# MODERN TRENDS IN PRENATAL DIAGNOSIS OF FETAL ANOMALIES

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والله ولى التوفي

## **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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#### **ABBREVIATION**

ACTH Adrenocorticotrophic Hormone

AIUM American institute of ultrasound in medicine

AFP - 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

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### 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<sup>th</sup> 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:**