, understanding and diagnosis of genetic disease has been accompanied by effective medical and surgical therapies in a small number of cases. For the majority of genetic conditions, therapeutic or preventive measures are nonexistent or disappointed limited. Consequently, the most useful means of reducing the incidence of these disorders is by preventing their transmission. With the accumulation of knowledge about genetic disorders, the probability of recurrence can be predicted with increased accuracy. At present the best means for reducing the number of children born with genetic defects is for health professionals to provide families with genetic information and services.

DEFINITION:

Genetic counseling can be defined as a process of communication and education that addresses concerns relation to the development and or transmissions of hereditary disorders.

The different etiologic factors for fetal malformations are:

1. Chromosomal abnormalities (numeric or structural)
2. Single gene disorders (cystic fibrosis)—1%
3. Polygenic or multi factorial disorders
4. Teratogenic disorders due to exposure of exogenous factors (drugs).

SCREENING METHOD

First Trimester Screening

Screening parameters are:

(A) Biophysical: (i) ultrasound measurement of nuchal translucency (NT),

(ii) Nasal bone,

(B) Biochemical: (i) free β-hCG, (ii) PAPP-A (Pregnancy Associated Plasma Protein-A).

Time of Test: Between 11 weeks and 14 weeks.

Values: PAPP-A—reduced; β-hCG—increased; NT—measurement increased in trisomy 21.

Second Trimester Screening:

It is done between 15 weeks and 22 weeks.

MSAFP: This test is done between 15 weeks and 20 weeks. MSAFP value of 2.5 multiples of the median (MOM) when adjusted with maternal weight and ethnicity is taken as cut-off point. Elevated MSAFP detects 85% of all neural tube defects. Cases with such high values are considered for high resolution ultrasound imaging and/or amniocentesis. Very low MSAFP levels are associated with increased rates of miscarriage, stillbirth and neonatal death.

Triple Test: It is a combined biochemical test which includes MSAFP, hCG and uE3 (unconjugated estriol).

Maternal age in relation to confirmed gestation age is also taken into account. It is used for detection of Down's syndrome. In an affected pregnancy, levels of MSAFP and uE3 tend to be low while that of hCG is high. It is performed at 15–22 weeks. It gives a risk ratio and for confirmation CVS/amniocentesis has to be done. The result is considered to be screen positive if the risk ratio is 1:250 or greater.

Quadruple (Quad) Screening includes four biochemical analytes:

(1) Maternal Serum Alpha Fetoprotein (MSAFP),

(2) Unconjugated estriol (uE3),

(3) dimeric inhibin-A and (4) hCG.

Quadscreen can detect trisomy 21 in 85% of cases with a false-positive rate of 0.9%. Levels of serum analytes in cases with trisomy 21: hCG—increased; uE3—reduced; inhibin A—elevated; MSAFP—reduced.

Adjustments are to be made for maternal age, weight and ethnic group.

Best screening procedure is combined first and second trimester procedures (ACOG).

	CHORIONIC VILLUS SAMPLING	**AMNIOCENTESIS**	**CORDOCENTESIS**
Time	Transcervical 10–13 weeks, Transabdominal 10 weeks to term	After 15 weeks (early 12–14 weeks)	18–20 weeks
Materials for study	Trophoblast cells	Fetal fibroblasts Fluid for biochemical study	Fetal white blood cells (others—infection and biochemical study)
Karyotype	result Direct preparation: 24–48 hours. Culture: 10–14 days	Culture: 3–4 weeks	Culture: 24–48 hours
Fetal loss	0.5–1%	0.5%	1–2%
Accuracy	Accurate; may need amniocentesis for confirmation	Highly accurate	Highly accurate
Termination of pregnancy when indicated	1st trimester—safe	2nd trimester—risky	2nd trimester–risky
Maternal	Very little	More traumatic;	Same as

effects following termination of pregnancy		physically and psychologically	amniocentesis

The Nature and Characteristics of Genetic Counseling

Genetic counseling has been defined as a communication process which deals with the human problems associated with the occurrence, or the risk of occurrence, of a genetic disorder in a family. This process involves an attempt by one or more appropriately trained persons to help the individual or family

(1) Comprehensive medical facts, including the diagnosis, the probable course of the disorder and the available management

(2) Appreciate in specified relatives

(3) Understand the options for dealing with the risk of recurrence

(4) Choose the course of action which seems appropriate to them in view of their risk and family goals and act in accordance with that decision

(5) Make the best possible adjustment to the disorder in an affected family member and/or to the risk of recurrence of that disorder.

BENEFICIARIES OF GENETIC COUNSELING:

- ✓ A couple who has a child with a congenital abnormality or inborn errors of metabolism, any genetic condition, any development delay or mental retardation.
- ✓ A couple whose close relatives have a child with genetic disorder, including those with a individual who is a known balanced translocation carrier.
- ✓ A couple that is consanguineous (closely related).
- ✓ Any women over the 35 years of age and man over 45 years of age.
- ✓ Couples of ethnic backgrounds in which specific illness are known to occur.
- ✓ Women who had three or more miscarriage.
- ✓ A person whose doctor or health care provider has recommended a genetic evaluation or genetic testing.

- ✓ Prenatal diagnosis of any genetic disorders.

PRINCIPLE OF GENETIC COUNSELING:

1. It is a therapeutic measures
2. It includes establishment of
3. accurate diagnosis, treatment of the affected individuals as well as the prevention of the occurrence of genetic disorder.
4. It requires a special aptitude of communication.
5. It must be non-directive.
6. The final decision is on the consultant.
7. Both the parents should be counseled together as far as possible.
8. Follow up session is always desirable.

TYPES OF GENETIC COUNSELING:

1. Prospective genetic counseling: usually premarital, especially when the couples are related. This allows true prevention of disease.
2. Retrospective genetic counseling: there is a way a history of either an offspring or relative being affected. At present this counseling is mostly using and this is the hereditary disorder has already occurred in the family.
3. Expanded family genetic counseling: usually given in chromosomal abbreations occurring secondary to balanced translocation or inversion and in x-linked condition.

STEPS OF GENETIC COUNSELING:

1. **Diagnosis:** the goal evaluation is to make a diagnosis of particular condition or syndrome
2. **Prognosis:** the next step after diagnosis is for a family to understand about the disease. The genetic counselor will explain how the condition has affected other people with same diagnosis. What kind of symptoms or problems may faced by the family.

3. **Treatment:** all the available treatment for genetic disease are explained and recommended.
4. **Inheritance and recurrence risks**: the genetic or the genetic counselor will be able to answer the questions of family member bout chances of passing this conditions.
5. **Genetic testing:** genetic counselor explains ad recommend various testing for the couples affected with genetic disorder.

NURSES ROLE IN GENETIC COUNSELING:

- **Preparation for Counseling**
  The initial interview or intake visit is often conducted ducted by a nurse. At this time a detailed family history is obtained including information about the social aspects and the meaning that disorder has for this particular person or family. The family is told what to expect during counseling process, procedures to be performed, and the personnel with whom they will be involved. The interview serves as an opportunity to assess the client's or family's needs and reduce anxiety. Most people who come for genetic counseling are nervous and apprehensive because they are not only concerned about a genetic condition but also know that the outcome of the counseling and the decisions made may significantly alter their lives, Therefore, a distraction-free atmosphere should be provided. This may require care for small children whose restlessness and behavior can divert attention from what is being said. An ample amount of time is allotted to provide the family with as much information as possible.

- **Preconception counseling:** ideally couples that have the potential to transfer genetic disorder to their offspring should be counseled before pregnancy occurs. When the pre-conception counseling is possible, the nurse should have a relaxed interview with the couples to discuss the issues related risk.
- **Prenatal counseling:** In prenatal counseling the nurse has to identify women or couples who are at risk or having the potentials for congenital disorder in their offspring.
- **Follow up care:** maintaining follow up care with the family after genetic counseling or therapy is one of the most important nurse responsibilities.

BIBLIOGRAPHY:

- Nima Bhaskar, “Midwifery and obstetrical nursing” second edition, EMMESS publications 2015. page no:95-108, 658-659.
- B.T. Basavanthappa, “essentials of midwifery and obstetrical nursing, Jaypee brothers medical publishers, 2011. Page no: 386-389.
- DC Dutta’s textbook of obstetrics including perinatology and contraception, Eighth edition, Jaypee brothers medical publishers, 2015. Page no: 102-106.

CLASS TEACHING ON GENETIC COUNSELING

Submitted To

Submitted By

Mrs. Jamuna rani
HOD,
Dept of OBG Nursing.
ICON.

P. RAJAM
M.Sc Nursing
I year
ICON

Submitted on:

NURSES ROLE IN GENETIC COUNSELING:

- **Preparation for Counseling**
  The initial interview or intake visit is often conducted ducted by a nurse. At this time a detailed family history is obtained including information about the social aspects and the meaning that disorder has for this particular person or family. The family is told what to expect during counseling process, procedures to be performed, and the personnel with whom they well be involved. The interview serves as an opportunity to assess the client's or family's needs and reduce anxiety. Most people who come for genetic counseling are nervous and apprehensive because they are not only concerned about a genetic condition but also know that the outcome of the counseling and the decisions made may significantly alter their lives, Therefore, a distraction-free atmosphere should be provided. This may require care for small children whose restlessness and behavior can divert attention from what is being said. An ample amount of time is allotted to provide the family with as much information as possible.

- **Preconception counseling:** ideally couples that have the potential to transfer genetic disorder to their offspring should be counseled before pregnancy occurs. When the pre-conception counseling is possible, the nurse should have a relaxed interview with the couples to discuss the issues related risk.
- **Prenatal counseling:** I prenatal counseling the nurse has to identify women or couples who are at risk or having the potentials for congenital disorder in their offspring.
- **Follow up care:** maintaining follow up care with the family after genetic counseling or therapy is one of the most important nurse responsibilities.

NURSES ROLE IN GENETIC COUNSELING:

- **Preparation for Counseling**
  The initial interview or intake visit is often conducted ducted by a nurse. At this time a detailed family history is obtained including information about the social aspects and the meaning that disorder has for this particular person or family. The family is told what to expect during counseling process, procedures to be performed, and the personnel with whom they well be involved. The interview serves as an opportunity to assess the client's or family's needs and reduce anxiety. Most people who come for genetic counseling are nervous and apprehensive because they are not only concerned about a genetic condition but also know that the outcome of the counseling and the decisions made may significantly alter their lives, Therefore, a distraction-free atmosphere should be provided. This may require care for small children whose restlessness and behavior can divert attention from what is being said. An ample amount of time is allotted to provide the family with as much information as possible.

- **Preconception counseling:** ideally couples that have the potential to transfer genetic disorder to their offspring should be counseled before pregnancy occurs. When the pre-conception counseling is possible, the nurse should have a relaxed interview with the couples to discuss the issues related risk.
- **Prenatal counseling:** I prenatal counseling the nurse has to identify women or couples who are at risk or having the potentials for congenital disorder in their offspring.
- **Follow up care:** maintaining follow up care with the family after genetic counseling or therapy is one of the most important nurse responsibilities.

NURSES ROLE IN GENETIC COUNSELING:

- **Preparation for Counseling**
  The initial interview or intake visit is often conducted ducted by a nurse. At this time a detailed family history is obtained including information about the social aspects and the meaning that disorder has for this particular person or family. The family is told what to expect during counseling process, procedures to be performed, and the personnel with whom they well be involved. The interview serves as an opportunity to assess the client's or family's needs and reduce anxiety. Most people who come for genetic counseling are nervous and apprehensive because they are not only concerned about a genetic condition but also know that the outcome of the counseling and the decisions made may significantly alter their lives, Therefore, a distraction-free atmosphere should be provided. This may require care for small children whose restlessness and behavior can divert attention from what is being said. An ample amount of time is allotted to provide the family with as much information as possible.

- **Preconception counseling:** ideally couples that have the potential to transfer genetic disorder to their offspring should be counseled before pregnancy occurs. When the pre-conception counseling is possible, the nurse should have a relaxed interview with the couples to discuss the issues related risk.
- **Prenatal counseling:** I prenatal counseling the nurse has to identify women or couples who are at risk or having the potentials for congenital disorder in their offspring.
- **Follow up care:** maintaining follow up care with the family after genetic counseling or therapy is one of the most important nurse responsibilities.