PARENTAL REACTION TO HAVING A CHILD WITH DOWN'S SYNDROME

A Thesis
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By

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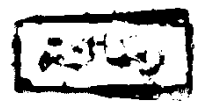

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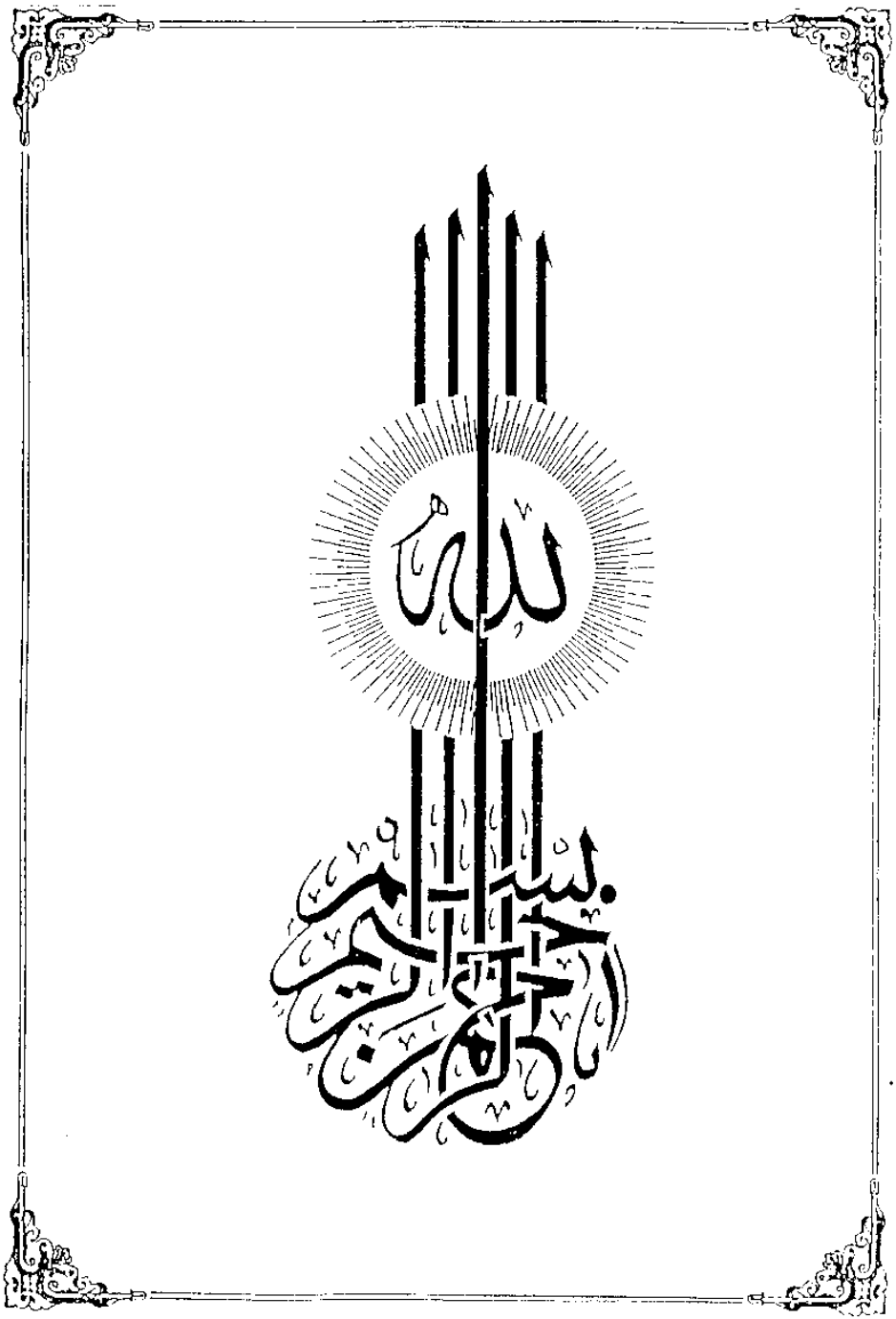

CONTENTS

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INTRODUTION

INTRODUCTION

Down's syndrome is a condition with life long implications for physical appearance, intellectual achivement and general functioning of the child. Parent whatever their race, religion of socioeconomic back ground are devastated to learn that their new baby has Down's syndrome. Frightene by the implications of this disorder with respect to the child appearance, intelligence, capacity for independent functioning, their joy in the new baby is compromised (Moos, 1977).

It is true that all parents find the birth of a Down's syndrome baby a great shock, they often confused and can not believe it is true. Depending on their experience, personality, values, judjments about life and children, they may have a number of reactions to the birth of the child (Cunningham, 1988).

The family may have a potent effect on the childs' development and the child may in turn have an impact on the well being of the family members. The provision of effective support for families of mentally handicapped children tests upon a sensitive understanding of the stresses which the family face. This understanding may depend on assumptions which have been made about the way in which family has been coping and whether the family is

emotionally overwhelmed by chronic grief, broken down by chronic stress or do not cope because they have no knowledge of what to expect. (Russell, 1985).

This explain how important is the determination and understanding of parental reactions to having a child with Down's syndrome who is primarly mentally retarded as that will help much in providing effective support for parents helping them to cope with the condition, plane for the future of the child and suggesting the better methods of behaving with the child.

AIM OF THE WORK

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Evaluation of children with Down's syndrome physically and intellectually.

Determination of parental reaction to having a child with this condition.

DOWN'S SYNDROME

Down's syndrome is one of the most common conditions associated with chromosomal aberrations occurring approximmately once in each 600 births (Benda, 1969). It's importance in the overall problem of mental retardation is indicated by the fact that it is found in 10 to 15 % of institutionalized mentally defective individuals.

The prevalence in population is less than the incidence at birth, because of the high rate of infant mortality of the syndrome, about 1 in six dying in the first year of life. Average expectation of the life is about 16.2 years. Penrose and Smith (1969), Lilienfeld and Benescott, 1969), although many persons with Down's syndrome survive to ages over 50 and 60 (Oster et al.,1975).

A recent increase in the incidence of Down's syndrome has been found, mainly affecting young mothers. It is concluded that this increase is due to the environment and an association with the contraceptive pills is thought (Read, 1982). Interestingly enough, oral contraceptive pills have only been used to a limited extent in Japan where no increase in age specific incidence could be shown. (Matsunage and Fujita, 1977).

AETIOLOGY

Chromosomal abnormality in patients with Down's syndrome was discovered by Le Jeune et al.,(1959). In the majority of cases the chromosomal abnormality is an additional small acrocentric chromosome or trisomy 21.

A high correlation exists between increasing maternal age and the non disjunction resulting in the presence of an extra chromosome in the offspring. Heteromorphisms on fluorescent staining have furnished cytologic proof for the parental origin of non disjunction in a number of instances. Evidence is accumulating that abnormal segregation is parental in origin in approximately on third of cases (Cohein and Nadler, 1983).

Three types of chromosomal aberrations are found in Dowr's Syndrome:

1. Non disjunction:

Patients have 47 chromosomes with an extra chromosome 21. The Karyotypes of the mothers are normal a non disjunction occurring during meiosis is held responsible for this disorder. Majority of Down's syndrome patients fall in this group.

2. Translocation:

There is fusion of two chromosmes mostly 21 and 15, resulting in 46 chromosomes in 46 chromosomes in affected

patients, despite the extra chromosome material.

3. Mosaicism:

Non disjunction occurring in any cell division after fertilization will result in Mosaicism where both normal and trisomic cells are found in various tissues. (Cytryn and Lourie, 1968).

The Risk of a Down's child:

The risk of a Down's child for any mother varies with:

a- her age.

b- her karyotype and that of the father.

c- Previous history of having Down's child on the another trisomies.

On the average, the recurrence risk of Down's child (or other trisomy) after the birth of one such child is about 1 percent regardless of maternal age.

There is an increasing risk of Down's child with advanced maternal age. The average maternal age at birth of a Down's child is about 34 years the Non disjunction group "age dependant while the translocation. While the average risk of a Down foetus is 1/1500 at the maternal age period of 20 - 29 years, it rises to 1/800 at 30 - 34 years and 1/260 at 35 - 39 years and 1/100 or more above 40 years old.

The recurrence risk in translocation Down depends on the karyotype of the child and parents not on the age of the mother. In a parent with a 21 - G translocation, the risk of producing 21 - trisomic offspring depends on whether the translocation is 21/22 or 21/21. In the former case the risks are similar to those in 21/D translocation (1 :3) while if it is 21/21 translocation the risk rises to 100 % of producing a 21 trisomic offspring.

Since mosaic 21 trisomy is the result of postfertilization error in cell Division, it is not correlated with advanced maternal age . However, they themselves may be at high risk of having Down's children if the mosaicism extend to the cells of the germ line. Occasionally females with 21 trisomy have children on the average 50 % of their children are Down's syndrome and 50 % a normal because they produce two types of gametes one with 23 chromosomes and one with 24 chromosomes with an extra 21 "secondary non-disjunction".(Kashaba and Salah, 1988).

Prevention of Down's Syndrome:

It is not possible to prevent nondisjunction or chromosomal rearrangement prior to conception, so that we will continue to be faced with the problems of detecting and preventing chromosomal abnormalities during gastation and preverting chromosomal abnormalities during gestation and prior to the birth of an abnormal and mentally retarded child.

Amniocentesis:

Is used in second triminster for detection of chromosomal abnormalities in certain pregnancies. It involves the aspiration of a small sample amniotic fluid culturing of fetal cells contained in the fluid and determination of the karyotype of these cells and thus of the fetus.

The major indications for the use of this technique for detection of chromosome abnormalities are:

* The birth of a previous child with chromosomal trisomy (cases in which the mother has had a previous trisomic infant or possibly a previous spontenious abortion or perinatal death in which the neonate or the abortus was trisomic.

* The detection of the presence of parental chromosome abnormality (one parent is a balanced translocation carrier particularly if rearrangement was detected as a result of a previously clinically abnormal infant.

* Maternal age, it should be offered to all mothers over the age of thirty four at the time of delivery. The safty and reliability of amniocentesis as a diagnostic technique has now been well established and it is generally accepted that it inrease the risk of spontenous abortion by between 1 in 250 and 1 in 500 over the risk, without

any intervention other risks of the test including fetal and maternal morbidity are very low. In comptent hands the test has been shown to have a near 100 percent reliability for detection of chromosomal abnormalities only.

The major disadvantage of the test is that it can prevent the birth only of a relatively small proportion of infants with a chromosome anomaly, namely those cases of Down's syndrome or other anomalies born to mothers who are over the thirty five or who had previous abnormal infants.

The posibility of an earlier prenatal diagnostic test now exsists called: Chorion villus sampling.(Clarke et al.1983).

Chorion Villus Sampling (CVS):

Involve transcervical sampling of villi of chorion frondusum between seven and ten weeks of gestation (Rodeck et al., 1983). Then simple direct chromosome studies within twinty four hours of sampling are posible. (Simoni et al., 1983), such test if it proves to be safe and reliable in clinical trials and if it is acceptabable to a wide segment of the population, may become available to a much larger group of mothers than can at present be offered amniocentesis.

This would allow the rapid testing of large numbers of pregnancies early in gestation with a possibility of terminaling abnormal pregnancies early in the first trimester.