حسام مغربى

شبكة المعلومات الجامعية

بسم اللّه الرحمن الرحيم

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التوثيق الالكتروني والميكروفيلم

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جامعة عين شمس

التوثيق الإلكتروني والميكروفيلم

قسم

نقسم باللّه العظيم أن المادة التي تم توثيقها وتسجيلها علي هذه الأقراص المدمجة قد أعدت دون أية تغيرات

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بعض الوثائق الأصلية تالفة

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شبكة المعلومات الجامعية

بالرسالة صفحات لم ترد بالأصل

RELATION BETWEEN MOLECULAR DIAGNOSIS OF CYTOGENETIC ANOMALIES AND SOME PRENATAL INFECTIOUS DISEASES

Thesis

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By

AMAL ABDEL RASHEED ALY WAFY

(M.B.,B.Ch., & M.Sc.)

SUPERVISORS

Prof. Dr.
MENA SAMY MESIHA
Professor of Microbiology
Faculty of Medicine
Tanta University

Prof. Dr.
AZIZA MAHMOUD EL TAYFA
Professor of Microbiology
Faculty of Medicine
Tanta University

Dr.
AFAF SAYED ZAMZAM
Assistant Prof. of Microbiology
Faculty of Medicine
Tanta University

Prof. Dr.
EZAT SAYED ELSOBKY
Professor of Genetics- Paediatrics Dept.
Faculty of Medicine
Ain Shams University

FACULTY OF MEDICINE
TANTA UNIVERSITY
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بسم الله الرحمن الرحيم

(سنريهم آياتنا في الآفاق وفي أنفسهم حتى يتبين لهم
أنه الحق ، أولم يكف بربك أنه
على كل شيء شهيد)

(سورة فصلت ، الآية ٥٣)

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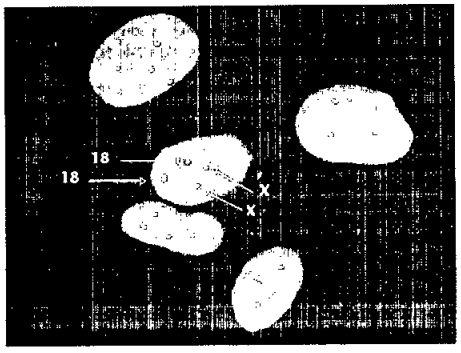
ABBREVIATION

AFP	: Alpha feto protein
AIDS	: Acquired immunodeficiency disease.
ALL	: Acute lymphocytic leukemia
AMA	: Advanced maternal age
AML	: Acute myelogenous leukemia
APL	: Acute promylocytic leukemia
ASD	: Atrial septal defect
Bp	: Base pair
CCD	: Couple change devise
CML	: Chronic myeloid leukemia
CMV	; Cytomegalovirus
CVS	: Chronic villus samplaing
EA	: Early aminiocentesis
ELISA	: Enzyme linked immunofluorscent assay
FISH	: Fluoroscence in situ Hybridization
FITC	: Fluorescin isothiocyanate
G	: long arm (grand)
HCG	: Human chrionic gonadotrophin
HIV	: Human immunodeficiency virus.
HLA	: Human leucocyte Antigen
HSV	: Herpes simplex virus
HTIV	: Human T-cell leukaemia virus 1 and 2
IgG	: Immunoglobulin G
ISH	: Insitu hybridization
IVF	: Invitrofertilization
LINES	: Long interspersed Nuclear elements
MMR	: Measles, Mumps and rubella.
MOM	: Multiple of the median
NT	: Nuchal thickness (translucency)
P	: Short arm (petit)
PCR	: Polymerase chain reaction

PDA	: Patent Ductus Arteriosus
PGD	: Preimplantation genetic diagnosis
PML	: Promyelocytic leukemia
PUBS	: Percutaneous umbilical blood sampling
RAC	: Retinoic acid receptor alpha
SINES	: Short interspersed nuclear elements
SRY	: Sex determining region on Y-chromosome
TDF	: Testis determining factor
TMPD	: Transient myeloproliferative Disorder
TMS	: triple marker screening
TORCH	: Toxplasma gondii, rubella herpes simplex
UE3	: Unconjugated estrial
UK	: United Kingdom
US	: Ultrasound
USA	: United states of American
VSD	: Ventricular septal defect
WCP	: Whole chromosome paint probe
YACS	: Yeast artifical chromosome

INTRODUCTION



INTRODUCTION

Pregnant women are exposed to many biological, e.g. microbial agents, which are potentially harmful to the fetus. TORCH infections acquired during pregnancy may result in severe fetal anomalies or even fetal loss (Ekblad, 1995).

The original TORCH complex described clinically include infections caused by toxoplama gonadii, rubella virus, cytomegalovirus and herpes simplex virus types 1 and 2. Diagnosis is confirmed by culture and identification of species-specific immunoglobulins (Epps and Pittelkow, 1995).

Kaur et al., (1999) demonstrated high frequency of seropositives for one or more of these infections during pregnancy, thus justifying routine prenatal TORCH screening.

Different variables influence the possibility that maternal viral infection may be transmitted to the fetus, although not all fetal infections result in fetal illness with consequent fetopathy. As concerns the fetus, prenatal diagnosis includes invasive techniques necessary for fetal tissue sampling. These techniques carry some risks. The fetal infectious risk, as determined by maternal clinico-serological profile and according to sonographic investigation, always should be weighed against the risks and benefits of invasive diagnostic procedures (Noia et al., 1998).

With the recent advances in molecular biology, many sophisticated technologies have been developed. The technique of fluorescence in situ hybridization (FISH) is one of them, and is being widely used not only in

research fields of molecular biology but also for diagnosis in clinical laboratories (Sasaki, 1996).

In situ hybridization (ISH), since its introduction in 1969 by Gall and Pardue has found multiple uses in molecular morphology due its unique capability of visualizing nucleic acids sequences without altering the cell's cytological, chromosomal or histological integrity. Fluorescence in situ hybridization (FISH) has established itself as a variation of the traditional hybridization process whereby the probes utilized are a fluorescently labeled and produce bright clear signals upon detection. FISH is involved in localizing and exploring chromosomal, genetic and genomic aberrations that are often directly correlated to disease causation and progression. This technique is used as an essential tool in prognostics, diagnostics and disease monitoring in medicine (Luke and Shepelsky, 1998).

Small supernumerary marker chromosomes are seldom found in prenatal diagnosis and the majority of them are difficult to identify. The only possibility to give a more precise prognosis is by establishing its origin. FISH is the best technique to identify the chromosomal origin (Sanz et al., 2000).

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