AN ESSAY ON:

EYE MANIFESTATIONS IN INBORN ERRORS OF METABOLISM

A THESIS
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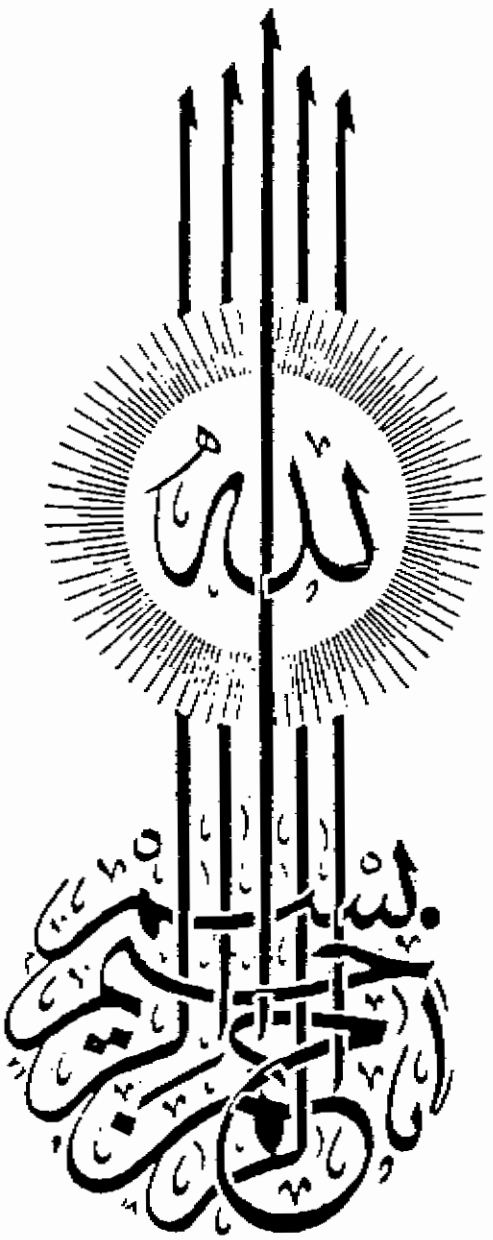

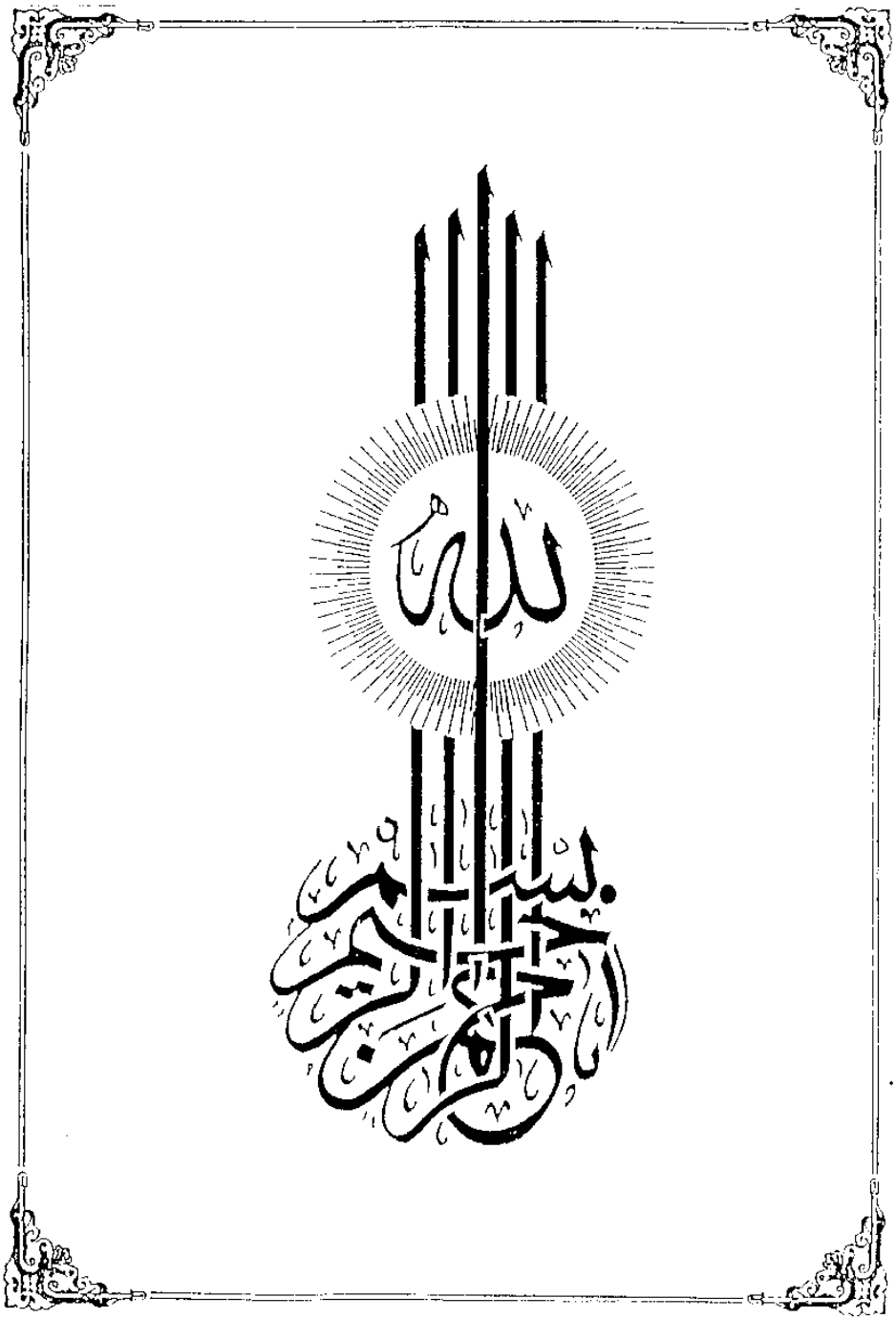

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Contents

Plate Index

INTRODUCTION

Introduction

Metabolism, by definition, comprises all the processes by which the living matter is built up (anabolism) or broken down (catabolism). Metabolic processes are controlled by two integrated inputs: the genes, which delimit the capacity of any given cell, and the environment, which determines how those genes will be expressed. It follows that all metabolic disorders result from some disturbance in the interaction between genetic and environmental factors (Rosenberg, 1980).

The term 'inborn errors of metabolism' was introduced in 1908 by Garrod who suggested that certain metabolic diseases of lifelong duration arose because the enzyme governing a single metabolic step was deficient through an inherited fault (Winder, 1977).

Inborn errors of metabolism may have their important clinical effects in almost any body system and be manifest in most aspects of pediatric medicine (Morrow and Auerbach, 1979). Eye is one of the most important organs affected by the inborn errors of metabolism.

The aim of our essay is to try to collect and summarize the important eye manifestations in inborn errors of metabolism, which may be helpful in diagnosis and follow-up.

It may be worthy to give a short account on the anatomy of the eye before discussion of our subject.

- Anatomy of the eyeball:

The eyeball (Plate I) is essentially a sphere, 2.5 cm in diameter. Its form is modified by a small anteriorly bulging segment, the transparent cornea. In its essential structure the eyeball can be resolved into three layers:

1. The outer fibrous supporting layer of sclera and cornea.
2. The middle vascular layer of choroid, ciliary body, and iris.
3. The inner layer of nerve elements, the retina.

There is a lens which lies behind the cornea.

(Woodburne and Russell, 1973)

The Chambers of The Eye:

1. Anterior Chamber:

This is the space bounded anteriorly by the posterior surface of the cornea and posteriorly by the lens, iris, and anterior surface of the ciliary body.

2. Posterior Chamber:

This is bounded anteriorly by the iris, posteriorly by the anterior surface of the lens and zonule, and peripherally by the ciliary processes.

Both chambers contain aqueous humor.

(Leeson and Leeson, 1981)

Plate I

- Diagram of the eye, Sectioned horizontally. (Leeson and Leeson, 1981).

3. Vitreous Chamber:

It lies behind the lens and ciliary processes. It is filled by the vitreous body.

(Woodburn and Russell, 1973)

Anatomy of the cornea:

The cornea forms the transparent anterior pole of the eyeball. It is about 11.6 mm in diameter. It is nearly 1 mm in thickness peripherally; but its central area is only 0.5 mm in thickness.

The cornea comprises of three main layers:

1. The epithelium is a regular layer 5-6 cells deep, and is continuous with the conjunctival epithelium.

2. The substantia propria or stroma, comprising 90% of the corneal thickness, is composed of about 60 layers of superficially - structureless fibrous lamellae, in two sets of bundles lying parallet to the surface and almost at right angles to one another.

3. The endothilium is a single layer of flattend granular polygonal cells.

* Bowman's membrane: (the anterior elastic lamina) is essentially an anterior condensation of substania propria. It offers a fair resistance, but once eroded it cannot be replaced, and an opaque scar will result.

* Descemet's membrane (the posterior elastic membrane) is a strong independent sheet separating the substantia propria from the endothelium. It readily regenerates after injury.

* The limbus is the transitional zone between cornea and sclera.

(Trevor and Poper, 1974)

Anatomy of the retina:

The retina is the innermost layer of the eyeball and comprisees an anterior, nonsensitive portion and a posterior, functional portion. The optical or neural retina lines the choroid from the papilla of the optic nerve posteriorly to the oraserrata anteriorly. It shows a shallow depression, the fovea centralis, situated about 2.5 mm to the temporal side of the optic papilla. Around the fovea is an area known as the yellow spot or macula lutea.

Layers of the retina:

In cross section, from externals to internal, the layers of the retina are as follows:

1. Pigment epithelium.
2. Layer of rods and cones.
3. External limiting membrane.
4. Outer nuclear layer.
5. Outer plexiform layer.
6. Inner nuclear layer.
7. Inner plexiform layer.
8. Ganglion cell layer.
9. Optic nerve fiber layer.
10. Internal limiting membrane.

(Leeson and Leeson, 1981)

REVIEW OF LITERATURE

HOMOCYSTINURIA

General aspects:

Homocystinurias are three biochemically and clinically distinct disorders, each characterized by increased concentration of the sulfur-containing amino acid, homocystine, in blood and urine (Rosenberg, 1980).

The basal metabolic defect:

In the most common situation, type I, the biochemical defect has been shown to be a deficiency of the enzyme cystathionine synthase, which condenses homocysteine with serine to form cystathionine (Morrow and Auerbach, 1979).

Genetics:

The occurrence of consanguinity and the familial patterns of inheritance indicate that this disorder is transmitted as an autosomal recessive trait (Nyhan, 1975).

Cardinal clinical features:

Patients with homocystinuria generally appear normal at birth. However, typical clinical features have been observed as early as one month of age.(Nyhan and Sakati, 1976).

Affected infants often show poor growth and failure to thrive. The skin tends to be coarse, wide-pored and becomes telangiectatic with aging. The hair is usually light in color. Malar flush, livido reticularis and other signs of vascular instability are commonly found (Schimke et al., 1967).

Many affected individuals have kyphosis, scoliosis,