بسم الله الرحمن الرحيم

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تم عمل المسح الضوئي لهذة الرسالة بواسطة / سامية زكى يوسف

بقسم التوثيق الإلكتروني بمركز الشبكات وتكنولوجيا المعلومات دون أدنى مسئولية عن محتوى هذه الرسالة.

ملاحظات:

- بالرسالة صفحات لم ترد بالأصل
- بعض الصفحات الأصلية تالفة
- بالرسالة صفحات قد تكون مكررة
- بالرسالة صفحات قد يكون بها خطأ ترقيم

Prevalence and Genetic Diagnostic Classification of Patients in Genetics Clinic, Pediatric Department, Ain Shams University

Thesis Submitted for Partial Fulfillment of M.Sc. Degree in Pediatrics

BY

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The genetic contribution to disease varies; some disorders are entirely environmental and others are wholy genetic. Many common disorders, however, have an appreciable genetic contribution but do not follow simple patterns of inheritance within a family. The term *multifactorial* or *polygenic* inheritance have been used to describe the etiology of these disorders ***(Kingston, 1989)***.

Genetic abnormalities are a common cause of disease, handicaps, and death among infants and children. They account for the primary diagnosis of 11 - 16% of patients admitted to the pediatric units of teaching hospitals ***(Shapiro, 1996)***.

In 1991, hospitalizations that were related to birth defects and genetic diseases accounted for 9% and 12% of hospitalization in South Carolina and California, respectively, among persons younger than 20 years ***(Paula et al., 1997)***.

When clinically appraising and managing the child with an inherited disorder, three phases are critical:

1. Recognizing that the condition is inherited.

2. Identifying the pattern of inheritance.

3. Clarifying the clinical nature of disorder, which includes understanding the risk of the disease occurrence in siblings or other members of the family.

(Shapiro, 1996)

The main feature of clinical genetics is the involvement of close relatives in the diagnosis of a hereditary disorder, and the possible consequences of the findings for future generations *(Galjarr, 1997)*.

The information gained from the human genome project and related genetic research will undoubtedly create significant changes in health care practice. It provides an overview of basic genetic concepts including inheritance patterns of single gene conditions, pedigree construction, chromosome aberrations, and the multifactorial basis underlying many common diseases of adulthood *(Middelton et al., 1997)*.

The first accurate observation and definition of the normal number of chromosomes in a human cell was accomplished by ***Hsu and Levan*** in *1956 (Hall, 1996)*.

Each human somatic cell contains two copies of the entire human genetic program or "genome", amounting to 6 billion base pairs (bp) of DNA. DNA is portioned into 46 (23 pairs) large fragments, each contained in a specific autosomal chromosome or the X or Y chromosome. The "gene" is the functional entity of information *(Shapiro, 1996)*.