GENODERMATOSES PREVALENT AMONG EGYPTIAN CHILDREN

THESIS

Submitted for Partial Fulfilment of
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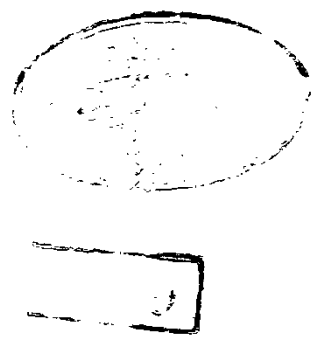

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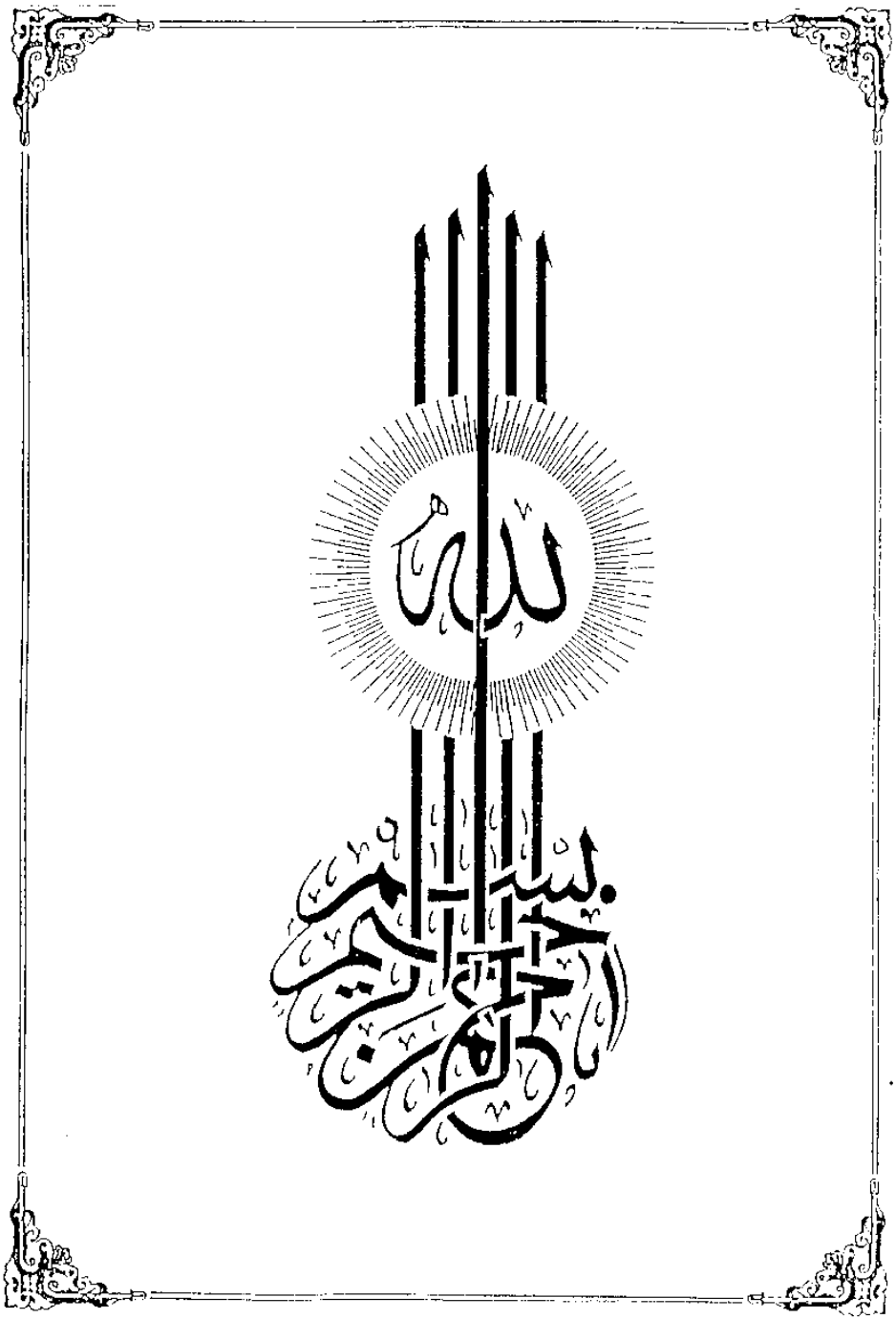

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Skin manifestations may reflect an inside disease in the body. Those genetically determined skin disorders in the production of which environmental factors play no obvious or decisive role are,for convenience, sometimes grouped together as the genodermatoses (Burton and Rook, 1986).

There are many dermatologic disorders that have a genetic etiology and others that have genetic predisposition. (Abnuelo,1987).

Not all hereditary disorders are congenital; they may not be apparent until later childhood or even old age. In general they tend to become manifest when the organ or tissue concerned first reaches its full functional development, but some are dependent on a wide variety of other factors, sometimes exogenous, or on the involutional changes of the ageing process. (Burton and Rook, 1986).

Pedigrees of families affected with heritable disorders due to mutations in single genes have provided imporant informations on how these conditions are inherited. Such informations are essential for accurate genetic counseling

of prospective parents who want to know their risk of producing an affected offspring (Carter and O'Keefe, 1985).

So, the aim of this work is to study skin diseases of genetic background and search for the thorough clincial picture of each disease entity and to detect the common category of genodermatoses in Egyptian children.

REVIEW OF LITERATURE

GENETIC PRINCIPLES

Understanding genetic principles and methods is essential for understanding the basis and the treatment of many skin diseases. Although means are not yet available for curing genetic diseases in terms of repairing the defect in the gene, many hereditary disorders are now amenable to satisfactory treatment aimed to averting the deleterious effects of the genes. (Goldsmith and MacKusick, 1987).

Types of Genetic Disorders

There are three general categories of genetic disorders (Table I). The chromosome disorders result from either a duplication or a deficiency of chromosomal material. The single gene defects are caused by mutaions in either one or Both genes of a pair and the multifactorial disorders by a combination of genetic and environmental factors. (Abuelo,1987).

Table (1) General categories of genetic disorders (Abuelo, 1987).

I	Chromosomal disorders
II	Single-gene disorders
	a) Autosomal dominant
	b) Autosomal recessive
	c) X-linked recessive
	d) X-linked dominant
III	Multifactorial disorders.

GENETIC PRINCIPLES AND CLASSIFICATION OF HEREDITARY SKIN DISORDERS

Chromosomal Abnormalities

There are both numerical and structural chromosome abnormalities. Abnormalities of chromosome number include trisomy (an extra chromosome) and monosomy (absence of a chromosome).

Abnormalities of structure include Translocations which are chromosoaml rearrangements; deletions of chromosomal material and duplications, that is, the presence of extra chromosomal material within achromosome (Abuelo, 1987).

Single - Gene Dirsorders

The single-Gene, or mendelian,disordes are transmitted by autosomal dominant, autosomal recessive, or X-linked genes. with regard to autosomal genes, each gene contributed by one parent, has acounterpart, or allele, from the other parent, if an abnormal gene causes a clinical abnormality despite the presence of its normal counterpart (i.e., in the heterozygous state), it is defined as a domiannt gene.

If , on the other hand, a single abnormal gene has no clinical effect, but a double "dose"(homozygosity) is required to produce the disorder, the gene has no clinical effect, the gene is defined as recessive. This difference between dominant and recessive genes is not always absolute. A heterozygous carrier of a recessive disorder should not have clinically apparent disease, but there may be biochemical abnormalities or other diferences that indicate the presence of the abnormal gene (Abuelo, 1987).

The characters of single gene disorders are collected in Tables 2a,b,c and figures 1,2,3.4.