# ESSAY ON THE VALUE OF PRENATAL SCREENING TESTS IN CHILDHOOD DISORDERS

*THESIS*

*Submitted for the Partial Fulfillment of*

*Master Degree in Pediatrics*


*PRESENTED BY*

ALAA EL DIN HUSSEIN ABUL-FETOUH

(M.B., B.CH.)

*SUPERVISORS*

Prof. Dr. MOHAMED FOUAD BADRAWY

Professor of Pediatrics,

Faculty of Medicine

Ain Shams University,

Dr. KHALID S. AWWAD

Lecturer of Pediatrics,

Faculty of Medicine,

Ain Shams University,

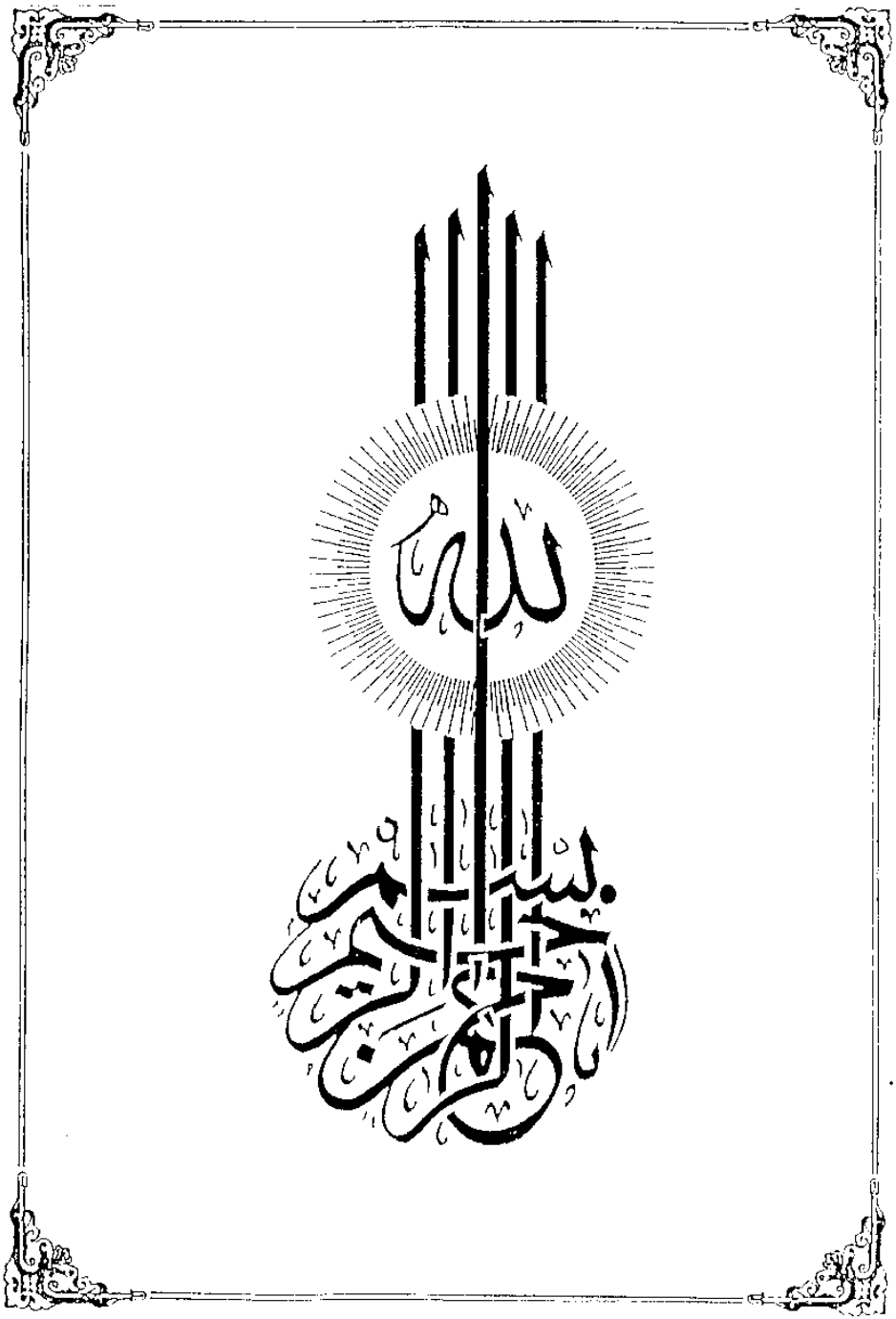

1988

# ACKNOWLEDGEMENT

*I would like to express my deep thanks and gratitude to* PROFESSOR DR. FOUAD BADRAWY, *professor of pediatrics, Ain-Shams university, for giving me the privilege of working under his supervision, for his encouragement and unfailing guidance throughout this thesis.*

*Also, I would like to express my deep thanks and gratitude to* DR. KHALID AWWAD, *lecturer of pediatrics, Ain-Shams university, for his kind patience, great support, great help and unending guidance in preparing and finishing this thesis.*

*Lastly, I owe my thanks and gratitude to every one who participated, in some way or the other, to let this thesis comes to such a final picture.*



## CONTENTS

## List of Abbreviations

**AF** : Amniotic Fluid.

**AFAFP**: Amniotic Fluid Alpha-Feto-Protein.

**AFP** : Alpha Feto-protein.

**CVB** : Chorionic Villi Biopsy.

**IUGR** : Intra-Uterine Growth Retardation.

**MoM** : Multiples of the Median.

**MSAFP**: Maternal Serum Alpha Feto-Protein.

**NTD** : Neural Tube Defect.

**US** : Ultra-Sonography, UltraSound.

## List of Tables

## List of Figures

# INTRODUCTION

# INTRODUCTION

Congenital abnormalities occur in 3-5% of live births. A great proportion of these infants will die in childhood or suffer chronic mental or physical disabilites. In addition, the birth of infants with such conditions has a destructive effect on the family and a profound impact on the community as a whole (Nicolaids & Rodeck, 1984).

Prenatal diagnosis is the ability to detect genetic disorders or other fetal defects early in pregnancy. It represents one of the most exciting advances in medicine, and is rapidly becoming an important tool of preventive medicine (Gerbie & Ellias, 1980).

It should be clear that prenatal diagnosis is not restricted to genetic disorders. It is the diagnosis of congenital conditions rather than solely genetic disorders which are of interest. In

fact, monitoring pregnancies for acquired illnesses has been a common obstetric practice for many years, and has been used in counselling couples regarding early termination of pregnancy (Nadler, 1987).

Prenatal diagnosis should be discussed early in pregnancy; this allows time for counselling and risk estimation so that couples can carefully consider associated risks, and their decision should a test prove positive. Diagnosis of fetal defects amenable to surgery allows optimal timing, place, and mode of delivery. A normal test result does not guarantee a normal baby; many fetal defects are undiagnosable with current techniques and the couple should be aware of the limitation of these tests (Donnai & Gowland, 1983).

Before undergoing prenatal diagnosis, the patient or, ideally, the couple should be informed regarding:

(1) The disorder(s) for which they are at increased risk,

(2) The occurrence or recurrence risks for the disorder(s), and

(3) Methods of heterozygote detection (if applicable), antenatal diagnosis, and alternative reproductive options, if relevant (Lippman-Hand & Fraser, 1979).

Many approaches to prenatal diagnosis have been tried. Attempts to visualize the fetus radiographically have, until recently, been disappointing because of the incomplete mineralization of the fetal bones early in gestation and the gestational hazards of ionizing radiation for the developing organism (Johnston & Forfar, 1978).

Since the late 1960s, tremendous advances have occured in the prenatal diagnosis of fetal anomalies. Initially, amniocentesis was the sole procedure utilized. Successful cultivation of the amniotic

fluid cells permitted the diagnosis of chromosome abnormalities and a variety of inborn errors of metabolism. This was rapidly followed by the use of ultrasonography, and determination of amniotic fluid alpha-fetoprotein for detection of neural tube defects. The later half of the 1970s witnessed the development of fetal blood sampling, fetal biopsy and the introduction of maternal serum alpha-fetoprotein screening (Slack, 1986).

The early 1980s witnessed two major developments that have had significant impacts on the utilization of antenatal diagnostic services:

(1) Chorionic villi sampling provided the opportunity to extend intrauterine diagnosis to the first trimester of pregnancy. It has a number of advantages over amniocentesis, which are primarily related to the earlier time at which it can be performed as well as to a more rapid laboratory diagnosis. The major drawback relates

to a risk factor which may be higher than that of amniocentesis. Almost all diagnoses that could be made in the second trimester can now be made reliably in the first trimester through this new diagnostic procedure (Brambati & Oldrini, 1986).

(2) The utilisation of the powerful techniques of modern molecular biology has made it possible to no longer rely solely on the expression of gene products. Three approaches-namely DNA hybridization, restriction endonuclease analysis, and linkage analysis - are rapidly expanding the number of disorders detectable in utero (Rodeck & Morsman, 1983).

Increasingly sophisticated instrumentation permits the detection of many fetal anomalies, and we are beginning to understand the natural history of many of these defects. The coming decade will see even more impressive advances in the area of prenatal diagnosis and treatment (Nadler, 1987).

## AIM OF THE ESSAY

It aims to catalogue the multiple, newly developed screening tests used for prenatal diagnosis of fetal disorders and their value in the prevention, early detection, better management and decreasing the incidence of a wide variety of childhood disorders.