



بسم الله الرحمن الرحيم

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مسئولية عن محتوى هذه الرسالة.

ملاحظات:

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**GENETICS, MATERNAL LIFESTYLE AND  
ENVIRONMENTAL ROIE IN PEDIATRIC  
INTELLECTUAL DISABILITY**

**Submitted By**

**Rana Mahrous Ali Mahmoud**

M.B.B.Ch., Faculty of Medicine, Cairo University, 2009  
Master in, Medical & Clinical Genetics, Faculty of Medicine,  
Ain Shams University, 2016

A Thesis Submitted in Partial Fulfillment  
Of  
The Requirement for the Doctor of Philosophy Degree  
In  
Environmental Sciences

Department of Environmental Medical Sciences  
Faculty of Graduate Studies and Environmental Research  
Ain Shams University

**2022**

APPROVAL SHEET  
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## Abstract

GDD/ID is characterized by significant limitations in intellectual functioning and adaptive behavior, and is frequently associated with developmental delay and various congenital abnormalities. A wide range of environmental factors, in addition to genetic determinants, can affect brain development. Because of the significant clinical and genetic heterogeneity, studying intellectual disability is challenging.

The aim of our study was to determine the possible role of maternal lifestyle and exposure to environmental risk factors in the development of intellectual disability and to detect chromosomal aberrations and/or copy number variants using various cytogenomics techniques in pediatric patients with intellectual disability.

Out of 40 selected patients with GDD/ ID with or without multiple congenital anomalies (MCA). Different cytogenomics techniques conducted on our patients revealed normal karyotype in 39 patient and 1 female patient had abnormal. MLPA subtelomeric screening, and microdeletion/ microduplication screening was done for all patients. Subtelomeric abnormality was detected solely in the patient with abnormal karyotype. Screening for microdeletion/ microduplication for all patients revealed 4

# Contents

<b>Subjects</b>	<b>Page</b>
• List of Abbreviations.....	II
• List of Tables .....	V
• List of Figures .....	VIII
• Abstract .....	1
• Introduction .....	3
• Aim of the Work.....	7
• Review of literature	
- <b>Chapter (1):</b> Intellectual disability “Environmental Factors” .....	8
- <b>Chapter (2):</b> Genetic Causes of Intellectual disability .....	32
- <b>Chapter (3):</b> Cytogenomic Techniques.....	46
• Patients and Methods.....	62
• Results.....	88
• Discussion .....	137
• Conclusion .....	160
• Recommendations.....	161
• Summary.....	162
• References .....	171
• Arabic Summary	
• Appendix	

## List of Abbreviations

<b>AAMR's</b>	American Association on Mental Retardation's
<b>AAIDD</b>	American Association on Intellectual and Developmental Disabilities
<b>ADHD</b>	Attention deficit disorder
<b>ADID</b>	Autosomal dominant intellectual disability
<b>ARID</b>	Autosomal recessive intellectual disability
<b>CD</b>	Cognitive disorder
<b>CNVs</b>	Copy number variants
<b>CGH</b>	Comparative genomic hybridization
<b>CMA</b>	Chromosomal microarray
<b>CMV</b>	Cytomegalovirus
<b>DNA</b>	Deoxyribonucleic acid
<b>DS</b>	Down syndrome
<b>DSM-IV</b>	Diagnostic and Statistical Manual of Mental Disorders, 4th edition.

<b>DSM-5</b>	Diagnostic and Statistical Manual of Mental Disorders, 5th edition.
<b>FA</b>	Folic acid
<b>FAS</b>	Fetal alcohol syndrome
<b>FASDs</b>	Fetal alcohol spectrum disorders
<b>FISH</b>	Fluorescence in situ hybridization
<b>GDD</b>	Global developmental delay
<b>ID</b>	Intellectual disability
<b>IUGR</b>	Intrauterine growth retardation
<b>IQ</b>	Intelligence quotient
<b>ISCN</b>	International System for Human Cytogenetic Nomenclature
<b>LSI</b>	Locus Specific probe
<b>MAPH</b>	Multiplex amplifiable probe hybridisation
<b>Mb</b>	Million base pairs
<b>MCA</b>	Multiple Congenital Anomalies

<b>MLPA</b>	Multiplex ligation dependent probe amplification
<b>MR</b>	Mental retardation
<b>NGS</b>	Next generation sequencing
<b>NS-ID</b>	Non syndromic intellectual disability
<b>NTDs</b>	Neural tube defects
<b>PCR</b>	Polymerase chain reaction
<b>S-ID</b>	Syndromic intellectual disability
<b>SNP</b>	Single nucleotide polymorphism
<b>TORCH</b>	Toxoplasmosis, Others (syphilis, varicella-zoster, parvovirus B19), Rubella, Cytomegalovirus (CMV), and Herpes infections.
<b>WES</b>	Whole exome sequencing
<b>X-CNVs</b>	X chromosome CNVs
<b>XLID</b>	X-linked intellectual disability

## List of Tables

Table No	Title	Page
<b>Table (1)</b>	cognitive disability classification	12
<b>Table (2)</b>	Relation between fetal pathogens and associated morbidity.	23
<b>Table (3)</b>	Recurrent copy number variants in the human genome and microdeletion syndromes that cause, or predispose to, intellectual disability or related disorders.	36
<b>Table (4)</b>	Common recognizable XLID Syndrome	42
<b>Table (5)</b>	Empirical recurrence risks in intellectual disability without a defined cause.	45
<b>Table (6)</b>	Examples of copy number variations (CNVs) and conveyed genomic disorders.	47
<b>Table (7)</b>	Methods and contexts for copy number detection	49
<b>Table (8)</b>	Comparison between multiplex ligation-dependent probe amplification (MLPA) assay and other methods for the detection of gene deletions/duplications	61
<b>Table (9)</b>	SALSA MLPA probemix P245 Microdeletion Syndromes-1A	76

<b>Table No</b>	<b>Title</b>	<b>Page</b>
<b>Table (10)</b>	Demographic findings in the patients' history.	88
<b>Table (11)</b>	IQ and severity of intellectual disability in the patients group	90
<b>Table (12)</b>	Number and percentage of relevant findings in the history and physical examination of patients.	91
<b>Table (13)</b>	Parental lifestyle and history of medical importance.	92
<b>Table (14)</b>	Pregnancy and delivery circumstances	94
<b>Table (15)</b>	Demographic characteristics and parental occupational exposure distributed across patient group and control group.	96
<b>Table (16)</b>	Parental chronic diseases and habits distributed across patient group and control group.	102
<b>Table (17)</b>	Relevant findings in the history distributed across both patients and control groups.	106

<b>Table No</b>	<b>Title</b>	<b>Page</b>
<b>Table (18)</b>	Mothers characteristics and habits at pregnancy time distributed across patients group and control group	109
<b>Table (19)</b>	One proportion z test comparing a reference value coming from the literature with the total number of participants in this study.	114
<b>Table (20)</b>	Karyotype and MLPA results in patients	120

## List of Figures

Figure No	Title	Page
<b>Figure (1)</b>	Intellectual disability classification. Multiple factors are involved in intellectual disability including genetic inheritance and environmental conditions.	17
<b>Figure (2)</b>	Bacteria, viruses and parasites known to influence fetal development	22
<b>Figure (3)</b>	Schematic diagram of the MLPA process. Step 1. Hybridization of the probes to the target DNA sequence. Step 2. Ligation of adjacent bound probes. Step 3. PCR amplification of the ligated probe	59
<b>Figure (4)</b>	PAXgene blood DNA tubes and PAXgene blood DNA kit ( <i>PreAnalytix, Hiden, Germany</i> ).	71
<b>Figure (5)</b>	The PAXgene blood DNA procedure ( <i>PreAnalytix, Hiden, Germany</i> )	72
<b>Figure (6)</b>	Thermocycler program for the MLPA reaction.	79
<b>Figure (7)</b>	Pie chart showing the severity of intellectual disability in the cases group.	90
<b>Figure (8)</b>	Distribution of both gender and residency across patient group and control group	101