

ESSAY ON PRENATAL DIAGNOSIS.

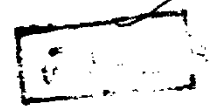
FOR PARTIAL FULFILLMENT FOR
MASTER DEGREE OF PEDIATRICS.

BY
HAFEZ ABDEL RAHMAN HAFEZ
M.B., B.Ch.
FACULTY OF MEDICINE
AIN SHAMS UNIVERSITY

6/8.92075
H.A



18525



UNDER SUPERVISION OF:
PROF. DR. MOHAMMAD A. AWAD-ALLAH
PROF. OF PEDIATRICS
FACULTY OF MEDICINE
AIN SHAMS UNIVERSITY

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1983

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بِسْمِ اللَّهِ الرَّحْمَنِ الرَّحِيمِ

”وَلَقَدْ خَلَقْنَا الْإِنْسَانَ مِنْ سُلَالَةٍ مِنْ طِينٍ . ثُمَّ جَعَلْنَاهُ نُطْفَةً
فِي قَرَارٍ مَكِينٍ . ثُمَّ خَلَقْنَا النُّطْفَةَ عَلَقَةً فَخَلَقْنَا الْعَلَقَةَ مُضْغَةً
فَخَلَقْنَا الْمُضْغَةَ عِظْمًا فَكَسَوْنَا الْعِظْمَ لَحْمًا ثُمَّ أَنْشَأْنَاهُ خَلْقًا
آخَرَ فَبَارَكَ اللَّهُ أَحْسَنُ الْخَالِقِينَ“

صَدَقَ اللَّهُ الْعَظِيمُ
(سورة التوحيث ١٢-١٤)



TO MY PARENTS

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LIST OF CORRECTIONS

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8	10	Brown (1958) (Donald and Brown).	Brown in 1958 (Donald and Brown).
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99	2&3	Ferguson-Smith et al 1971	Ferguson-Smith, 1971
101	10	Ryan, Lee, and Nadler, 1972.	Ryan et al, 1972
101	14,15	Russel, Russel, and Littlefield, 1972	Russel et al, 1972
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146	13	vluid	fluid
147	11	bluid	fluid
147	15	embrylogy	embryology
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172	3	vluid	fluid

I N T R O D U C T I O N

INTRODUCTION

From implantation to delivery, the human fetus lives for 38 weeks in the uterus, an encapsulated organism, shut off from the external world, dependant upon the umbilical and placental circulation.

Since the beginning of the second half of this century, knowledge of the fetus and his environment has increased remarkably. The intra-uterine life was considered as a mystery because of the inaccessibility of the fetus. Recently, a revolution has occurred in this field so as to facilitate not only intrauterine fetal monitoring but also diagnosis of so many diseases of the fetus in the prenatal period.

The term of prenatal diagnosis applies to diagnostic procedures employed for the identification of disease in the fetus when interruption of the pregnancy is under consideration, and in instances in which direct treatment of the fetus may be possible. (Vaughan et al, 1979).

The main procedures achieved in prenatal diagnosis are amniocentesis, ultra-sonography, amnioscopy,

fetoscopy, amniography, fetography, fetal heart monitoring, and placental biopsy. The use of these methods has led to diagnosis of so many diseases of the fetus prenatally some of which can help in Genetic counseling.

Genetic counseling is a process of communication dealing with the human problems associated with the occurrence or risk of occurrence of a genetic disorder in family.as in consanguineous marriage, Rh incompatibility, carrier detection and in determination of paternity. Those who commonly receive it are couples whose first child has just been born with a birth defect or medical problem. Older parents also are frequently concerned about genetic risks and wish to learn about prenatal diagnosis. Others seek information prior to marriage or before having children, because of medical problems of their relatives. Indeed, pre-marital and pre-conceptual periods are the optimum way to try to decrease the incidence of most genetic disease (Vaughan et al., 1979).

So this essay aims to catalogue the multiple, newly developed technics used for prenatal diagnosis as well as the diseases which can be diagnosed prenatally by them, some of which can help in genetic counseling.

REVIEW OF LITERATURE

I- HISTORY

HISTORY

Prenatal diagnosis is one of the most rapidly developing areas in medicine, not only in regard to the number of patients evaluated, but also to the number of conditions which may be diagnosed. Prior to the 1960s, prenatal diagnoses were made infrequently. During the 1960s, however, techniques for chromosomal and biochemical analyses of amniotic fluid cells were developed, and routine prenatal evaluation became a reality (Nadler and Gerbie, 1970).

Subsequently, expanded utilization and application of prenatal diagnosis have occurred, due in part (1) to other technologic advances, including α -feto-protein (AFP) assay techniques for detecting neural tube defects, the application of ultra-sound for the diagnosis of physical abnormalities, and the development of fetoscopy for direct visualization of the fetus, skin biopsy, and the sampling of blood, and (2) to the increased use of radiologic examinations (Stephenson and Weaver, 1981).

As a result of the widespread publicity, concerning prenatal diagnosis, many physicians are now

being consulted in regard to the diagnosability of specific disorders.

As of January 1981, 182 separate fetal conditions have been reported to be diagnosed in utero (Tables 6 & 7). In this chapter, a brief historical review of the various techniques used will be dealt with.

Amniocentesis is one of the most widely used of the prenatal diagnostic procedures and allows for the detection of a number of biochemical and chromosomal disorders.

It has been used diagnostically since the mid-1930s for detecting fetal distress and, more recently, for following the progress of Rh hemolytic disease. In the mid-1950s it was found that fetal cells in the amniotic fluid could be used to determine fetal sex (by observing the sex chromatin) and blood type. In the late 1960s techniques were developed for obtaining Karyotypes (Steele and Breg, 1966), and enzymatic assays on fetal cells, and prenatal diagnosis began to revolutionize genetic counseling (Nadler, 1968).

Amniotic cells which originate from fetal skin, amnion, and the gastro-intestinal, respiratory, and genito-urinary tracts (Huisjes, 1978) are found within the amniotic fluid. These cells, after culture, may be analyzed for fetal enzyme activity or for fetal chromosomal constitution by employing techniques similar to those utilized for fibroblast analyses. Although several enzyme deficiencies, such as hexosaminidase A deficiency in Tay Sachs disease (O'Brien et al, 1970) and iduronate sulfatase deficiency in Hunter syndrome (Liebaers et al, 1977), have been diagnosed from uncultivated cells or the amniotic fluid alone, most investigators are of the opinion that cultured cells provide the greatest diagnostic reliability. The major disadvantage of using cultured cells, however, is that 10 days to 4 weeks are frequently required before the cultures are ready to be analysed. This time period may represent a significant delay in establishing diagnoses. Micro-techniques, which require smaller numbers of cells for analyses, and thus less culture time, are being developed (Niermeijer et al, 1975).

Concentrations of AFP are determined from amniotic fluid directly. The concentration of this protein in this fluid varies throughout pregnancy, reaching a peak