



ISSN 2278 – 0211 (Online)

## Who Is the Catalyst of Your Life?-"Genes"

**Vasanthakohila K. R.**

Professor, HOD, Community Health Nursing Sree Balaji College of Nursing,  
Bharath University, Chennai, India

**V. Hemavathy**

Principal, Sree Balaji College of Nursing, Bharath University, Chennai, India

**Abstract:** The cell cycle or cell-division cycle is the series of events that takes place in a cell leading to its division and duplication. Genes are working subunits of DNA. There are between 50,000 and 1,00,000 genes, and every gene is made up of thousands, even hundreds of thousands, of chemical basis. Genetic disease in humans is caused due to abnormalities in genes or chromosomes. Prenatal diagnosis is any of various diagnostic techniques to determine whether a developing fetus is affected with genetic disorder.

**Keywords:** Cell, Cell division, Gene, Genetic disorders, Chromosomes

### 1. Introduction of Cell

The cell is one of the most basic units of life. There are millions of different types of cells. There are cells that are organisms on their own, such as amoeba and bacteria cells and there are cells that only function when part of a larger organism, such as cells that make up the human body. The cell is the smallest unit of life in our bodies. All the cells have a "outer membrane" called the plasma membrane, protecting it from the outside environment. The cell nucleus contains the cell's DNA, the genetic code that coordinates protein synthesis.

### 2. Cell Division

**MITOSIS:** Mitosis is the process of forming identical daughter cells by replicating and dividing the original chromosomes, in effect of making a cellular Xerox. Mitosis is divided into following four stages

- Prophase
- Metaphase
- Anaphase
- Telophase

Prophase – The DNA molecules progressively shorten and condense by coiling to form chromosomes.

Metaphase- the spindle fibres attach themselves to the centromeres of the chromosomes and align the chromosomes at the equatorial plate

Anaphase- the spindle fibres shorten and the centromere splits, separated sister chromatids are pulled along behind the centromere.

Telophase- the chromosomes reach the poles of their respective spindles. Nuclear envelope reforms before the chromosomes uncoil. The spindle fibres disintegrate.

Cytokinesis is a process of splitting the daughter cells apart. Where mitosis is the division of the nucleus, cytokinesis is the splitting of the cytoplasm and allocation of the golgi, plastids and cytoplasm into each new cell. At the time of cytoplasmic division, organelles like mitochondria and plastids get distributed between the two daughter cells.

### 3. Meiosis

Sexual reproduction only in Eukaryotes. It is a special type of nuclear division which segregates one copy of each homologous chromosome into each new gamete.

#### 3.1. Chromosomes

Thread like structures that are visible under light microscope. Each chromosome differs in size.

### 3.2. Components of Chromosomes



Figure 1

It consists of

- Deoxyribonucleic acid
- Protein
- RNA
- Uracil is replaced in Thymine

### 3.3. Types of Nucleic Acid

The majority of chromosomal DNA is double stranded helix. But it is single stranded at the end of chromosomes, it is called TELOMERE. Deoxyribonucleic acid has nucleotides called PURINES&PYRIMIDINES.

Purine bases are Adenine (A) & Guanine (G)

Pyrimidines bases are Thymine (T) & Cytosine (G)

The two nucleotide chains of DNA are held together by two types of molecular forces. Adenine base on one strand always pairs with Thymine (A-T) or (T-A).Guanine base on one strand always pairs with Cytosine (G-C) or(C-G).

## 4. Genes & Genetics

Gene is defined as a segment of DNA which carries the genetic information

Genetics is the science that studies the structure and behavior of gene.

### 4.1. Concept of Gene

T.H.Morgan (1910) proposes that genes are arranged on a chromosome in linear sequence like beads on a string. Each gene occupies a specific place called its "locus". It is a basic unit of structure which is not sub divisible by recombination and is the smallest unit of genetic material capable of independent mutation .Gene is called unit of function and unit of structure defined by recombination and mutation.

### 4.2. Gene Mutation

A gene mutation is a permanent change in the DNA sequence that makes up a gene. Mutation range in size from a single DNA building block to a large segment of chromosomes.

### 4.3. Causes of Mutation

- DNA fails to copy accurately
- External influences
- Viruses
- Radiation
- Chemicals

### 4.4. Effect of Mutation

Some mutations do not have any noticeable effect on the phenotype of an organism. This happens in many situations perhaps the mutation occurs in a stretch of DNA with no function or perhaps the mutation occurs in a protein coding region, but ends up not affecting the amino acid sequence of the protein.

### 4.5. Chromosomal Abnormalities

Chromosomal Abnormalities occurs when there is a change within normal chromosome of human and also caused by errors in numbers or structure of chromosome. Chromosomal Abnormalities usually result from an error that occurs when an egg or a sperm develops .Egg and Sperm cell each contains 23 chromosomes. They join together to form a fertilized egg with 46 chromosomes. When this cell with wrong number of chromosome joints with a normal cell, the regulating embryo has a chromosomal abnormality.

### 4.6. Causes of Chromosomal Abnormalities

- Error in cell division
- Elderly primi mother(>35 years)
- Environmental factors
- Medicines

- Medical conditions

#### 4.7. Common Chromosomal Abnormalities:

##### 4.7.1. Down Syndrome or Mongolism



Figure 2

This is also called Trisomy 21 syndrome it is defined as congenital condition characterized by physical malformations and some degree of mental handicap.

Occurs in children of mothers who are in older child bearing age

- Signs & Symptoms are
  - Microgenia (small chin)
  - Flat nasal bridge
  - Oblique eye
  - Flat and broad face
  - Short neck
  - White spot on iris known as brush field spot
  - Excessive space between large toe and second toe
  - Obese
  - Maximum height for male is 154 cm
  - Maximum height for female is 144 cm

##### 4.7.2. XYY Syndrome (Aneuploidy)

Also called super male syndrome. Mainly caused by abnormal number of sex chromosomes and occur in 1 in 1000 new born boys. Here extra 'Y' chromosome is present in each male.

- Signs & Symptoms Are
  - Very tall than average
  - High risk for learning abilities, poor motor skills development. eg Walking, Sitting
  - Behavioural problems e.g. Temper tantrum, Aggressive in nature, Emotional immaturity
  - Delayed development in speech & language

##### 4.7.3. Turner Syndrome

Occurs in females, about 1 in 2000 live birth. Here only one 'X' is present

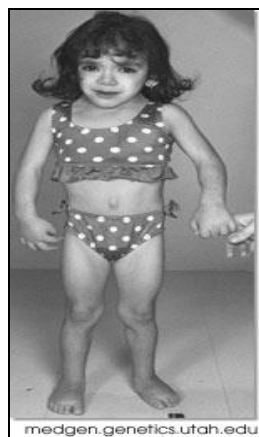


Figure 3

- Signs & Symptoms are
  - Swollen hands and feet
  - Wide web neck
  - Small breast ,drooping eye lids
  - Infertility
  - Short height
  - Absence of menstruation
  - Vaginal dryness(pain during intercourse)

#### 4.7.4. XXX Syndrome or Super Female

They are also called meta females occur 1 in 1000 females and caused by error in cell division.

- Signs & Symptoms are
  - Tall stature
  - Microcephaly
  - Vertical skin folds that may cover the inner covers of eyes
  - Delayed motors skills ,speech, language,
  - Low self –esteem
  - Struggle with personality & psychological problems

#### 4.7.5. Klinefeltersyndrome

A congenital chromosome abnormality in which each cell has three sex chromosomes. XXY is present rather than usual XX or XY making a total of 47 instead of 46.

- Signs & Symptoms are
  - Lack of facial and body hair, Enlarged breasts
  - Small firm testes, small penis
  - Abnormal body proportions (long legs, short trunk)
  - Tallness with extra long arms and legs
  - Social and learning disabilities (common)
  - Personality impairment
  - Speech and language problems—Children with KS often learn to speak later than other children do. They may have a difficult time reading and writing.
  - Normal to borderline IQ
  - Lack of ability to produce sperm (common)

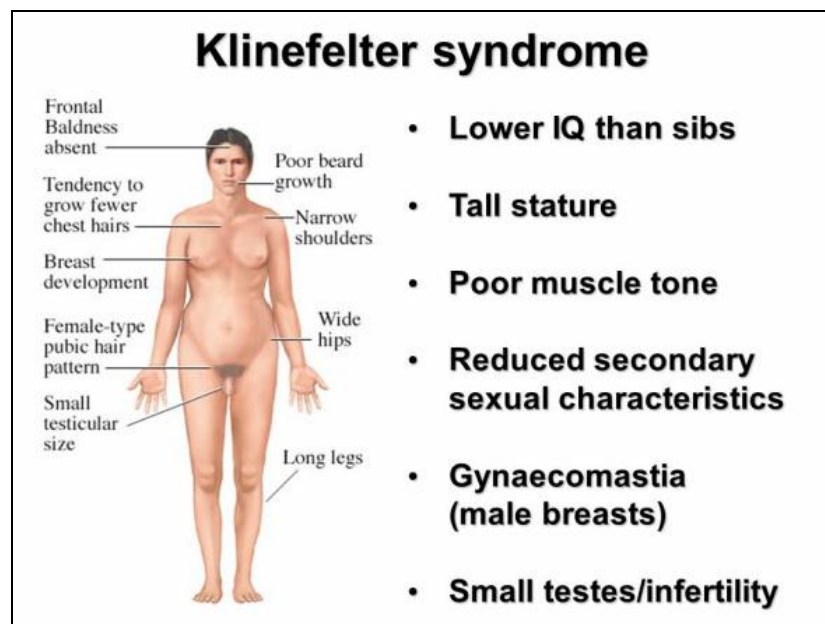


Figure 4

## 4.8. How to Overcome This?

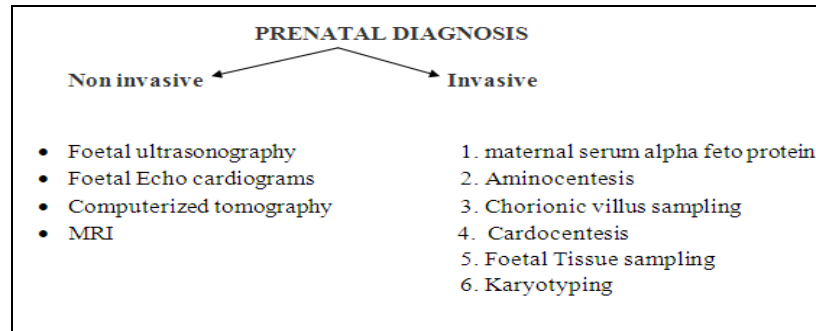


Figure 5

- Genetic Councelling
  - Directive counseling
  - Advocacy counseling
  - Informative counseling
  - Supporting counseling

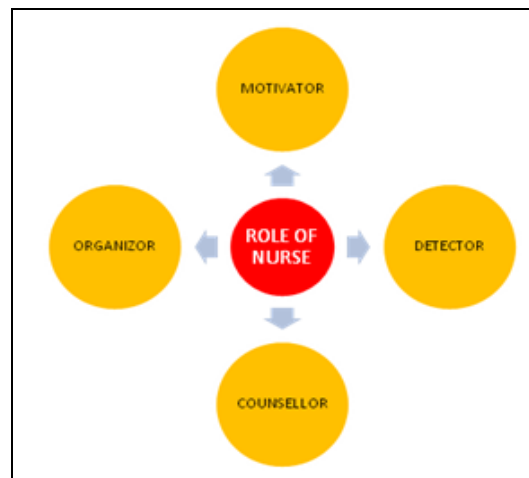


Figure 6: Role of Nurse

## 5. Conclusion

Genetics is affecting all type of population. The way in which, nurses think about planning health care must be seen how now through a genetic eyes or lens, and nurses must learn to think genetically. Genetics is very important in the field of nursing in order to find out the genetic abnormalities in an earlier stage and to advice for better future. Genetics related nursing practice includes the care of clients who have genetics conditions, persons who may be predisposed to develop or pass on genetic conditions and person who are seeking genetics information and referral for additional genetics services.

## 6. References

1. V.Deepa Parvathi "Genetics for Nurses" 1<sup>st</sup> edition 2013,Dorling KindersleyPvt Ltd,U.P,Page no 81-98
2. Dr.Purnima S Rao "Text book of Pathology & Genetics for Nursing" 1<sup>st</sup> edition 2008, Emmess Medical Publishers Bangalore,Page no 231-236
3. RamadossNayak,Sharada Rai "Text book of Pathology &Genetics for nurses" 1<sup>st</sup> edition 2013, Jaypee Brothers Medical Publication(p) LTD, NewDelhi Page no295-308
4. Shebeer.P.Basheer ,S.Yaseen Khan "A Concise text book of Advanced Nursing Practice",1<sup>st</sup> edition 2012, Emmess Medical Publishers Bangalore, Page no115-159 .
5. SamtaSon "Text book of Advanced Nursing Practice", 1<sup>st</sup> edition 2013, Jaypee Brothers Medical Publication(p) LTD, NewDelhi Page no246-270
6. www.Pub Med.com
7. Swww.google.com