Detection of Prenatal Abnormal Chromosomal Syndrome among Pregnant Women with High Risk of Birth Defect during the First Trimester

Submitted By

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For the Fulfilment of the requirement of

Doctor of Philosophy (Ph-D) Degree in Sciences (Biochemistry)

Faculty of Science

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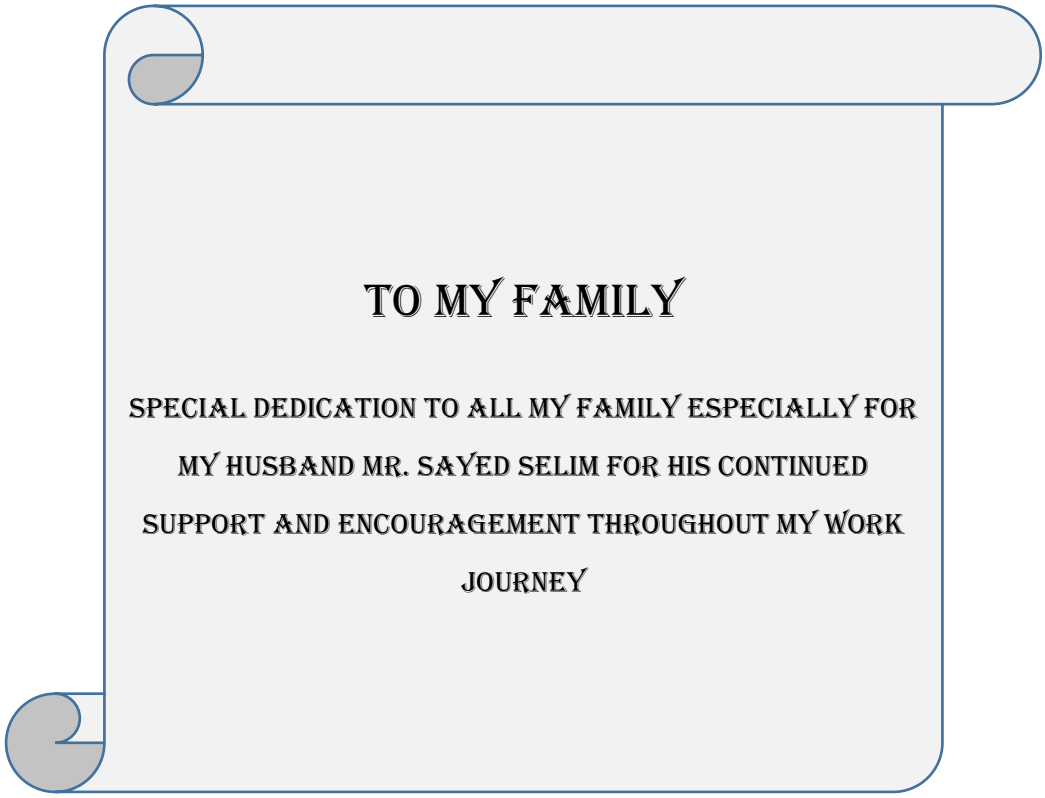
DEDICATION


TO MY FAMILY
SPECIAL DEDICATION TO ALL MY FAMILY ESPECIALLY FOR MY HUSBAND MR. SAYED SELIM FOR HIS CONTINUED SUPPORT AND ENCOURAGEMENT THROUGHOUT MY WORK JOURNEY

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ABBREVIATIONS AND SYMBOLS

aCGH	**Array Comparative Genomic Hybridization**
ACOG	American College Of Obstetrics And Gynaecology
ADAM12	Disintegrin And Metalloproteinase Domain-Containing Protein 12
AF	Amniotic Fluid
AMR	Analytical Measurement Range
AUTO	Autosomal prope
AS	Angelman Syndrome
BACs	Bacterial Artificial Chromosomes
BMI	Body Mass Index
BOBs	Bacterial Artificial Chromosomes On Beads
CFTS	Combined First Trimester Screening
CDC	Cri due Chat syndrome
Chr	Chromosome
CI	Confidence Intervals
CNVs	Copy Number Variants
CRL	Crown-Rump Length
CV	Coefficient Of Variation
CVs	Chorionic Villis
DGS	Digeorge Syndrome
DM I	Diabetes Mellitus, Type I
DM II	Diabetes Mellitus Type II
DS	Down Syndrome
eFTS	Enhanced First Trimester Screening
ES	Edwards Syndrome
EVTs	Extravillous Trophoblasts
FGR	Fetal Growth Restriction
FISH	Fluorescence In Situ Hybridization

FMF	Fetal Medicine Foundation
FP	False Positive
FPR	False Positive Rate
FTS	First Trimester Screening
GA	Gestational Age
G-banding	Giemsa Banding
GCM1	Glial Cell Missing 1
GHBP	Growth Hormone Binding Protein
GMU	Gulf Medical University
LGS	Langer-Giedion Syndrom
HCG	Human Chorionic Gonadotropin
IGF	Insulin-Like Growth Factors
IGFBPs	Insulin-Like Growth Factor Binding Proteins
IQ	Intelligence Quotient
ITA	Invasive Trophoblast Antigen
IVF	*In Vitro Fertilisation*
Kb	Kilobyte
LMP	Last Menstrual Period
Mb	Megabyte
MDS	Miller-Dieker Syndrom
MoMs	Multiples Of Median
mRNAs	Messenger Rnas
MSAFP	Maternal Serum Alpha Fetoprotein
NB	Nasal Bone
Ng	Nano Gram
NT	Nuchal Translucency
NTD	Neural Tube Defect
PAPP-A	Pregnancy Associated Plasma Protein A
Pb	Petabyte
PE	Preeclampsia
PGDM	Pre Gestational Diabetes Mellitus

PGH	Placental Growth Hormone
PGS	Pre-Implantation Genetic Screening
PLGF	Placental Growth Factor
PMP22	Peripheral Myelin Protein 22
POC	Product Of Consumption
PP13	Placental Protein 13
ProMBP	Preform Of Eosinophil Major Basic Protein
PS	Patau Syndrome
PWS	Prader-Willi Syndrome
QF-PCR	Quantitative Fluorescent Polymerase Chain Reaction
SGA	Small Gestational Age
SMS	Smith-Magenis syndrome
SNP	Single Nucleotide Polymorphism
T13	Trisomy 13
T18	Trisomy 18
T21	Trisomy 21
TRACE	Time Resolved Amplified Cryptate Emission
uE3	Unconjugated Oestriol
US	Ultrasound
VCFS	Velocardiofacial Syndrome
VEGF	Vascular Endothelial Growth Factor
WBS	Williams–Beuren Syndrome
WHS	Wolf-Hirschhorn Syndrome
WS	Williams Syndrome
β –hCG	Beta Human Chorionic Gonadotropin

ABSTRACT