Genetics & Its Practical Application in Nursing

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Abstract: -Genetics is of increasing importance in health care, as more is now known about the basic facts of inheritance. This article written introduces the general principles of genetics and its practical application in nursing and links these to real examples to allow nurses to apply this knowledge in their routine practice. Whatever may be the area of nursing, weather neonatology, intensive care or geriatrics it will be useful for them in their practices. As genetics gains greater predictive power & becomes increasingly incorporated in daily health care, it will be more imperative that all nurses have a strong genetic basis.

Keywords: Genetics, Nursing, Genetics Practical application

Introduction:-

• Nearly all diseases are now recognised to have a genetic component. The past several years have transformed genetic nursing practice from hidden speciality with a visible contribution to the genetic & overall health of individuals & families.

• The recent development of commercial testing for susceptibility genes (e.g. predisposition genes for breast, ovarian cancer syndrome & colon cancer) has had a great impact of nursing’s role in the identification & management of individuals at risk for developing many diseases. This development has led to tremendous changes in genetic nursing practice. As a result of this, the scope of genetic knowledge application in nursing is limitless.

• According to Forsman, “Nursing can ignore genetics no longer. The time for meaningful action is now.”

• In 1865 Johann Gregor Mendal was the first to describe the elements of hereditary genes.

• Scientific discoveries during the last several decades have provided more information about how genes function & how they contribute to human health & disease.

• Currently more than 10,371 identified genetic disorders are known to be inherited in a predictable pattern in families.

• With the knowledge of genetics, nurses can collect appropriate family information, provide current & appropriate information & support patients, families & communities as they integrate this new information & technology into their daily lives.

Concept of Genetics

The term ‘Genetics’ was first introduced by Bateson in 1906. It has been derived from the Greek word ‘Gene’, which means ‘to become’ or ‘to grow into’. Therefore, genetics is the science of coming into being.

Genetics is that branch of biological sciences which deals with the transmission of characteristics from parents to off-spring. Medical genetics has focused on the inheritance of hereditary disorders affecting only a small portion of the population.

Genetic services have been primarily associated with prenatal genetic counselling, identification of paediatric disorders associated with birth defects & dysmorphology, & in some cases rare adult onset single gene disorder.

Recent genetic & technological advances are helping us to better understand how genetic changes impact human variation as well as the development of cancer, Alzheimer’s disease, diabetes & other multifactorial diseases that are prevalent in adults.

Characteristics & Structure of Genes:-

• The term ‘Gene’ was introduced in 1909. Prior to him Mendel had used the word factor for a specific, distinct, particular unit of inheritance that takes part in expression of a trait.

• Presently, a gene is defined as a unit of inheritance composed of a segment of DNA or chromosome situated at a specific locus (gene/locus), which carries coded information associated with a specific function & can undergo crossing over as well as mutation.
Specific Features of Genes:-

- The term gene is often used to refer genetic material on a chromosome that code for a trait. E.g. one person has a gene for hair colour.
- It is a unit of recombination or capable of undergoing crossover.
- A unit of genetic material, which can undergo mutation.
- A unit of hereditary connected with somatic structure or function that leads to a phenotype expression.
- A gene is the basic physical & functional unit of hereditary.
- Gene which are made up of DNA, act as RNA instructor to make molecules called proteins.
- Every person has two copies of each gene, one inherited from each parent. Most genes are the same in all people, but a small number of genes less than (1% of the total) are slightly different between people. Alleles are forms of the same gene with small differences in their sequence of DNA bases. These small differences contribute to each person’s unique physical features.
- The physical development & phenotype of a person can be thought of as a product of genes interacting with each other & with environment.
- Total set of genes in a person are known as Genome.
- Gene is basically an instruction for human body, each gene has a specific purpose & very single function of the human body is coded in one or more genes.
- A person’s unique genetic constitutes called the Genotype are made up of about 30,000 to 40,000 genes
- A person’s phenotype, the observable characteristics of his or her genotype includes the physical appearance & other biological, physiological & molecular traits.

Functions of Genes:-

- Genes are components of genetic material & are thus unit of inheritance.
- They control the morphology or phenotype of individual.
- Replication of genes is essential for cell division.
- Genes carry the hereditary information from one generation to next.
- They control the structure & metabolism of the body.
- Reshuffling of genes at the time of sexual reproduction produce variation.
- Different linkages are produced due to crossing over.
- Genes undergo mutation & change their expression.
- New genes are consequently new traits develop due to reshuffling of different parts of genes.
- Genes change their expression due to position effect.
- Differentiation or formation of different type of cells, tissues & organs in various parts of the body is controlled by expression of others.
- Development or production of different stages in the life history is controlled by genes.

What is a chromosome?

- In the nucleus of each cell, the DNA molecule is packed into a thread-like structure called Chromosome.
- Each chromosome is made up of thread-like DNA tightly coiled many times around proteins called Histones.
- Chromosomes are packaged by proteins into a condensed structure called Chromatin.
- Each chromosome has a constriction point called centromere, which divides the chromosome into two section or “arms”, the short arm of the chromosome is labelled the “p-arm”, the long chromosome is labelled the “q-arm”.
- The location of the centromere on each chromosome gives the chromosome its characteristic stage, & can be used to help describe the location of specific genes.
- In humans each cell normally contains 23 pairs of chromosomes, for a total of 46.
- 22 of these pairs, called autosomes, look the same in both males & females.
- The 23rd pair, the sex chromosomes differs, between males & females.
- Females have two copies of the ‘X’- chromosome while males have one ‘X’ & one ‘Y’ chromosome.
Basic types of chromosomes

1. **Telocentric**: telocentric chromosomes have the centromere at the very end of the chromosome. Humans do not possess telocentric chromosomes but they are found in other species such as mice.

2. **Acrocentric**: acrocentric chromosomes have a centromere which is severely offset from the centre leading to one very long and one very short section human chromosome 13, 14, 15, 21 and 22 are acrocentric.

3. **Sub metacentric**: the centromere is displaced towards one end, creating a long arm and a short arm. Human chromosomes 4 to 12 are sub metacentric.

4. **Metacentric**: the centromere is located approximately in the middle, & so the chromosome has two arms of equal length. Human chromosome 1 & 3 are metacentric.

Functions of chromosomes

- Chromosomes contain genes. All the hereditary information is located in the genes.
- Chromosomes control the synthesis of structural proteins & thus help in cell division & cell growth.
- They control the cellular differentiation.
- By directing the synthesis of particular enzymes, chromosome control cell metabolism.
- Chromosome can replicate them or produce their carbon copies for passage to daughter cells & next generation.
- Some chromosome called sex chromosomes (e.g. X & Y) determines the sex of the individual.
- Mutations are produced due to change in gene chemistry.

Chromosomal Aberrations (chromosomal Mutation)

1. Chromosomal aberrations are disruptions in the normal chromosomal content of a cell, and are a major cause of genetic conditions in humans.

2. Change in **number** of chromosomes is known as **aneuploidy or numerical aberrations**.

3. Change in **arrangement** of genes in the chromosomes is known as **structural aberration**.

4. Chromosomal aberrations may involve **changes in single chromosome**, known as **intra-chromosomal aberrations**.

5. Chromosomal aberrations may involve **changes in two chromosomes** known as **inter-chromosomal aberrations**

6. All these are termed as chromosomal abnormalities.

7. Some chromosomal abnormalities do not cause disease in carriers, such as translocation, or chromosomal inversions, although they may lead to a higher chance of having a child with chromosomal disorder.

8. Abnormal numbers of chromosomes or chromosomal sets, aneuploidy, may be lethal or give rise to genetic disorders.

9. The three major singlechromosomemutations (intra-chromosomal Aberration):

   1. **Deletion**: - loss of part of chromosome. E.g. deletion of short arm of chromosome 5 Cri du chats.

   2. **Duplication**: - extra copies of a part of a chromosome. E.g. duplication of certain segment of chromosome 22 resulting in anal atresia and various congenital malformations.

   3. **Inversion**: - reverse the direction of a part of the chromosome. For example, about 40% are chromosome 9. May or may not result in visible effects.

The two major two-chromosomal mutations; (inter-chromosomal aberration)

1. Insertion or ring chromosome. For e.g., ring chromosome 14 is associated with psychomotor delay, mental retardation &dysmorphic craniofacial features. It is very rare.

2. Translocation: - part of a chromosome breaks off and attaches to another chromosome. In balanced translocation no genetic material added or lost. Balanced reciprocal translocation usually does not cause problems. For e.g., translocation trisomy 21 or down syndrome may result from the presence of 46 chromosomes that include a translocation chromosome.

Mechanisms of inheritance

- The basic features of mechanism of inheritance are as follows:-

1. Genes or chromosomes are physical basis of inheritance.
2. Person inherits half of the genetic information from each parent.
3. Every parent has two copies of genes; each parent contributes one copy of gene to their offspring.
4. Genes for different traits are inherited separately from one another. For example, the gene for hair colour is not linked with the gene for height. A child may have his mother’s hair colour but may not her height. For the most part, each trait is inherited separately.

Patterns & Mechanisms of Inheritance in Humans
1. Pedigree
2. Patterns of inheritance
   - Mendelian patterns of inheritance
     - Autosomal dominant
     - Autosomal recessive
     - Sex linked inheritance
   » X-linked dominant
   » X-linked recessive
   » Y-linked (Holandric) inheritance
   - Non-Mendelian pattern of inheritance
     - Co-dominant pattern of inheritance
     - Mitochondrial pattern of inheritance
     - Multifactorial pattern of inheritance.

Pedigree
- An important tool used by geneticist to study human inheritance in the pedigree.
- A pedigree is a pictorial representative of a family history; a family tree that outline the inheritance of characteristics.

Mendelian pattern of Inheritance
1. Autosomal Dominant: - one mutated copy of the gene in each cell is sufficient for a person to be affected by an autosomal dominant disorder. **Autosomal dominant disorder tends to occur in every generation of an affected family.**
   - Examples are: Huntington disease, colon cancer, hereditary breast & ovarian cancer.
2. Autosomal recessive: -
   - Two mutated copies of the genes are present in each cell when a person has an autosomal recessive disorder. An affected person usually has unaffected parents who carry a single copy of the mutated gene (and are referred to as carriers).
   - Examples: cystic fibrosis, sickle cell anaemia, thalassemia
3. X-linked Dominant: -
   - X-linked dominant disorders are caused by mutations in genes on the X chromosome.
   - Females are more frequently affected than males, and the chance of passing on an X-linked dominant disorder differs between men & women. Families with an X-linked dominant disorder often have both affected males & affected females in each generation.
   - A striking characteristic of X-linked inheritance is that fathers cannot pass X-linked traits to their sons (no male-to-male transmission).
4. X-linked recessive: -
   - X-linked recessive disorders are also caused by mutations in genes on the X chromosome. Males are more frequently affected than females, & the chance of passing on the disorder differs between men & women.
   - Families with an X-linked recessive disorder often have affected males, but rarely affected females, in each generation.
   - A striking characteristic of X-linked inheritance is that fathers cannot pass X-linked traits to their sons (no male-to-male transmission).
5. Y-linked (Holandric) Inheritance: -
   - Most Y-linked genes manifest their effect with one copy & show male-to-male transmission exclusively.
   - All the sons of an affected male would eventually develop trait.
   - Although the age at which they develop do varies.

Non-Mendelian pattern of Inheritance
1. Codominant pattern of inheritance: -
   - In codominant inheritance, two different versions (alleles) of a gene can be expressed, and each version makes a slightly different protein.
Both alleles influence the genetic trait or determine the characteristics of the genetic condition.

Example: ABO blood group

2. Mitochondrial Pattern of Inheritance:-

This type of inheritance, also known as maternal inheritance, applies to genes in mitochondrial DNA. Mitochondria, which are structures in each cell that convert molecules into energy, each contain a small amount of DNA.

Because only egg cell contributes mitochondria to the developing embryo, only females can pass on mitochondrial conditions to their children.

Mitochondrial disorders can appear in every generation of a family & can affect both males & females, but fathers do not pass mitochondrial traits to their children.

3. Multifactorial Pattern of Inheritance:-

This is a common cause of many birth defects as well as common adult onset conditions such as diabetes, heart disease & cancer.

Multifactorial inheritance conditions are believed to be the result of multiple mutations & environmental influence that combine to cause birth defects or disease.

Genetic conditions with a multifactorial cause tend to cluster in families but do not follow the characteristic pattern of inheritance seen with single gene disorder.

Examples: congenital heart disease, cleft lip/palate, neural tube defect, congenital hip dislocation, diabetes and high blood pressure.

Practical Application of Genetics in Nursing

a. All nurses have a role in the delivery of genetic services & management of genetic information.

b. Nurses require genetic knowledge to identify, support, refer & care for persons affected by or at risk for genetic disorders.

c. Nurses can offer care that protects patients & families from the risk associated with genetic information, including addressing family issues.

d. Nurses are also needed to refer patients to genetic specialist & assist in making choice of genetic health care.

e. Genetic nursing is practiced in different environments such as maternity, paediatrics, medical-surgical, psychiatric & community health nursing.

f. Genetic nursing is a holistic practice that includes assessing, planning, implementing & evaluating the physical, spiritual, ethical & psychosocial aspects of patients & families who have genetic concerns.

Genetic Nursing includes the following:-

I. Client & family assessment to identify genetic risk factors. In assessment takes detailed family history & construct a pedigree, analyse the assessment data. In addition, interpret information collected.

II. Planning & implementation of care during diagnosis & management of genetic disorders. In care, provide genetic education & develop & carry out a plan of care to address genetic concerns.

III. Information, counselling & support services to persons affected by or at risk for genetic disorders.

IV. Meeting referral needs

V. Long-term follow up.

Major Practical Applications of Genetic in Nursing

I. Understands genetic basis of disease

II. Early and effective diagnosis of genetic disorders.

III. Contributes towards health promotion with genetic aspects

IV. Prevention of genetic conditions

V. Management & care in genetic disorders

VI. Genetic information & counselling.

VII. Referral services

VIII. Social & ethical issues in genetics

Understands genetic basis of disease:

- With knowledge of genetics, nurses will understand that a large proportion of total disease have genetic basis. In addition will learn about:
  
  - Role of different genes in causation of genetic disorders & defects.
  
  - Good or bad genes for health-illness continuum

  - Normal & abnormal cell division & its genetic regulation.

  - Mechanism of disease inheritance from generation to next generation.

  - Genetic factors are playing role in an individual’s health.

  - Basic mechanisms of inheritance & transmission of chromosomes & genes,
including the concept of variation & mutation.

- Genetic contribution towards different diseases, disorders & defects.
- Genetic contributions to common & complex conditions such as breast cancer, colorectal cancer, heart disease & hypercholesterolemia, mental illness certain behavioural traits & Alzheimer's disease.

Early & effective diagnosis of genetic disorders

- Genetic knowledge of nurses will equip them with;
- Information about genetic risk, genetic testing & screening, & the implications, both positive & negative results.
- Interpretation of the results genetic tests.
- Interpretation of genetic risk
- Awareness of the possibility of an inherited or genetic component for a client condition & knowledge of cardinal features of familial predisposition such as early age of disease onset, multiple family members with the same diagnosis & predisposing risk factors.

Contributes towards health promotion with genetic aspects

- Learning about genetics nurses will enhance their understanding about:
  - Relationship of health & disease in relation to genetics, including how genetics & the environment interacts & how genes interacts with genes.
  - Healthy prenatal environment will ensure minimal risk of genetic defects among newborns.
  - Environmental interaction of an individual is an important factor in reference to gene or chromosomal mutation, which may have positive or negative impact on health of an individual.

Prevention of Genetic Condition

- Prevention is major principle of any medical discipline, similarly knowledge of genetics will enhance nurse's understanding that;
- Several genetic disorders can be prevented with prompt & early diagnosis & treatment. For example, phenylketonuria (PKU) related mental retardation could be prevented with early newborn screening & diagnosis & diet management.
- The genetic disorders can be prevented by selected interventions. For example, risk of neural tube defect can be minimized with administration of folic acid in first trimester of pregnancy.

Management & care in genetic disorders

- Knowledge of genetics will empower the nurses to manage & care for patients with genetic disorders in their routine health care practice by building up their understanding about;
- Genetic approaches to the therapy of genetic & complex diseases.
- Care management of adults with childhood genetic disorders
- Care management of persons with adult genetic disorders such as Huntington disease.
- Ways in which genetic knowledge is used in diagnosis & treatment application.

Genetic information & counselling

- Knowledge of genetic will help them to;
- Development of non-judgemental attitudes about genetics & related disorders.
- What information needs to be collected before providing genetic counselling.
- Role of a nurse in delivering genetic information & counselling.
- Application of traditional nursing skills such as patient education, confidentiality, & counselling about genetic information. The concept of non-directive counselling can be included.

Referral Services

- In developing countries, there is less awareness about genetic disorders & health care facilities offering services for testing & management of genetic disorders. Nurses are the primary health care providers who can direct them to right place for their diagnosis & management. So that, genetic information will equip nurses to provide effective referral services to their genetic clients.
- Services available to manage the genetic disorders at local or national level.
- Knowledge about referral possibilities knowing not only who should be referred but also how & to whom it should be done.
- Ways to access resources relating to genetics for patient & self-education & the need to keep them up-to-date.

Social & Ethical Issues in Genetics
There are several social & ethical issues, which play important role in care of patients with genetic disorders. Therefore, study of genetics will make nurses to build:

- An awareness of social, legal, & ethical issues related to genetics, including effects on individuals, groups, & societies, some of which are unique to genetic conditions.

Conclusion:

- Nursing practice in genetic-related health care blends the principles of human genetics with nursing care in collaboration with other professionals, including genetics specialists, to foster health improvement, maintenance & restoration.

In any practice setting nurses will carry out five main activities in genetic-related nursing practice:

- Collect & interpret relevant family & medical histories
- Identify patients & families who need further genetic evaluation & counselling
- Refer them to appropriate genetics services
- Offer genetic information to patient & families
- Collaborate with genetics specialist; & participate in the management & coordination of care of patients with genetic conditions.

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