STUDY OF THE EYE MANIFESTATIONS WITH GENETIC SKELETAL DISORDERS

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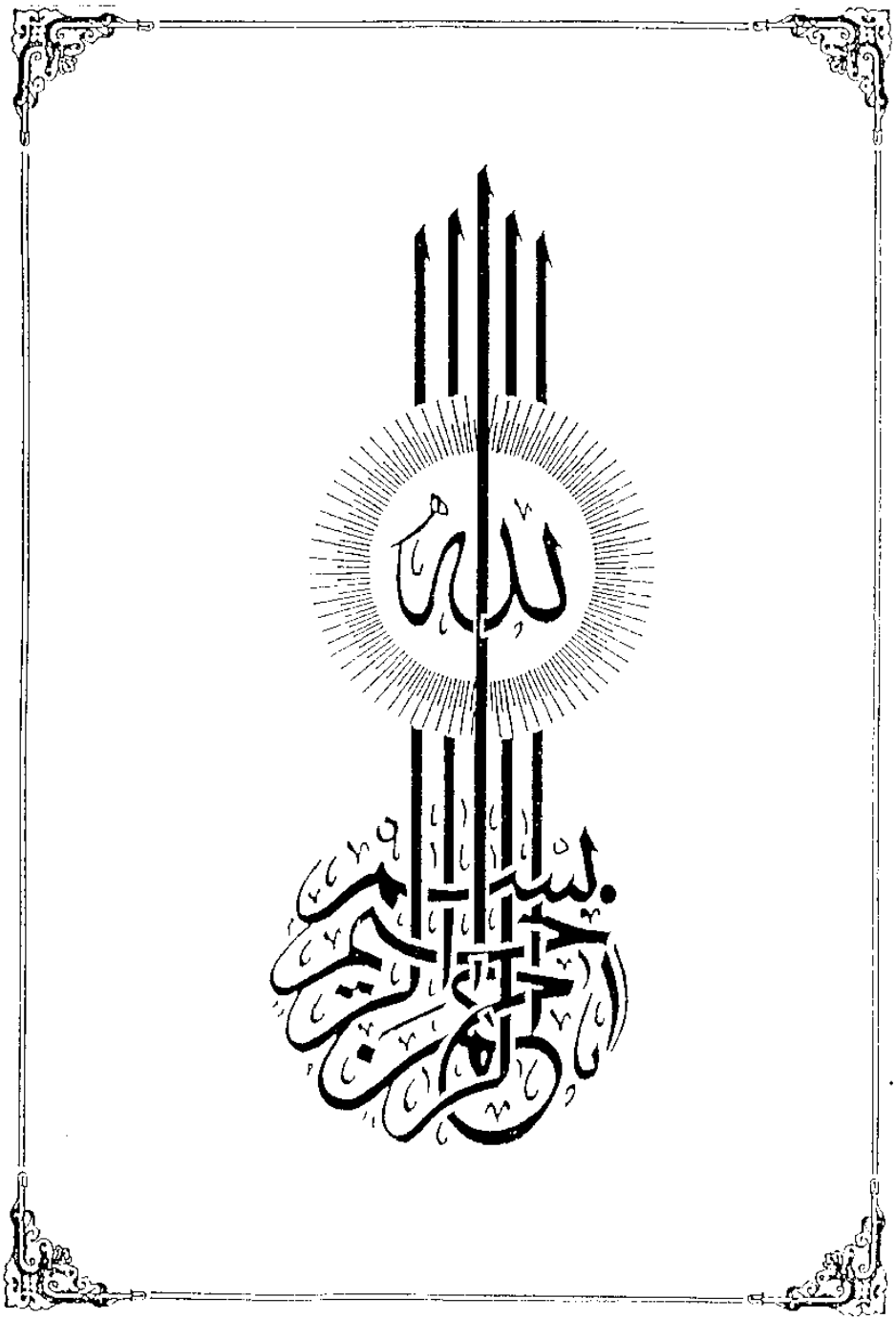

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LIST OF ABREVIATIONS

%	Percent
+/-	Before a chromosome number indicates a gain or loss of whole chromosome, after a chromosome number indicates a gain or loss of that part.
1st	First
2nd	Second
3rd	Third
A.D.	Autosomal dominant
A.R.	Autosomal recessive
X^2	Chi-square test
CNS	Central nervous system
CT	Computerized tomography
C.T	Connective tissue
C.V.S.	Cardiovascular system
Cm	Centimeter
mm	Millimeter
E.C.G.	Electrocardiogram

E.E.G	Electroencephalogram
gm	Gram
I.Q	Intelligence quotient
M.R.	Mental retardation
S.D.	Standard deviation
STORCH	Syphilis, Toxoplasmosis, rubella, cytomegaloviros and herpes simplex.
Cong.	Congenital
↑	Increase
↓	Decrease
V.S.D	Ventricular septal defect
A.S.D.	Atrial septal defect
Exam.	Examination
&	and
Chr.	Chromosome
No.	Number
D.M.	Diabetus mellitus
Aberr	Aberration

Rt	Right
Lt.	Left
Yr.	Years
gp	group
Synd.	Syndrome
+ve	positive
-ve	negative
Consang	Consanguinity
Post.	Posterior
Ant.	Anterior
Hge	Haemorrhage
G.I.T.	Gastro-intestinal tract
bet.	Between
p	Short arm of chromosme
q	Long arm of chromosme
pt	Patient

Introduction and Aim of the Work

INTRODUCTION AND AIM OF THE WORK

It has been noticed that many genetically determined disorders as connective tissue disorders, metabolic disorders, genetic syndromes and chromosomal aberrations, show eye manifestations together with skeletal manifestations e.g. Marfan syndrome is a serious connective tissue disorder, it is presented by skeletal manifestations in form of upper segment/ lower segment ratio at 2SD below mean for age, pectus deformity and kyphoscoliosis. The eye anomaly, ectopia lentis may be the key for diagnosis [*Tsipauras, 1990*].

Also congenital craniofacial abnormalities frequently require ophthalmic evaluation, e.g. in craniosynostosis as in Grouzon syndrome, hypertelorism and exophthalmos are present as eye manifestations which could aid in early diagnosis and early management [*Etheridge, 1983*].

Our aim in the present work is to study the prevalent forms of congenital eye anomalies associated with skeletal disorders to find out if these abnormalities are related to certain teratogens, metabolic disorders, connective tissue disorders, genetic syndromes or chromosomal aberrations, and to provide data that could be beneficial in early diagnosis and management of cases and in adequate genetic counselling.

Review of Literature

EMBRYOLOGY, ANATOMY AND PHYSIOLOGY OF THE EYE

Embryology of the Eye:

A short general out line of the embryology of the eye is essential to appreciate its anatomy and understand much of its patholgoy. The central nervous system is developed from the neural groove which invaginates to form the neural tube running longitudinally down the dorsal surface of the embryo.

The eye develops in the 22nd day embryo as a diverticulum from the lateral aspect of the fore brain the diverticulum grows out laterally toward the side of the head, and then becomes slightly dilated to form the optic vesicle, while the proximal portion becomes constricted to form the optic stalk. At the same time, a small area of surface ectoderm overling the optic vesicle thickens to form the lens placode. The lens placode invaginates and sinks below the surface ectoderm to form the lens vesicle. Mean while the optic vesicle becomes invaginated to form a double layered optic cup. The inferior edge of optic cup is deficient and this notch is continuous with a groove on the inferior aspect of optic stalk called the optic fissure. Vascular mesenchyme now grows in the optic fissure and takes with it the hyaloid artery. Later the fissure becomes narrowed by growth of its margins around the artery, and by the 7th week the fissure closes, forming a narrow tube, the optic canal, inside the optic stalk. By the 5th week, the lens vesicle loses contact with the surface ectoderm and lies within the mouth of the optic cup, the edges of which form the future pupil [*Sadler, 1985*].

Retina:

The retina is divided into two developmental layers.

1- The pigmented layer

2- The neural layer

The pigmented Layer:

This is formed from the outer thinner layer of the optic cup, as a result of the development of pigment granules in the cells in the 5th week of development.

The Neural Layer:

This is formed from the inner thicker layer of the optic cup.

By the 6th month of development, all the layers of the nervous portion of the retina have developed, including the rods and cones. It is thus seen that the inner layer of the optic cup may be divided into small numerous portions near the edge of the cup and a large photosensitive portion, and the two are separated by a wavy line, the ora serrata [*Ehler and Brown, 1983*].

Optic Nerve:

Walton and Robb, (1970) mentioned that the ganglion cells of the retina develop axons which converge to a point where the optic stalk leaves the posterior surface of the optic cup. Gradually the inner layer encroaches on the cavity of the stalk until the inner and outer layers fuse. The cells of the optic stalk form neuroglial supporting cells to the axons. The cavity of the stalk disappears, the

stalk, together with the optic axons form the optic nerve, the hyaloid artery and vein become the central artery and vein of the retina.

Lens:

As the lens vesicle sinks beneath the level of the surface ectoderm, the cells forming the posterior wall rapidly elongate, loose their nuclei and form a transparent lens fibers. The laminated arrangement of the lens fibers occurs as a result of additional fibers being produced and added to the outer surface of the lens by the division of the cells in the equatorial region of the lens.Mean while, vascular mesenchyme is growing into the optic cup and surrounds the developing lens. The mesenchyme immediately adjacent to the lens becomes the lens capsule, which in the earliest stages receives an abundant arterial supply from the hyaloid artery. Later this blood supply regresses and it disappears before birth [*Waardenburg, 1961*].

Ciliary Body and Suspensary Ligaments of the Lens:

The mesenchyme situated at the edge of the optic cup differentiates to form:

1) The connective tissue of the ciliary body.
2) The smooth muscle fibers of the ciliary muscles.
2) The suspensory ligament of the lens

[*Calhaun, 1983*].

The two layers of ectoderm forming the edge of the optic cup grow onto the posterior surface of the ciliary muscle and become folded to form the ciliary processes.