ROLE OF DIAGNOSTIC IMAGING IN EARLY DETECTION OF INTRAUTERINE DOWN SYNDROME

Essay

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List of Abbreviations

AC	Abdominal circumference
BPD	Biparietal diameter
CBS	Cystathionine B synthetase
CF	Cystic fibrosis
CM	Cisterna magna
CPCS	Choroid plexus cysts
CRL	Crown rumb length
DS	Down syndrome
EB	Echogenic bowel
FL	Femur length
FTS	First Trimester Screening
GA	Gestational Age
GARTF	glycinamide ribonucleotide transformylase .
HL	Humeral length
HUIFN	Human interferon
IPS	Integrated prenatal screening
kHz	Kilohertz
LFA-1	lymphocyte function associated antigen
MHz	Megahertz
MR	Magnetic resonance
MRI	Magnetic resonance imaging
MVM	Mild ventriculomegaly
NB	Nasal bone
NF	Nuchal Fold
NST	Nuchal skin fold thickness
NT	Nuchal translucency
PAIS	phosphoribosy aminoimidazole synthestase
PFKL	phosphofructokinase
PRGS	Phosphoribosy glycinamide synthetase
PUJO	Pelvi ureteric junction obstruction
RFLPs	Restriction fragment length polymorphisms
ROI	Region of interest
RPD	Renal pelvic dilatation

List of Abbreviations (Cont.)

SD	Standard deviation
SDS	Standard deviations
SOD	Superpxide Dismutase
SSFSE	Single shot fast spin echo
T21	Trisomy 21
TCD	Trans cerebellar diameter
VSDS	Ventricular septal defects

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Introduction

Development of human embryo is a complex and evolutive process that requires accurate evaluation to detect any fetal malformation that may result in lethal or morbid outcome of pregnancy. Early and accurate antenatal diagnosis of fetal malformations and differentiation between a lethal and a non-lethal variety has important implications for the management of a pregnancy and prediction of fetal outcome ***(Lee et al., 2002).***

Down syndrome (Trisomy 21) is the most common autosomal Trisomy encountered in pregnancies that extend beyond the first trimester. Down syndrome is named after JOHN LANGDON DOWN, the first scientist to describe the pattern of congenital anomalies characterizing individuals with trisomy 21 ***(Baladini and Volpe, 2007)***.

Ultrasound screening in pregnancy can be seen as the offer to check the largest number of pregnancies, by the largest number of operators for simple and reproducible criteria in order to make important choices on the management of pregnancy and delivery. Most established screening programs claim high sensitivity (for low false-positive rate) for different conditions such as Down syndrome ***(Baladini and Volpe 2007).***

The use of MRI was first described in pregnancy in 1983. Its initial application was for maternal and placental abnormalities. Fetal MRI was initially limited because of fetal motion artifact. In the 1990s, fetal MRI became practical because of the development of single-shot rapid acquisition sequence with refocused echoes. This high-quality T2-weighted sequence has a slice acquisition time of <1 second and essentially "freezes" fetal motion As a result, excellent fetal imaging can be obtained without maternal or fetal sedation ***(Opin 2001)***.

The risks and benefits of all imaging studies that are carried out during pregnancy need to be discussed with the patient. In the case of magnetic resonance imaging (MRI) there are theoretical risks of teratogenicity, but no proven effects in humans. This study reviews safety concerns pertaining to MR examinations during pregnancy and illustrates embryonic and fetal anatomy in the first and second trimester ***(Levine 2005)***.

Aim of The Work

To emphasize the role of various diagnostic imaging modalities (U.S and MRI) in early detection of Down Syndrome during first and second trimesters of pregnancy to help in the decision of therapeutic abortion.

(I) Normal Anatomy of the Fetus by Ultrasound Examination

An ultrasound examination is in the unique position of being both a screening test and a diagnostic test for fetal anomalies ***(Sadler, 2004)***.

The majority of second trimester scans are performed at 20-22 weeks, although a more limited assessment of fetal anatomy can be made, using the transabdominal technique, from 15 weeks ***(Sadler, 2004).***

I. Central nervous system :

A-The lateral ventricles :

The lateral ventricles are the easiest parts of the ventricular system to visualize with ultrasound. These are bilateral and have anterior, posterior and inferior horns. They contain the choroid plexus, which is normally apparent with ultrasound only in the posterior horn. The inferior horn is rarely seen in normal fetuses because it is lost in the echoes from the base of the skull **(Fig. 1)** ***(Filly, 1991).***

The anterior horn :

The anterior horns of the lateral ventricles are visualized in the same section as that required for the BPD. The medial and lateral borders of the anterior horn lie in very close proximity at this level ***(Filly, 1991).***

The posterior horn or atrium :

The posterior horn, or atrium, contains choroid that is readily identified as a rounded, hyperechoic structure. The ultrasound appearance of the posterior horn differs from the anterior horn in that it has obvious depth may give valuable information **(Fig. 1)** ***(Hehr and Muenke, 1999)***.