

**Tishk International University**  
**Faculty of Nursing**



# **GENETICS**

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# INTRODUCTION TO GENETICS



# INTRODUCTION

- ▶ During mid 19<sup>th</sup> Century, Gregor Mendel observed that certain features pass from parents to their children/offspring.
- ▶ A child usually looks like their parents and is due to inheritance of certain characteristics from parents to children .
- ▶ This transmission of characteristics from parents to children is known as **heredity**.
- ▶ The basic unit of heredity is **gene**, which consist of portion of DNA molecules.
- ▶ The term gene was coined by Johannsen in 1909.



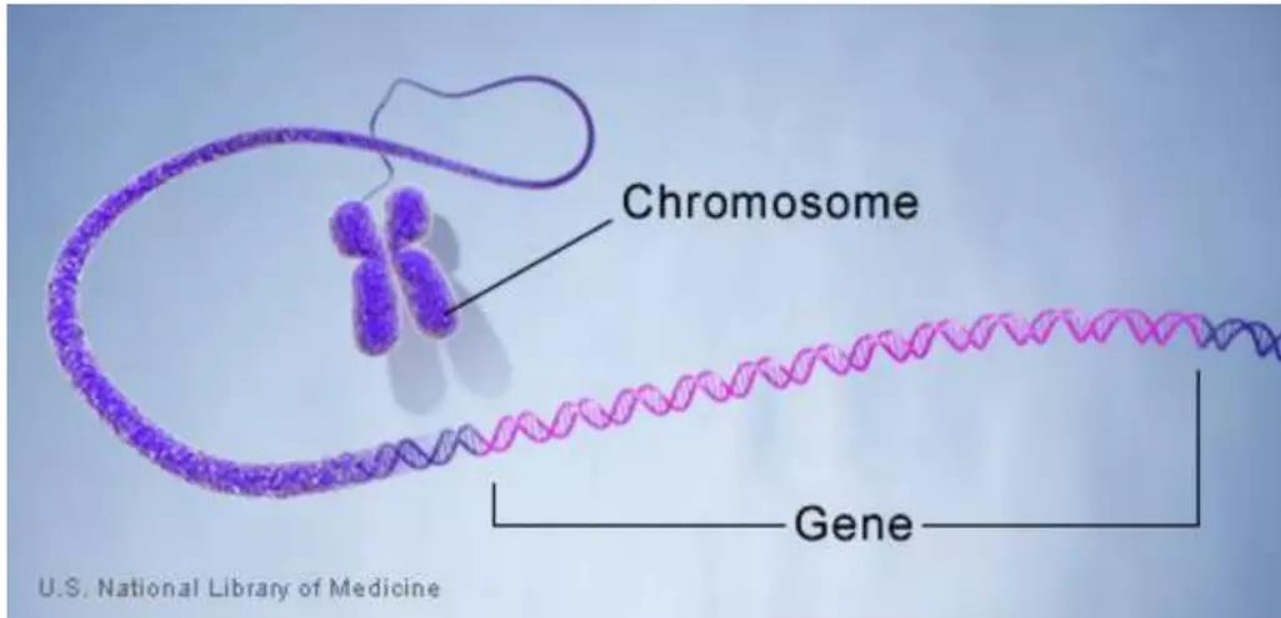
# GENETICS

- ▶ Genetics is the study which deals with the science of genes, heredity and its variations in living organism.
- ▶ **Gregor Mendel is the father of Genetics**
- ▶ The term Genetics was coined by William Bateson

# Terminologies

- ▶ GENE
- ▶ ALLELES
  - ▶ Dominant
  - ▶ Recessive
- ▶ Chromosomes

# Gene

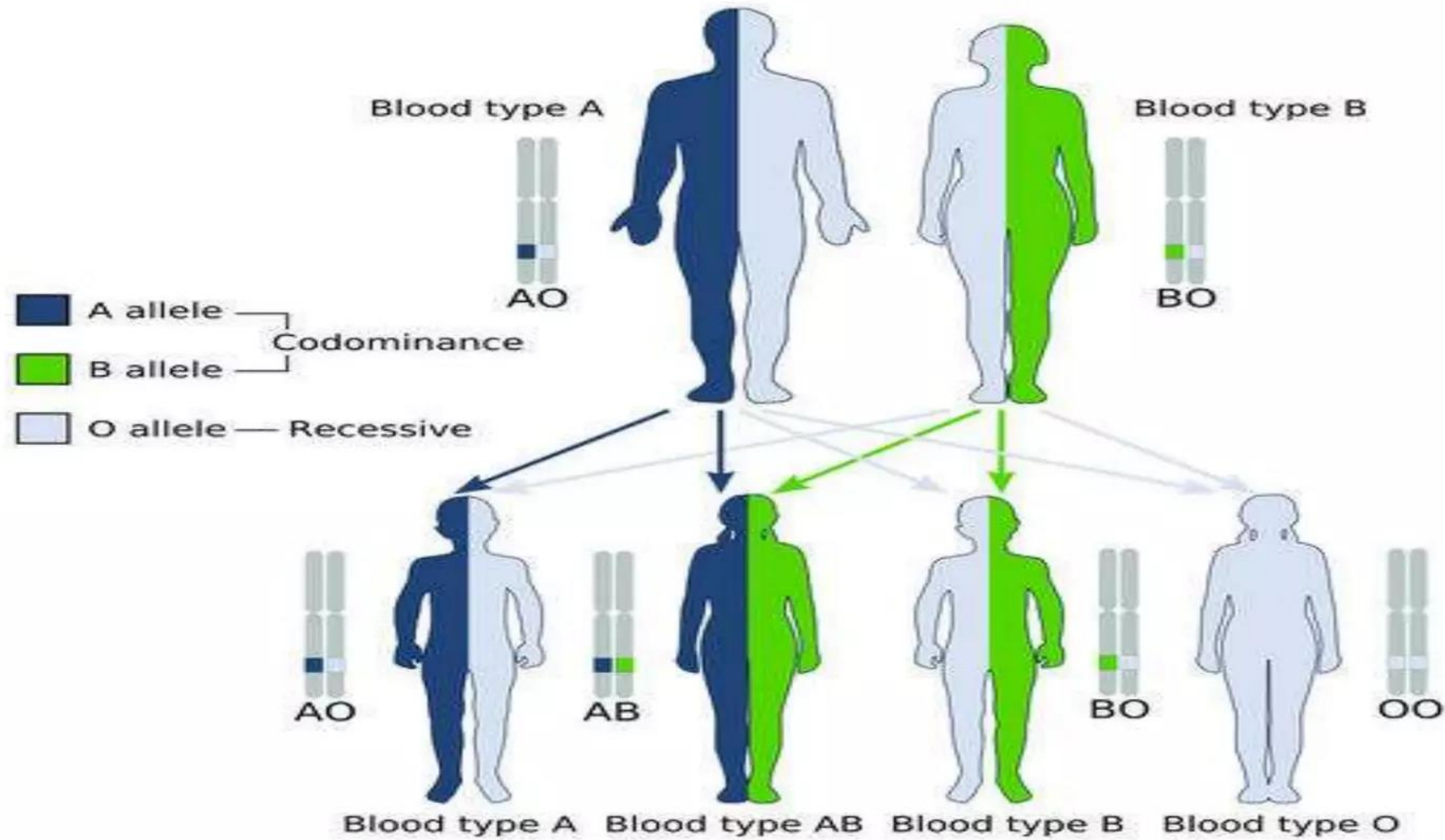


# Gene

- ▶ Gene is defined as a segment of DNA (Deoxyribonucleic Acid) which carries the genetic information.
- ▶ Gene is the basic physical and functional unit of heredity
- ▶ DNA has also segment which do not contain gene.
- ▶ The human genome contains about 30000 – 40000 genes and each gene varies in size.

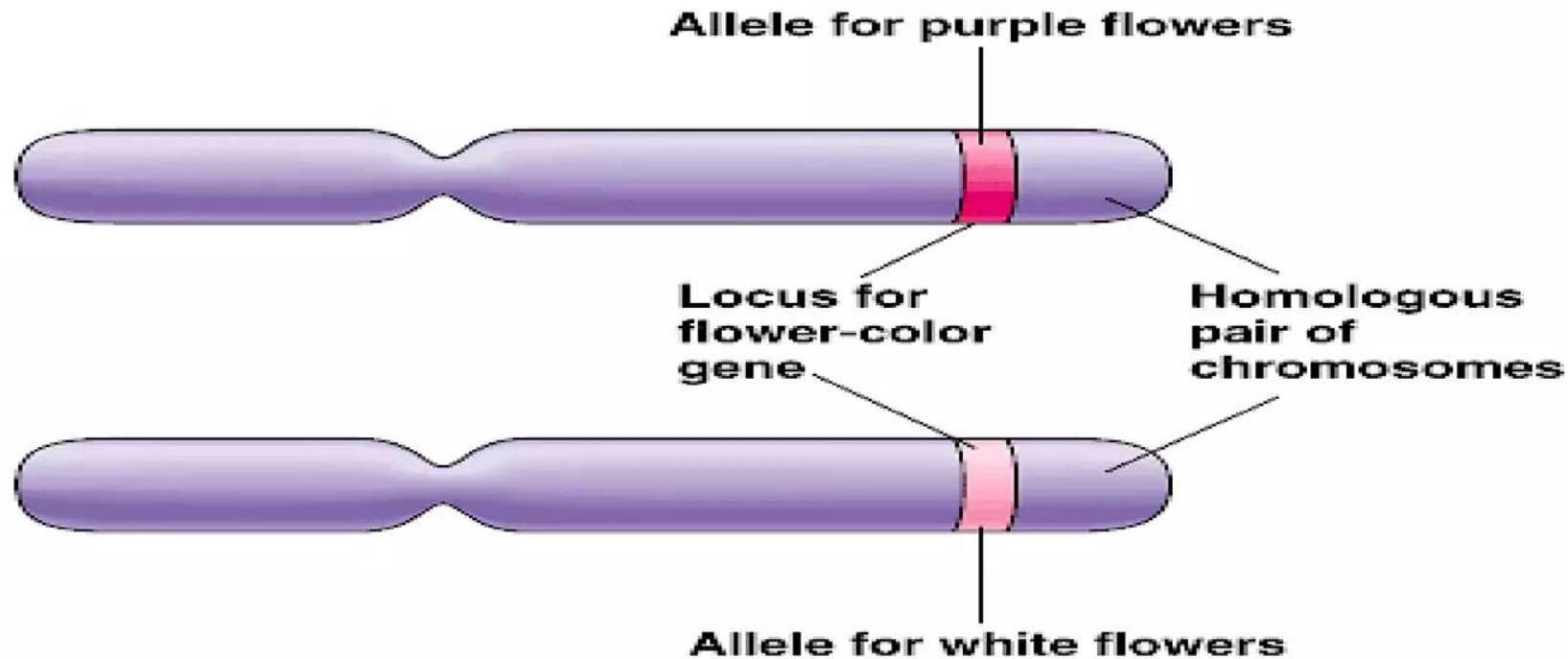


# Alleles

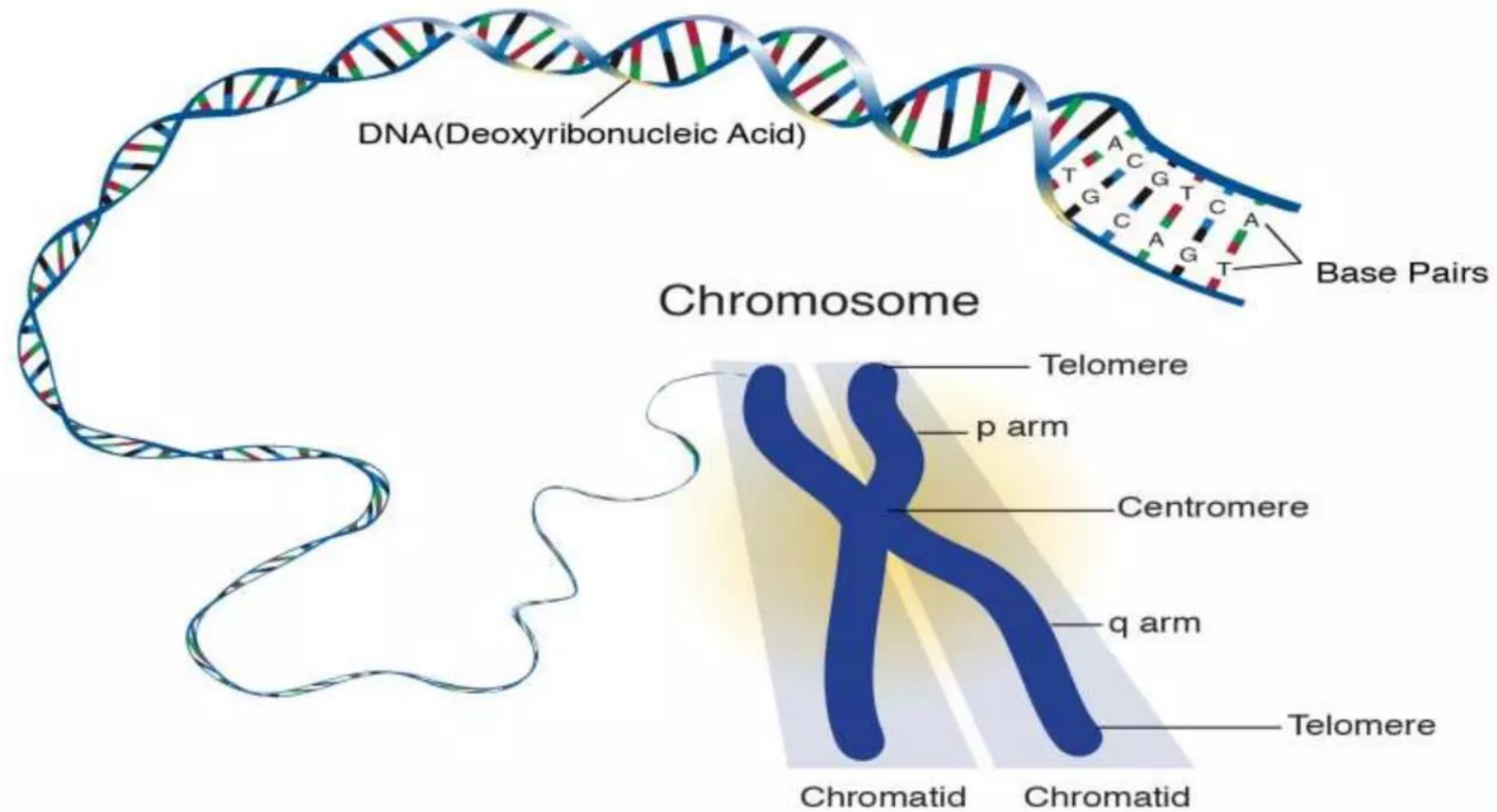


# Alleles

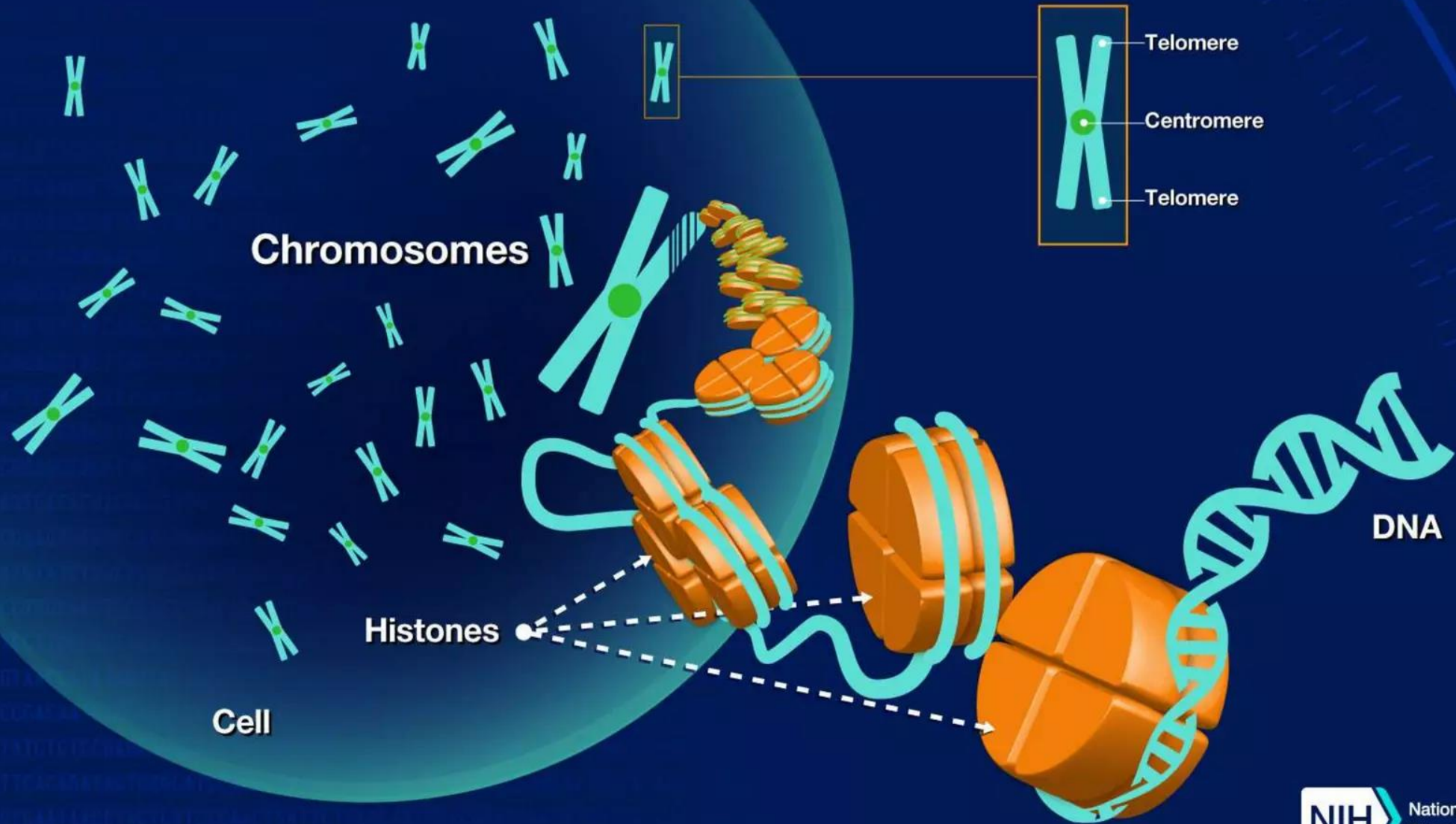
- ▶ An allele is one of two, or more, forms of a given gene variant.
- ▶ Each allele determines a single inherited characteristics in an individual.



# CHROMOSOMES



# Chromosome



# Chromosomes

- ▶ Chromosomes are thread-like structures located inside the nucleus of animal and plant cells. Each chromosome is made of protein and a single molecule of deoxyribonucleic acid (DNA).
- ▶ Passed from parents to offspring, DNA contains the specific instructions that make each type of living creature unique.
- ▶ The term chromosome comes from the Greek words for color (chroma) and body (soma). Scientists gave this name to chromosomes because they are cell structures, or bodies, that are strongly stained by some colorful dyes used in research.

# PRACTICAL APPLICATIONS OF GENETIC IN NURSING



# Role of Nurse in Genetics

- ▶ Nurses came across individuals or families affected by the genetic diseases.
- ▶ Nurses are a vital links between patients and health care services.
- ▶ Nurses should have a basic sound knowledge of genetics.
- ▶ The important role of nurses in genetic include -

# Genetic Counseling and Interviewing

- ▶ Interviewing patients or individuals with suspected genetic disorders
- ▶ Taking a detailed clinical history along with relevant family history (over three generations) from patients or parents of child with genetic disorder.
- ▶ Refer those with genetic disorder to the concerned doctor.



# Planning, Screening or Gene Based Testing Programs

- ▶ Provide health education related to genetics and genetic testing.
- ▶ Drawing and interpreting a pedigree chart.
- ▶ Ability to recognize the possibility of a genetic disorder based on the pedigree chart.
- ▶ Assessment of a genetic risk especially in conjugation with genetic testing options.

# Monitoring

- ▶ Follow up of positive newborn screening test.
- ▶ Monitoring individuals with genetic disorders
- ▶ Working with families under stress due to a genetic disorder.

# Care

- ▶ Developing an individualized plan of care and services of affected patient.
- ▶ Participating in public education about genetics.
- ▶ Maintain the privacy and confidentiality of the patients genetic information.

# Educational Role

## **Genetic Aspect**

- ▶ When a genetic condition is identified, it leads to stress and shock in the individuals and his family.
- ▶ The nurses have a major role in counseling, reducing their fears, getting the consent for genetic testing and arranging the tests and offering post test advice.

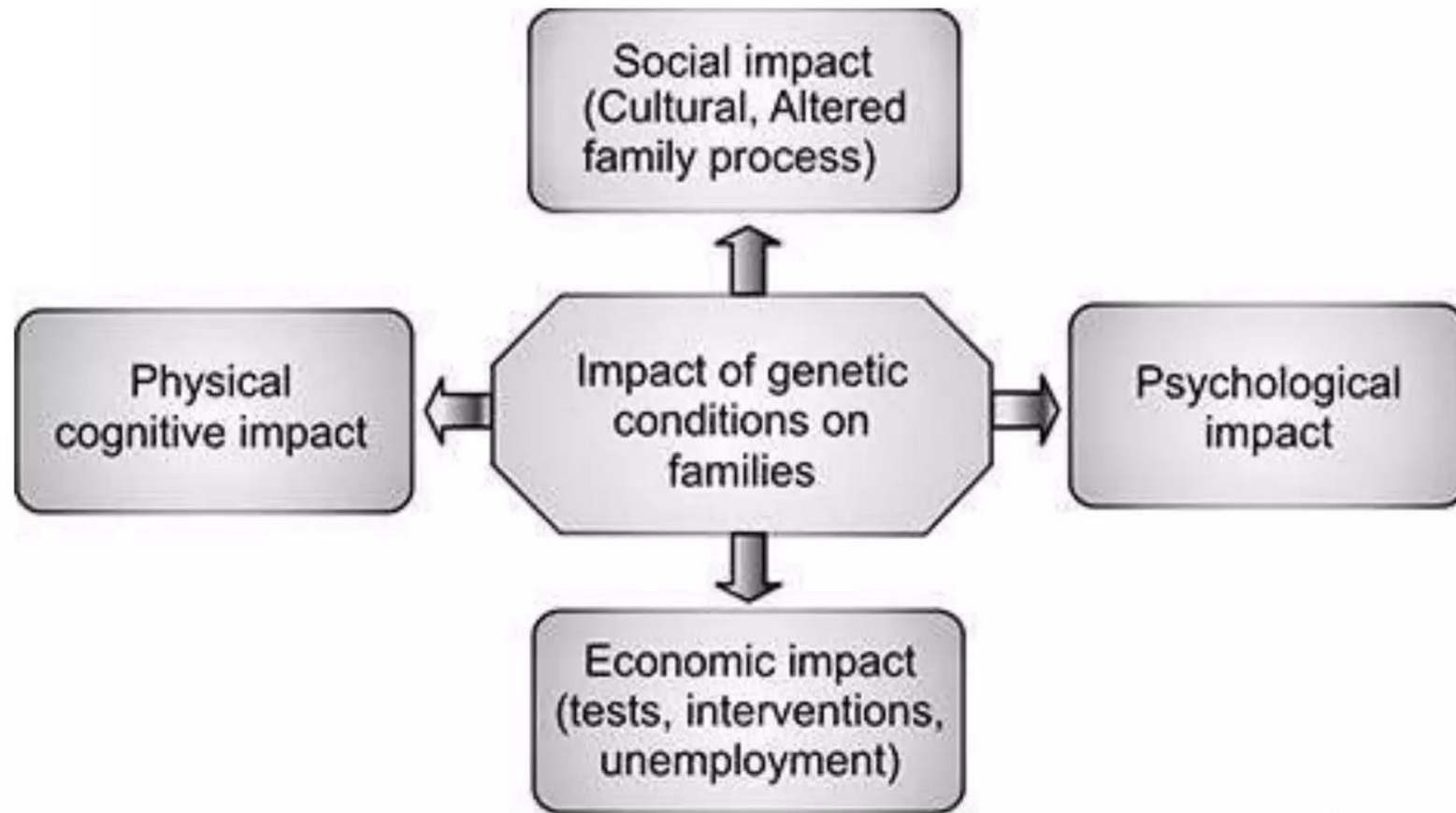
## **About Transmission of genetic condition within families**

- If an individual is identified to have a genetic condition, nurses should educate the family members, who are likely to be affected and provide advice, counseling and screening for them.

# Educational Role

- Educate how genetics and environmental factors influence health and disease.
- Nurses should be able to identify the Mendelian patterns of inheritance of genetic conditions in families in the form of a pedigree (family tree)

# Impact of Genetic Conditions on Families



# Impact of Genetic Conditions on Families

- Guilt – Parents with genetic disorder tend to feel guilty, when they come to know that they might have passed on a condition to a child.
- Depression – When an individual comes to know that he/she has a genetic condition and the decision not to have a children or decision to terminate a pregnancy, may result in depression or loss of peace of mind.