

# GENETICS IN ORTHOPAEDICS

*ESSAY*

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*Orthopaedics*

*By*

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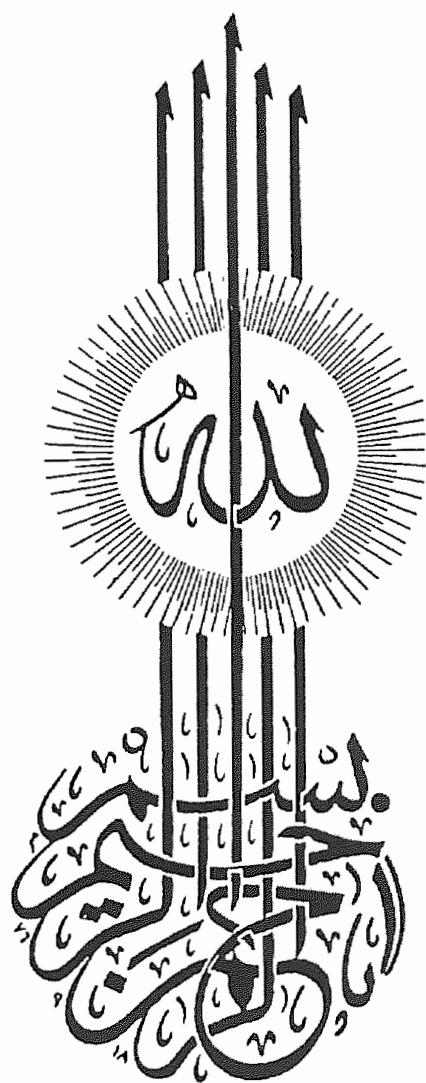
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TO MY  
MOTHER

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#### References of Figures

1. *Zitelli, Basil J. and Davis, Holly W. (1992):*  
Atlas of Pediatric Physical Diagnosis, 2nd. edn.  
Gower Medical Publishing, New York - London.
2. *Emery AEH, Mueller RF (1992):*  
Elements of Medical Genetics, 8th. edn.  
Churchill Livingstone, Edinburgh.
3. *Apley, A. Graham and Solomon, L. (1982):*  
System of Orthopaedics and Fractures, 6th. edn.  
Butterworth & Co. (Publishers) Ltd.



# *INTRODUCTION*

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## INTRODUCTION

The constitution of an individual is determined by the interplay of many factors, some of these factors were at work before the child was born, others are acquired after birth.

All people are recognizably human, but no one is exactly like anybody else.

The hereditary transmission of physical & moral qualities in humans was suggested as early as 1823 by W. Lawrence<sup>(1)</sup>.

But in the last ten years, the science of genetics has been developed progressively up to date & its relations to many diseases in different subjects of medicine have been revealed.

# *CHAPTER (I)*

## *CHROMOSOMAL BASIS OF INHERITANCE*

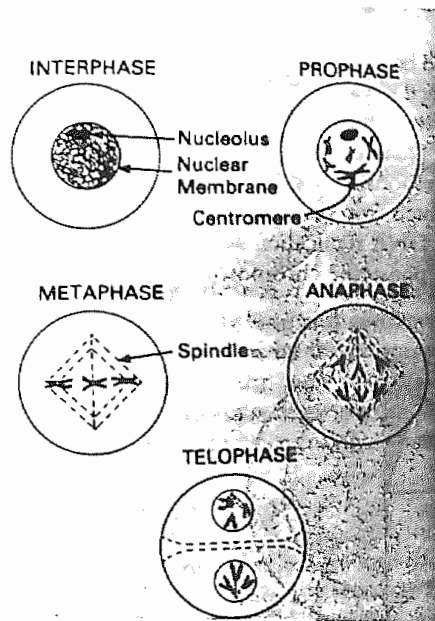


Fig. (1): Stages of mitosis.

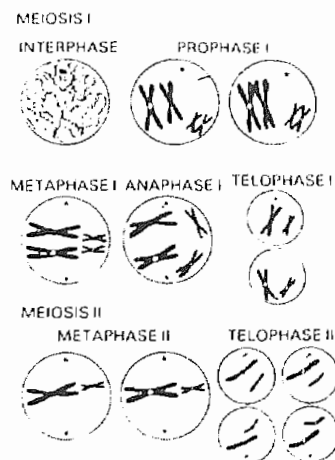


Fig. (2): Stages of meiosis.

\* The Normal Human Chromosomes:

The chromosomes are the portions of the chromatin material of the nucleus which carry the genetic information. When a cell divides, the nuclear material is seen to form a number of small, deeply staining bodies, the chromosomes (chromos = colour, soma = body). Chromosomes are composed of deoxyribonucleic acid (DNA) on a framework of protein (histone). Segments of the DNA molecules comprise the genes, the units of heredity. Each gene has a precise position known as a locus. The genotype of an individual is his full set of genes. The phenotype is the expression of those genes as physical, biochemical and physiological traits. The genome is the full set of genes.

In the human being, there are 46 chromosomes in each somatic cell. These form 23 homologous pairs; one member of each pair has been received from one parent. This is the diploid number, 46. The haploid number, 23, is present in the ovum and sperm.

In some abnormal conditions there are more than two sets of chromosomes in the cell, a condition called polyploidy, this may be triploidy i.e. 69 chromosomes or tetraploidy i.e. 92 chromosomes. Twenty two of the twenty three pairs of chromosomes are similar in males and females and

# THE CHROMOSOMAL BASIS OF INHERITANCE

## \* Chromosomes in cell division:

With the exception of some highly differentiated cells as the neurones, all the cells of the body are potentially able to divide into two essentially similar cells.

## \* There are two types of nuclear division:

1. Mitosis which occurs in somatic cells resulting in two daughter cells with the same number of chromosomes and exactly the same amount of genetic material as the parent cell.
2. Meiosis occurs in the germinal cells during the formation of the gametes and has the effect of reducing by half the number of chromosomes and consequently the amount of genetic material in the daughter cells.

This consists of two divisions, the first is a reduction and the second is essentially the same as a mitotic division. The final result is that each ova or sperm contains the haploid chromosome number i.e. 23 chromosome<sup>(2)</sup>.

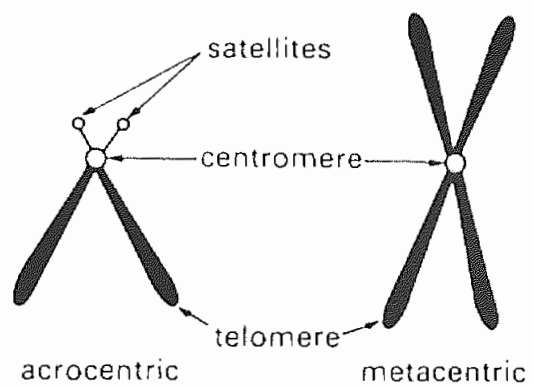


Fig. (3): Diagram of an acrocentric and a metacentric chromosome.