HEREDITARY DEAFNESS

Thesis Submitted For The Partial Fulfillment of the Degree of M.S. in Pediatrics

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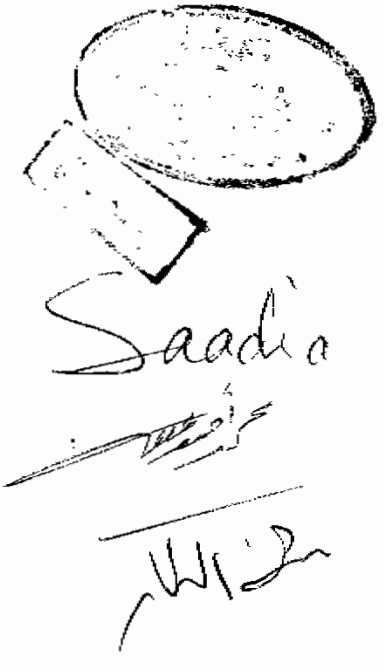

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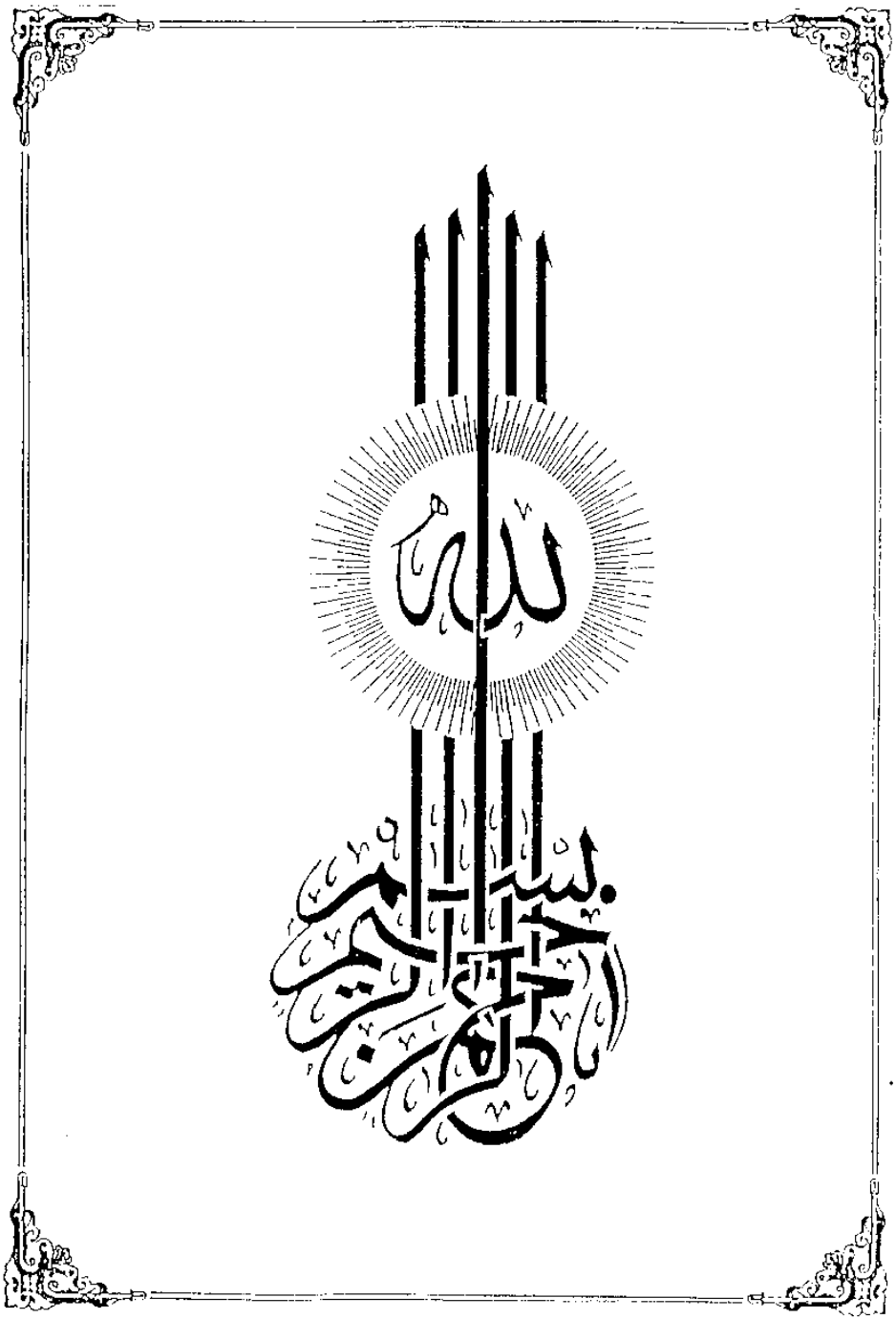

CONTENTS

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INTRODUCTION

1- **Introduction:**

Deafness can result from a wide variety of genetically determined abnormalities.

From time to time every medical geneticist is faced with a deaf child, the burden of disability is great, and the importance of early detection of significant hearing loss is very important, even mild hearing loss if not detected early can significantly retard the acquisition of language, effect intellectual development, deaf child have deminished contact with other people and outside world. Deafness impedes communication with other and places stress upon interpersonal relationships, leads to social isolation, unhappiness, even depression may result.

So this problem needs the full attention of those concerned with amelioration and prevention of genetic diseases.

2- **Aim of the essay:**

a- to concentrate upon the most common and important types of inherited deafness.

b- to outlines the clinical genetic diagnostic approach to childhood deafness.

c- this may help early detection.

d- prevention if possible.

e- to guard against complications of deafness.

DEVELOPMENT OF THE EAR

DEVELOPMENT OF THE EAR

The development of the ear and its associated structures is largely established within the first trimester of pregnancy, some clinical anomalies will arise from aberrations in this process. (Edwards 1978).

During the early stages of fetal development, a series of six visceral arches appears on the lateral aspect of the head. These mesenchymal arches form ridges in the overlying ectoderm and corresponding projections in the entoderm of the pharynx.The ridges became separated from one another by series of furrows where ectoderm and entoderm came into contact with one another. The ectodermal furrows form the visceral clefts. The entodermal furrows form the pharyngeal pouches.

The Auricle Develops from a series of six tubercles which form around the margins of the first visceral cleft (Ballantyne-Groves 1978).

External ear pinna arises from fusion of the six tubercles around the dorsal end of the 1st mandibular cleft, the external auditory meatus from the cleft itself at eighth week, (Harrison & Pracy 1984).

The tympanic membrane is formed from the membrane which intervenes between the developing meatus and the first branchial poutch from the foregut. It is formed at the 28th week as the deep meatus becomes canalized, (Edwards 1978).

The Eustachian tube, tympanic cavity are developed from the entoderm of the tubotympanic recess between the first and second visceral arches during 7th and eighth weeks.

The ossicles develops from the mesoderm of dorsal ends of the cartilages of the first and second branchial (visceral) arches and are invaginated into the tympanic cavity at the 6th week. By the seventh week the first (Mandibular) and second (hyoid) arches have developed Meckel's, Reichert's cartialges from which the head short and long process of incus and the handle of the malleus, stapedial foot plate arise (Harrison & Pracy, 1984).

The inner ear: is developed from ectoderm in the region of the hindbrain. The ectoderm invaginates to form an auditory pit which is later converted into auditory vesicle.

The membranous labyrinth is formed from the vesicle. The mesoderm surrounding it becomes the cartilaginous ear capsule which finally ossifies to form the bony Labyrinth.

The inner ear has reaches its full adult size and form by the end of the fourth fetal month. (Harrison, Pracy 1984).

FUNCTIONAL ANATOMY OF THE AUDITORY SYSTEM

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The human ear is a remarkable instrument. It usually performs its task of gathering the pressure fluctuations of an acoustic signal and transforming the information into a series of neural impulses. (Meyerhoff 1984).

The auditory system is subdivided into peripheral and central components. The peripheral component consists of three subsystems: the outer, middle and inner ear. The auditory nerve (cranial nerve VIII) is considered part of the peripheral system.

A. The central system includes the brain stem cochlear nuclei, superior olive, lateral lemniscus, inferior colliculus, medial geniculate body, auditory representation in the cerebral cortex and diversity of interconnections and pathways. Sound awareness may be brainstem phenomenan, although the appreciation of sound, its pitch, intensity, and discrimination, occurs in the temporal lobe of the cerebral cortex (Meyerhoff, Saunders 1980), Fig. 4 (mcNaught, Callander).

B. **The peripheral system:**

External Ear:

The auricle or pinna and the external auditory meatus or canal together constitute the outer ear, the canal is relatively straight in man, having a diameter of 0.7 c.m., length of 2-3 c.m. The convolution of the pinna are of considerable importance for the perception of sound directionboth horizontally and vertically. (Evans 1982) and the most important functional property of the external ear is the role it plays in sound localization. The external auditory canal is S-shaped angulating backward and upward in the lateral one third with a forward and downwards angulation in the medial two thrids. the lateral one third is cartilaginous whereas the medial two thirds is bony.

the external auditory canal not only serves as a conduit for sound energy but also it acts as a tube resonator, and also affords a measure of protection for the tympanic membrane and other middle ear structures. (Evans 1982).

Middle Ear:

the tympanic cavity (middle ear) is the space medial to the tympanic membrane and lateral to the osseous labyrinth it is approximately 15 m.m. high, 15 m.m. wide and 2.6 m.m. deep. The middle ear contains