

# Essential Roles in Etiology and Prevention of Disabilities

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## Abstract

In this study 100 numbers disable persons and their files were evaluated. The most of them were mental retard. Some of them had congenital disability. Also some of them had disturbance in vocal and Hearing or vision. Hereby between of study group had seizure. Rarely of them were Down syndrome (trisomy 21) patient. However some of genetically disease such PKU (Phenyl ketone uria) and FMF (Familial Mediterranean fever) and major  $\beta$  thalassemia and hemophilia were reported.

**Keywords:** disable persons; hearing; vision

## Introduction

The most of disabilities are made through Family marriages. Also pregnancy in very young women and very old women are etiology of some congenital disabilities. However Physical and motor disabilities such as club foot were reported from very young ages women who were in their first pregnancy. It might be drug excessive consumption and malnutrition and stress and x-ray other interfering factors lead to disabilities in pregnancy duration. Some of researches showed that seizure and fever illness were made mental retardation in duration of newborn and child. Some of tests are important in Medical Genetic counseling in premarriage and before pregnancy. They are classified as a predictive test, which means its results report the likelihood of chromosomal abnormalities. These tests are double marker, triple marker and quad marker [1-5].

## Methods and Results

Some of molecular and cellular clinical laboratory tests were made on samples of study groups. These tests were confirmed their illness. So connexin gene mutation disease was reported to hearless people through PCR (Polymerase chain reaction) test. This study showed that the children who had defect in above mentioned gene they were deaf. So some of them received inner cochlear implant. It is requirement to use some minerals and vitamins including folic acid with a physician's prescription before deciding to pregnancy. The use of defroxamine drug to treatment of iron overload in major  $\beta$  thalassemia patients is necessary. Also antiseizure

drugs such phenobarbital and sodium valproate are recommended. However the FMF patients received colchicine drugs. [6-10]

## Discussion

The double marker test, also known as maternal serum screening, is part of a more comprehensive screening called the first trimester screening. Levels of  $\beta$ -hCG and PAPP-A may be either higher or lower than "normal" in pregnancies with chromosomal abnormalities. The triple marker blood test is conducted in the second trimester. It examines the serum levels of three hormones: Alpha-fetoprotein (AFP), Unconjugated estriol, Human chorionic gonadotropin (beta-hCG). Triple marker down syndrome is most commonly used to detect Down Syndrome (a chromosomal disorder) between 15 to 18 gestation weeks. The quad marker screen is a blood test that provides useful information about a pregnancy. That can help estimate baby's risk of Down syndrome, Edward's syndrome (trisomy 18), and neural tube defects (NTD). The blood sample is sent to a laboratory and tested for the presence of the following four things that are normally found in the baby's blood, brain, spinal fluid, and amniotic fluid: Alpha-fetoprotein: A protein produced by the baby's liver and other organs, Unconjugated estriol: A protein produced in the placenta and in the baby's liver, Human chorionic gonadotropin: A hormone produced by the placenta, Inhibin-A: A hormone produced by the placenta. [1,2,8,11,12]

## Conclusion

Prevention from very close familial marriage will be important role in lacking of disabilities percentage. Also prevention of arbitrarily drugs consumption in pregnancy duration are necessary. Medical Genetic counseling in premarriage and before pregnancy are recommended [1, 2].

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