# Cytogenetics in Gynecology and Obstetrics

## Essay

Submitted For Partial Fullfilment of master degree in Obstetrics and Gynecology



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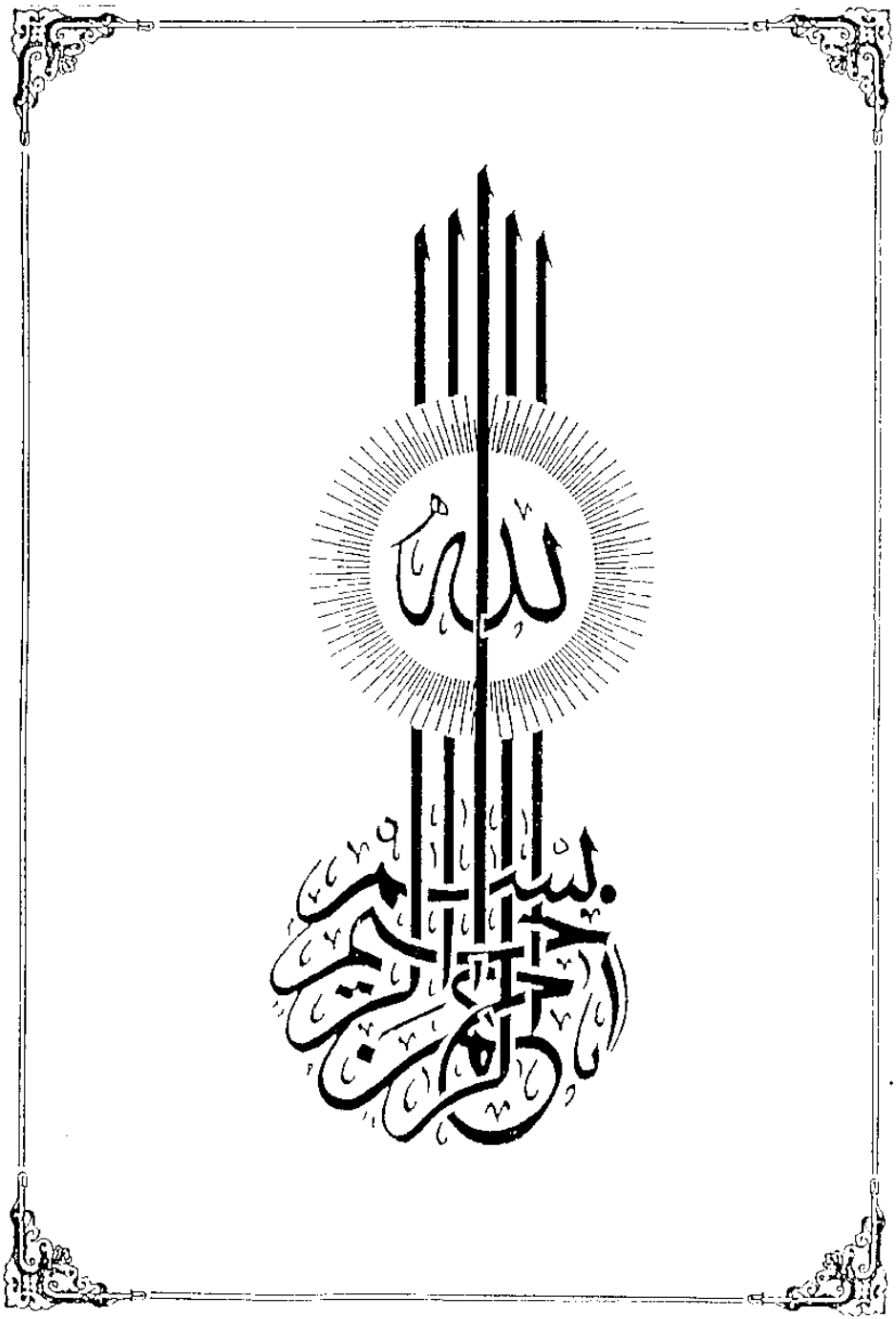
**Cairo 1994**

بسم الله الرحمـــن الرحيـــم

" * الله يعـلم ما تحمـل كل أنثـــي
وما تغيض الأرحـــام وماتـــزداد
وكـــل شـــيء عـنـده بمقـدار *
عالم الغيب والشهادة الكبير المتعال* "

صـــدق اللـــه العظيـم

( آية ٨ : ٩ سورة الرعـــد )



# Acknowledgement

No words can sufficiently express the deep gratitude that I feel towards my Prof. Dr. Mohamed Farouk Mohamed Mahfouz Fikry, Professor of Obstetrics and Gynecology. Faculty of Medicine, kind help and patience that were much beyond Praise.

I owe much of my basic and advanced knowledge in the field of Cytogenetic practice in Gynecology and Obstetrics to the eminent work of Prof. Dr. Farouk Fikry " Chromosomal Sexing and Counting in Obstetrical and Gynecological practice " That was a Thesis submitted in Part Fulfilment for the degree of M.D, in Gynecol. & Obstet . Presented to the department of Gynecol & Obstet. Faculty of Medicine Ain Shams University. Cairo. 1966.

Without the guidance of this pioneer work, the establishment of my essay " Cytogenetics in Gynecol& Obstet." could not be realized.

I would like to express my deep gratitude to Dr. Ihab Hassan Abdel Fattah. Lecturer of Obstet & Gynecol. Faculty

of Medicine. Ain shams University, for his unforgetable valuable guidance, Help, advice, and meticulous revision all through this work.

# CONTENTS

## TABLE OF ABBREVATIONS

The following table lists symbols used to designate parts of chromosomes and certain rearrangements and abbreviations in the essay.

(Some symbols Recommended by the Paris Conference (1971 and Supplement 1975).

| | |
|---|---|
| Centromere | Cen |
| Short arm | p |
| Long arm | q |
| Isochromosome | i |
| Deletion | del |
| Translocation | t |
| Reciprocal translocation | rec |
| Mosaicism | mos |
| Chimerism | chi |
| Ring | r |
| Dicentric | dic |
| Duplication | dup |
| Inversion | inv |
| Break without reunion (e.g teminal deletion) | : |
| Break and join | :: |
| From ... to ..... | → |
| complementary deoxyriboucleic acid | c DNA |
| milli sieverts | m Sv. |
| restriction fragment length poloymorphisms | RFLPs |
| Recombinant deoxyribonucleic acid | r DNAA |
| ribonucleic acid | RNA |
| mesenger ribonucleic acid | m RNA |

***(Simpson and Martin. 1977).***

# INTRODUCTION& AIM OF THE WORK

# INTRODUCTION

Genetic problems are frequently encountered initially by the obstetrician gynecologist as the primary physician to women. During the routine prenatal evaluation the obstetrician may become aware of specific risk factors, such as advanced maternal age or family history of specific genetic disorders. Patients frequently pose questions about the risks and the nature of genetic and non genetic birth defects. Subsequently, the obstetrician is often the first to observe the presence of a serious congenital defect in the child, necessitating propor diagnosis, management and interpretation to the parents.

Thus the obstetrician must be familiar both with the fundamental principles of medical genetics and with those disorders of greatest relevance to feto - maternal health and reproductive function.

# Aim of the work

This study aims to review the old and recent litratures concerning the major principles of cytogenetics and survey the contribution of cytogenetic disorders to reproductive problems and effects of environmental factors on the development of genetic disorders.

# BASIC PRINCIPLES

# Historical background

N.B. The refrence of this chapter is Emery. Elements of medical genetics 8th edition 1992 (1:9).

The history of medical genetics can be divided into three phases. The first followed the rediscovery of Mendel's laws around the trun of the century, and was concerned with analysis of the inheritance patterns of genetic disorders and with their division into autosomal and X-linked dominant and recessive traits. The second phase started in the late 1940s, when it was discovered that mutant genes could be followed trough the proteins they controlled. The classic example is sickle cell anaemia, a recessively inherited disease which can be tracked by measurement of an abnormal haemoglobin molecule in red blood cells. The third phase, sometimes called the newgenetics' , stems from technologies developed in the late 1970s which allow direct analysis of genes themselves. Although each phase has added to the power of the preceding one, the potential of gene analysis for the diagnosis of genetic disorders is almost without limit.

We will discuss in some detail some points as regard the history of medical genetics

It is a fact that the interst in human genetics did not spring up overnight. The inheritance of the bleeding disorder, haemophilia, was mentioned in the Talmud some 1500 years ago. The development of the embryo and its gradual increase in size and complexity throughout the whole period of gestation was mentioned in koran several centuries earlier.

In the 17th century, Regnier de Graaf described the Gaafian folicle as containing the unfertilized egg or ovum which contributed half the heriditary material with the sperm. In the 18th century, Pierre Louis Moreau de Maupertuis studied certain heriditary traits in man such as polydactly and albinism and from pedigree studies showed that these two conditions were inherited in different ways. Otto's account in 1803 of haemophilia in a new hampshire family was apparently the earliest clear discription of the clinical features and mode of inheritance of this disease : it was transmitted by healthy carrier females to their sons but never by an affected father to his son, so called sex - linked inheritance.

Perhaps the real founder of human genetics was Joseph Adams In 1814 he published a book entiteled A Treatise on the Supposed Heriditary Properties of Diseases, in which he