ROLE OF ULTRASOUND IN DIAGNOSING FETAL ANOMALIES

Essay

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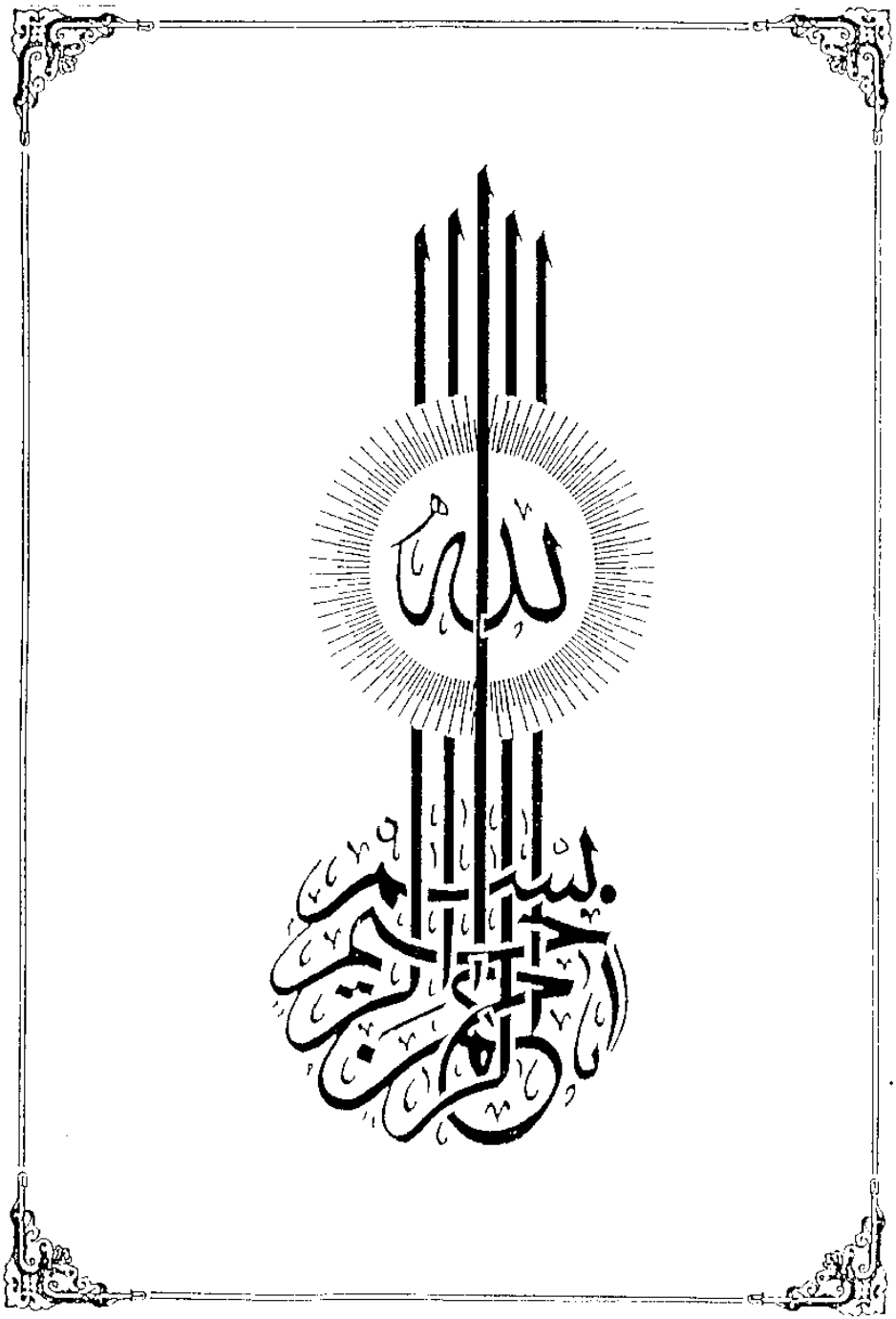
This work is dedicated with my love to my dear parents.



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Introduction

Introduction

The risk of a newborn infant having a congenital abnormality, be it major or minor, is two per cent. Congenital abnormalities account for approximately 25% perinatal mortality in the UK. Significant anomalies occur in 32% stillbirths and 1.6% live births ***(Ellis and Rodeck, 1988)***. An incidence up to 15.5% of liveborn infants was also reported including major, minor, and multiple anomalies, sequences, and syndromes, ***(Chung and Myrianthopoulos, 1987)***. Major anomalies are found at a higher rate in those babies who die in the perinatal period and the defects frequently contribute to their death ***(Mueller et al., 1983)***.

Magnitude of the problem: One measure of the importance of congenital anomalies is their contribution to infant death. While perinatal mortality due to prematurity, asphyxia, and infection has decreased due to improvements in their treatment and in perinatal care, however at the same time, the contribution of congenital anomalies to the overall perinatal death rate has increased ***(Romero et al., 1991)***. In the United States, congenital anomalies and prematurity were cited as the leading cause of infant mortality ***(Oakely, 1981)***.

As regards the impact of congenital disease on morbidity, it has been estimated that one of every four hospitalized children is affected by a disease that is at least partially genetically determined, and 1 of every 20 is affected by diseases that are completely genetic in origin ***(Emery and Rimoin, 1983)***.

As regards the impact of those defects on the individuals or their families, there are; the emotional upheaval, the financial burden, and the loss of physical or intellectual abilities associated with birth defects ***(Weaver, 1987)***. The incidence of divorce and sibling social maladjustment is greater in families of children with spina bifida than in families of infants without congenital anomalies. ***(Lorber, 1978 and Main, Mennuti, 1986)***. The impact on the individual depends mainly on the severity, the location, and the long-term problems associated with the defect ***(Weaver, 1987)***.

Ultrasound has undergone a transformation that has allowed to answer not only the basic question whether the patient is pregnant, but also whether a fetal anomaly is detected ***(Callen, 1988)***.

The cardinal principle behind the diagnosis of congenital anomalies with ultrasound is recognition of a departure from normal fetal anatomy. Congenital anomalies are generally recognized with ultrasound by one of the following means: 1. absence of a normal anatomic structure, 2. a disruption of the contour, shape, location, sonographic texture, or size of a normal anatomic structure, 3. presence of an abnormal structure, 4. abnormal fetal biometry, or 5. abnormal fetal motion ***(Romero et al., 1991)***.

When a fetal anomaly has been thoroughly documented, the fetal diagnosis may be used to alter obstetric management. After appropriate genetic counselling of the patient and with informed consent:

1. The pregnancy may be terminated because of the presence of a lethal anomaly, for example, anencephaly.

2. The anomaly may be appropriately treated or corrected after a term delivery, for example, omphalocele.

3. The timing or type of delivery may be altered, for example, a Cesarean section for hydrocephalus.

4. The anomaly may be treated in utero, for example, catheter drainage of the obstructed fetal bladder into the amniotic cavity ***(Hill et al., 1988)***.

5. Delivery may be arranged to be accomplished at a facility capable of immediate repair, for example, the diaphragmatic hernia and handling of the resultant respiratory problems ***(Weaver, 1987)***.

Aetiology of Fetal Anomalies

Chapter One
Aetiology of Fetal Anomalies

Most birth defects can be caused by *(Nevin, 1982, Iams, 1982)*: (1) genetic abnormalities (either chromosomal or single gene disorders), (2) environmental factors, or (3) multifactorial conditions caused by the interaction of several abnormal genes, each with small detrimental effects and environmental influences.

(I) Genetic Abnormalities

a) Chromosomal disorders:

Since about 10% of spermatozoa and about 50% of ova are cytogenetically abnormal, it is not surprising that approximately 7.5% of all conceptions have a visible chromosome abnormality *(Boue et al., 1985, Wramsby et al., 1987)*. The vast majority (95%) of these abnormal conceptions abort spontaneously and account for the 50% incidence of chromosomal abnormalities amongst early abortions. This proportion declines with advancing gestational age to become 5% in late spontaneous abortions. Amongst liveborn infants about 1 in 160 has a major chromosomal disorder *(Tolmie, 1989)*.

Many chromosomal abnormalities have been identified and show a wide spectrum of clinical manifestations. Some result in spontaneous abortions and stillbirths (trisomy 16); others produce a severely abnormal infant who usually dies shortly after birth (trisomies 13 & 18); others lead

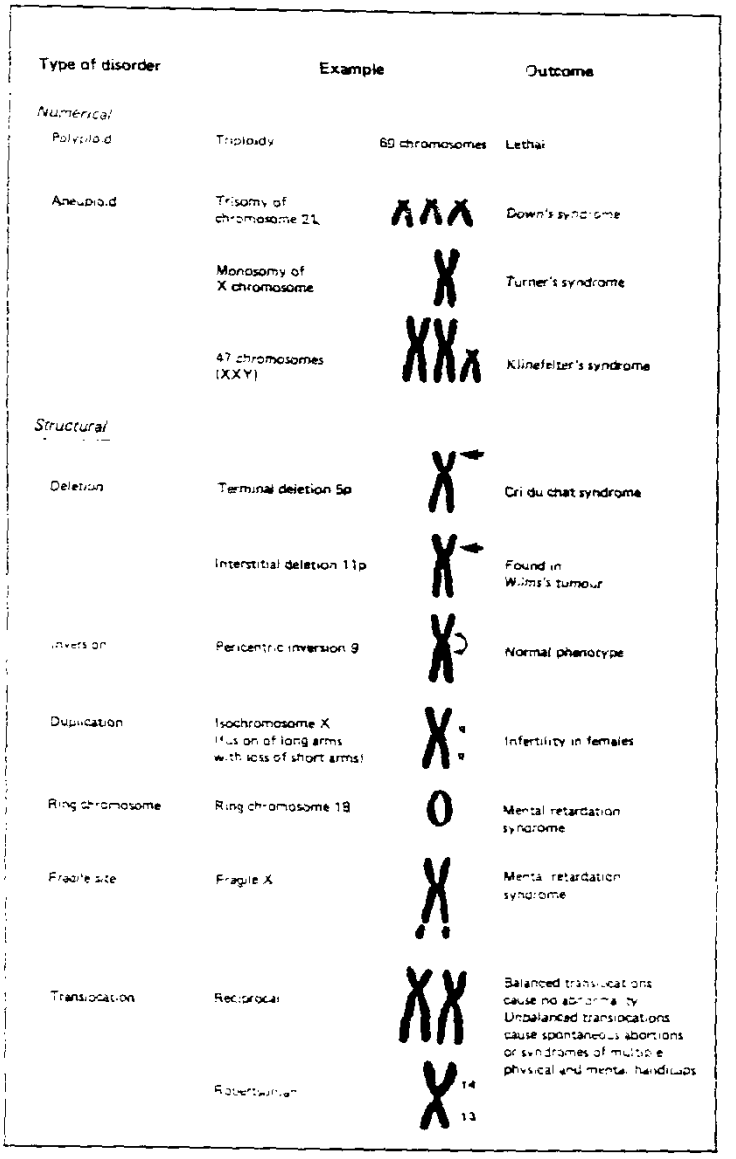

Fig. (1): Types of chromosomal disorders.
(Kingston, 1989).

to a severely mentally retarded child (trisomy 21 = Down's syndrome) and still others, although permitting relatively normal physical development, may be associated with some degree of intellectual impairment and sterility (Turner's and Klinefilter's syndromes) ***(Borgaonkar, 1980)***.

Chromosomal abnormalities may involve the x-chromosomes or the autosomes. Chromosomal aberrations may be numerical or structural ***(Iams, 1982)***:

1. ***Numerical anomalies*** are represented by a deficiency or excess of one or more entire chromosomes and result in a chromosomal complement that is not a multiple of the haploid number of 23 and is called aneuploidy (e.g. 47 + 21 or trisomy 21). Aberrations involving entire haploid sets of 23 are called polyploidy (e.g. 69 poly ploidy) which is very rare among live-born infants ***(Iams, 1982)***.

Aneuploidy of autosomes is always associated with fetal abnormalities. Numerical anomalies of sex chromosomes may not have an obvious phenotype expression. Autosomal trisomies for chromosomes 21, 13 and 18 are the most common. Sex chromosome trisomies are not as harmful as autosomal trisomies. Monosomy in which only one pair of homologous chromosomes is present is never seen in live born infants. Monosomy for the x chromosome (45, x) is compatible with live birth and normal intellectual function ***(Iams 1982)***.

Numerical anomalies are almost all mutations, while structural rearrangements due to breaking and rejoining of fragments between or within chromosomes are often inherited ***(Hutchins and harvey, 1984)***.

2. ***Structural abnormalities*** may or may not result in phenotypic changes ***(Iams, 1982)***. Structural anomalies may be due to partial loss (deletion) of chromosome material, or rearrangement of genetic material as in interchange (translocation) chromosomes, inversions or other abnormalities ***(Smithells, 1978)***.

Translocations of fragments between chromosomes where the total amount of genetic matter is normal are said to be "balanced" and rarely show their presence clinically ***(Evans, 1977)***. About a quarter of the offsprings will carry the same translocation; about a quarter will inherit normal chromosomes; and about half will possess an unbalanced set ***(Hutchins and Harvey, 1984)***. This latter group declares itself clinically.

Deletions, where a substantial part of a chromosome is lost, are nearly always lethal ***(Hutchins and Harvey, 1984)***.

Some of the chromosomal abnormalities show an increased incidence with advanced maternal age. The most notable examples being trisomy 21 (Down's syndrome) and the 45, x-genotype (Turner's syndrome) ***(Nevin, 1982)***.

The precise type of chromosome abnormality may be critical in defining the risk of reccurence in the family. For example the risk of a further affected child with Down's syndrome is approximately 1% if the disorder is due to simple trisomy 21, but as high as 20% or more if the mother is the carrier of a chromosome translocation involving chromosome 21 *(Bobrow, 1988)*.

B) Single gene disorders:

They have typical Mendelian patterns of inheritance *(McKusick, 1978)*. The mode of inheritance depends upon whether the mutant gene is located on an autosome or sex chromosome, and whether it is dominant or recessive. Accordingly, such diseases can be grouped as autosomal dominant, or recessive, x-linked dominant or recessive.

Disorders due to dominant genes are relatively few and a great proportion of them result from a fresh mutation *(Smithells, 1978)*. For recessive disorders, detection of the carrier state is desirable by means of certain laboratory investigations. Carrier detection can be valuable in counselling those couples at risk of having affected offspring *(Nevin, 1982)*.

(2) Environmental Disorders

Few congenital defects can be confidently attributed to a single external agent. The rubella virus and the drug thalidomide are the best known examples *(Smithells, 1978)*.