

Cytogenetics in Gynecology and Obstetrics

Essay

Submitted For Partial Fulfillment of master degree in Obstetrics and Gynecology

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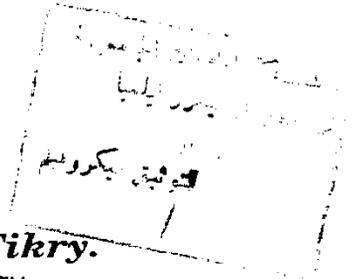
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بِسْمِ اللّٰهِ الرَّحْمٰنِ الرَّحِیْمِ

**"* اللّٰهُ یَعْلَمُ مَا نَحْمَلُ كُلُّ اُنْثَى
وَمَا تَغِیْضُ الْاَرْحَامُ وَمَا تَزِدَاد
وَكُلُّ شَیْءٍ عِنْدَهُ بِمَقْدَارٍ *
عَالَمِ الْغِیْبِ وَالشَّهَادَةِ الْكَبِیْرِ الْمَتَعَالِ *"**

صَلَّى اللّٰهُ الْعَظِیْمِ

(آیة ۸ : ۹ سورة الرعد)



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TABLE OF ABBREVIATIONS

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 terminal deletion)	:
Break and join	::
From ... to	→
complementary deoxyribonucleic acid	cDNA
milli sieverts	m Sv.
restriction fragment length polymorphisms	RFLPs
Recombinant deoxyribonucleic acid	rDNA
ribonucleic acid	RNA
messenger ribonucleic acid	mRNA

(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 proper 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 literatures 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 reference 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 turn 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 through 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

Historical background

It is a fact that the interest 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 the Koran several centuries earlier.

In the 17th century, Regnier de Graaf described the Graafian follicle as containing the unfertilized egg or ovum which contributed half the hereditary material with the sperm. In the 18th century, Pierre Louis Moreau de Maupertuis studied certain hereditary traits in man such as polydactyly 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 description 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 entitled *A Treatise on the Supposed Hereditary Properties of Diseases*, in which he