سامية محمد مصطفى

شبكة المعلومات الجامعية

بسم الله الرحمن الرحيم

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قسم

نقسم بالله العظيم أن المادة التي تم توثيقها وتسجيلها علي هذه الأقراص المدمجة قد أعدت دون أية تغيرات

يجب أن

تحفظ هذه الأقراص المدمجة بعيدا عن الغبار

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بالرسالة صفحات لم ترد بالأصل

بسم الله الرحمن الرحيم

« سبحانك لا علم لنا إلا ما علمتنا إنك أنت العليم الحكيم »

سورة البقرة ، آية (٣٢)

GENETIC STUDY OF NON-ACQUIRED CHILDHOOD BLINDNESS

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To My Family

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CHAPTER I

INTRODUCTION

INTRODUCTION

Genetic study of non-acquired childhood blindness

Visual loss of any type in infancy and childhood is a cause of great concern to the patient and parents, therefore usually leads to prompt medical referral because impaired vision affects the development of the child (King, 1990)[1].

Vision is generally agreed to be the coordinating sense. Young blind children are at a disadvantage that the impairment will interrupt the normal developmental process to such a degree that the child's capacities of normal progress towards maturity and independence are seriously restricted (Zinkin, 1979)[2].

The structures of the eyes are observable to a very large extent, and a great number of ocular morphological abnormalities have been described . Some of these look dissimilar but may be due to the same hereditary cause, while others which look identical may be due to separate genetic mutations (Warburg, 1983)[3].

The normal visual acuity (V.a) is 6/6 in each eye and failure to achieve this requires explanation (King, 1990)[1].

The medical definition of blindness varies in different countries. According to the International Association for prevention

of blindness (I APB) it is defined as "visual acuity of 20/200 (6/60) or less in the better eye with best correction, or visual acuity of more than 20/200 if the widest diameter of the field vision subtends an angle no greater than 20 degrees" (Warburg, 1990)(4).

Hereditary disease of the eye, with rare exceptions, demonstrates bilateral, symmetric involvement. When unilateral ocular disease is seen, other causes, such as birth defect, intrauterine or antenatal infection, and inflammatory disease should be evaluated first, before considering a hereditary etiology. (Heckenlively and Daiger, 1997)(5)

For the handicapped blind children a comprehensive rehabilitation program is always needed, to prepare the blind persons for a full and well adjusted independent life, as a member of his family, community and in his leisure time (WHO, 1982)(6).

Prevention of childhood blindness requires increasing knowledge of genetic eye diseases and provision of accurate genetic counseling (Arnould and Hussels 1997)(7)

Genetic counseling in cases of infantile blindness requires a thorough examination of the whole patient and not only his eyes. The relatives must be asked for minor expression of the trait in question. This is almost importance for giving advice regarding the propabilities of fetal involvement and prognosis to the affected individual, his parents and other relatives (Warburg, 1990)(4).

Anatomy and physiology of the eye

The eye is the special organ of sight, through which we communicate with the outer world, where objects are percevied by them and brought to the brain for interpretation.

Idealy the eye enjoys distinct vision without an effort or fatigue, if incident parallel rays emitted from a distant object are brought to a focus upon the retina, which is considered as the receptive neural layer, and to it all the other structures of the eye and its adnexa are subsidiary serving the functions of protection, nutrition, dioptric adjustment and motor mechanisms (Eugen, 1976)(8).

Light rays to reach the retina must pass through transparent optical media; the cornea, anterior chamber, lens and vitreous (Duke-Elder, 1969)(9). In order to form a focussed image, there must be an accomodative mechanisms to change the power of the eye (Robert, 1981)(10). In this mechanism the crystalline lens increases its curvature to increase the focal power of the eye thus adjusting it to clear vision of near objects (Hugh, 1980)(11). Another important mechanisms takes place which is the convergence, where the amount of convergence increases if the object get nearer and vice-versa (Robert, 1981)(10).