MODERN TRENDS IN PRENATAL DIAGNOSIS OF FETAL ANOMALIES

ESSAY

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الوسائل الحديثه فى تشخيص العيوب الخلقية

للاجنة قبل الولاده

رسالة مقدمة من

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وعنوانها

الوسائل الحديثه فى تشخيص العيوب الخلقية للاجنة قبل الولاده

MODERN TRENDS IN PRENATAL DIAGNOSIS OF FETAL ANOMALIES

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والله ولى التوفيق

Abstract

MODERN TRENDS IN PRENATAL DIAGNOSIS OF FETAL ANOMALIES is devided in to

Non-invasive methods for diagnosis of fetal anomalies Which include
Diagnosis of fetal anomalies by 2D sonography
Diagnosis of fetal anomalies by 3D and 4D sonography
Magnetic resonance imaging

Maternal Serum Markers
Invasive Diagnosis of Fetal Anomalies
Amniocentesis Chorionic Biopsy Cordocentesis Fetal Cells in Maternal Blood Pre implantation Genetic Diagnosis

Key wards :

Prenatal Diagnosis Fetal Anomalies

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CHAPTER 4

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ABBREVIATION

ACTH	Adrenocorticotrophic Hormone
AIUM	American institute of ultrasound in medicine
AFP	α- Fetoprotein
2DUS	2-Dimensional Ultrasound
3DPD	3D power Doppler
3D US	3-Dimensional Ultrasound
CAD	Cerebellar Antero-posterior Diameter
CCAM	Cystic Adenomatoid Malformation
CDUS	Colour Doppler Ultrasound
CNS	Central nervous system
CRL	Crown-Rump Length
CTD	Cerebellar Transverse Diameter
STIC	Spatio-temporal image correlation
CVS	Chorionic Villus Sampling
DHEA-S	Dehydroepiandrosterone Sulfate
DS	Down's syndrome
ESHRE	**European Society of Human Reproduction and Embryology**
FADS	Fetal Akinesia Deformation Sequence
FASTER	**First and Second Trimester Evaluation for Aneuploidy Risk**
FISH	Fluorescent In-Situ Hybridization
FSH	Follicular Stirnulation Hormone
HASTE	Half-Fourier acquired single-shot turbo spin-echo
HCG	Human Chorionic Gonadotropin
ICSI	Intracytoplasmic Sperm Injection
IQ	Intelligence Quotient
IUGR	Intra uterine Growth Restriction

IVF	In vitro Fertilization
LH	Luteinizing Hormone
MACS	Magnetic Activated Cell Sorting
MCD	Malformation of Cortical Development
MOM	Multiple of the Median
MRI	Magnetic Resonance Imaging
NMR	Nuclear Magnetic Resonance
NT	Nuchal Translucency
NTD	Neural Tube Defect
OSCAR	One-Stop Clinics for Early Assessment of Fetal Risk
PAPP-A	Pregnancy-associated Plasma Protein A
PCR	Polymerase Chain Reaction
PDUS	Power Doppler Ultrasound
PGD	Preimplantation Genetic Diagnosis
PUBS SS-	Percutaneous Umbilical Blood Sampling
FSE	Single-shot, fast spin-echo
SURUSS	Serum Urine and Ultrasound Screening Study Thyroid-
TSH	stimulating Hormone
TTTS	Twin-Twin Transfusion Syndrom
TUI	Tomographic Ultrasound Imaging
UE3	Unconjugated Estriol
VEGF	Vascular Endothelial Growth Factors

Introduction

Embryonic Background

A basic understanding of embryology is necessary to appreciate the mechanism of producing congenital anomalies. There are three basic stages in human development.

1. The first stage begins with fertilization and lasts to third week (post-conception), until organ formation that is called embryonic stage.
2. The second stage is the organogenesis, which is the period of organ formation and lasts from the third to eight week (post-conception).
3. The third stage (the final stage) is called the fetal stage; it is primarily the stage of growth and maturation of previously formed structures. The embryo during this stage is appropriately called the fetus ***(Moore, 1982).***

Most birth defects were thought to arise during organogenesis. The usual period for the development of malformation is from the beginning of organogenesis to about the eights week ***(Briggs et al., 1990)***

Errors of Morphogenesis:

Congenital anomaly consists of a departure from the normal anatomic architecture of an organ or system. Anomalies may result from an intrinsically abnormal primordium of an organ, or from normal primordium that is affected during development by extrinsic forces ***(Spranger et al., 1982).*** Growing interest in prenatal development, coupled

with the need for a uniform nomenclature to refer to errors in morphogenesis, led an international working group to propose a set of terms useful in classification of anatomic congenital anomalies. Individual alteration of forms or structure can be classified as malformation, deformation, and disruptions ***(Spranger et al., 1982).***

Malformation:

Malformation is a morphologic defect of an organ or a larger area of the body resulting from an intrinsically abnormal development process, the term intrinsically abnormal development process refers to an abnormality in the primordium of the organ, this abnormality may not be identifiable in early stages of development. The typical example is a limb bud that appears normal in early embryonic life but later develops an extra digit. Malformations can be considered as the result of a development arrest of the primordium (incomplete morphogenesis), redundant morphogenesis or aberrant morphogenesis, although malformations often occur during the embryonic period until the 9^{th} week, some may also arise during later stages of development. A general principle is that the earlier the malformation is initiated, the more complex is the resulting anomaly ***(Moore, 1982)***

Deformation:

This refers to an abnormal form, shape or position of a part of the body, the primordium of the organ is normal but

development is affected by mechanical forces that are extrinsic or intrinsic to the fetus. For example, a club foot deformity may be the result of intrauterine constraint due to oligohydramnios (extrinsic force) or lack of movement due to neural defects associated with spina bifida (intrinsic force). Four main factors influence the pathogenesis of deformations; pressure, fetal plasticity, fetal mobility, and rate of fetal growth ***(Dunn, 1976; Hall, 1985),***

Deformations tend to occur late in gestation, as during this time there is rapid fetal growth in a potentially constraining intrauterine environment, removal of the mechanic force responsible for the deformation result in normalization or improvement of the anomaly. Spontaneous resolution after birth occurs in approximately 90% of deformations ***(Dunn, 1976).***

Table 1 - Comparison between malformation and deformation

	malformation	Deformation
Time of occurrence	Embryonic period	Fetal period
Level of disturbance	organ	Region
Incidence before 20th week	5%	0.1%
Incidence after 28th week	3.7%	2%
Perinatal mortality	41%	6%
Spontaneous correction	-	+
Correction by posture	-	+

(Cohen, 1982)

Disruption: