Hearing Loss and Genetics in Humans

An Essay

Submitted in partial fulfillment of Masters Degree In Audiology

By

Mona Ahmed El-Akkad

(MB, Bch)

Supervised By

Prof. Dr. Ahmad Sameh Farid

Professor of ENT and Audiology
Head of Audiology Department
Faculty of Medicine
Cairo University

Prof. Dr. Mohamed Ibrahim Shabana

Professor Of Audiology
Faculty of Medicine
Cairo University

Faculty of Medicine
Cairo University
2002

SUMMARY

This thesis was planed to study different theoretical aspects of human genetic hearing loss.

A- The studying included the academic basis of old and recent sciences of inheritance which comprise: Mendel's inheritance pattern, chromosomes (nature and karyotyping) and chromosomal constitutional molecules; nucleoproteins.

Nucleoproteins are the subject science Molecular Biology previously termed Genetic Engineering. Molecular biology studies included the king molecule of the whole body DNA. The DNA in all cells of a human is the same whatever the function of the is. Studying, diagnosis, and treatment of any genetic disease needs detailed uncomprehensive of DNA; chemistry, function, expression and mutations. Molecular Biology investigations depend on different recent techniques. A fantastic tremendous technique controlling genetic social and medical status of every person, societies, nations, and Man is the **gene project.** Gene project includes accurate and detailed identification of DNA sequences. Referring every special collection of sequences to its specific site (locus) on the chromosome is **gene mapping.** Gene mapping facilitates diagnosis of mutations in genetic diseases.

In addition, the gene project will point to the genetic I.D fingerprint. The genetic finger print sequence resides in the non-codon areas of DNA.

B- The clinical aspect of the study:

1. Definitions of hearing loss.
2. Monthly maternal observations of infants to pick and criteria of shortage in hearing of the baby.
3. Measurements of degrees of hearing loss.
4. Prevalence.
5. Types of hearing loss and syndromes; classified both according to clinical presentation as well as according to types of inheritance.

C- Clinical aspects of clinical biology:

1- Identification of genes involved in deafness.

2- Special considerations of the protein connexin 26, its normal expression and physiological

Function. Chromosomal loci and gene mutations.

D- Diagnosis and treatment of genetic hearing loss:

1- Clinical examination.

2- Usual investigations.

3- Genetic testing and evaluation (interpretation).

4- Treatment is double based:

a- Genetic counseling: this includes family planning combined decision of the physician and family, supports group and follow-up.

b- The final conclusion might be gene therapy, which means recombinant DNA application or surgical according to the judgment of the expert physician.