ECOLOGICAL AND GENETIC FACTORS INFLUENCING AUTISTIC CHILDREN

Submitted By

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B.Sc. of Veterinary Science, Faculty of Veterinary Medicine,
Cairo University, 1985
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Ain Shams University, 1997

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

Department of Environmental Medical Sciences
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APPROVAL SHEET

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May God bless all of them and this work.

Aim of the Work

Aim of the study

The aim of this work is to find out the influences of some environmental factors and genetic factors on the development of autism on some Egyptian children.

Abstract

Abstract

1- Background:

Autism is a complex neurodevelopmental disorder defined by impaired social interaction, communication, deficits, restricted interests and repetitive behavioral patterns.

There are probably many combinations of genes and environmental factors that contribute to the constellation of autistic traits.

2- Patients and Methods:

This study was carried out on patients following up at the Medical genetic center, Ain Shams University Hospitals.

- The study was conducted on about (52) Autistic patients, age ranging from (3-12) years both males and females were included.
- Group of relatives consists of available (mother, grandparents) whom are apparently healthy individuals, but most of them showed a history of medical, neurological and psychiatric disorders as noticed throughout the pedigree construction test.
- Control group about (42) patients, was selected from apparently healthy individuals with no history suggesting medical, neurological or psychiatric disorders with ages ranging between (3-12) years.
- The cases and controls were subjected to history taking sheet that include age, family history of other autistic patients, birth difficulties, birth place and medications received.
- They were also exposed to pedigree construction and findings showed cases with autistic behaviour and psychic disorders.
- Diagnosed cases with Autism were subjected to (CARS) (Child Autism Rating Scale) test which showed significant cases.
- Also the experimental group under study of (autistic children) were subjected to the following investigations:

1- DNA analysis:

In which (DNA) samples were taken from peripheral lymphocytes to undergo (DNA) extraction and the:

* ***Glutathione-s-transferase test:***

  Which was genotyped by polymerase chain reaction (PCR) in which its absence was found in some studied cases.

2- Heavy Metals test:

Including (lead and Mercury) it was detected by the atomic absorption apparatus in blood of studied cases of children which showed non significancy in both detected metals (lead & mercury).

3- Pseudocholine esterase test:

Was measured in blood of autistic cases revealing non significancy.

* Results and Conclusion:

Increased exposure to some parameters measured above showed no evidences of significancy.

List of Abbreviations

AAP	:	American Academy of Pediatrics.
AD	:	Autistic Disorder.
ADHD	:	Attention Deficit Hyperactivity Disorder.
AL	:	Aluminium
AS	:	Angelman Syndrome.
ASD	:	Autism Spectrum Disorder.
ATA	:	Atmospheric Altitude above Sea Level.
BCHE	:	Plasmacholesterase
Ca	:	Calcium
CARS	:	Childhood Autism Rating Scale.
CAST	:	Childhood Asperger Syndrome Test.
CDC	:	Center of Disease Control.
CGH	:	Comparative Genetic Hybridization.
CNS	:	Central Nervous System.
DHA	:	Docosahexaenoic Acid.
DNA	:	Deoxy-ribo Nucleic Acid.
DSM III R	:	3rd analytical diagnosis guide
DSM IV	:	4th analytical diagnosis guide
EPA	:	Eico sapentanoic Acid.
EPA	:	Environmental Protection Agency.
FDA	:	Food and Drug Administration (USA).
FISH analysis:		Fluorescence Insitue Hybridization.
GIT	:	Gastro Intestinal Tract.
GLA	:	Gamma Linolenic Acid.
GSH	:	Glutathione - S - transferase Hormone.
GSTP1	:	Glutathione - S - Transferase p1.
Hg	:	Mercury
I/U	:	International unit
ICD	:	International Classification for Disabilities.
ICD	:	International Classification for Disabilities.

IG : Immuno Globulin.
IgG : Immuno Globulin G.
IQ : Intelligence Quotient.
JNK : Jun N - terminal Kinase.
LNA : Alpha Linolenic Acid.
Mg : Magnesium
MMR : Measles, Mump, Rubella.
Mn : Manganese
$MRCP_2$ mutations: Methyl – DNA binding protein.
MRI : Magnetic Resonance.
Nacl : Sodium chloride
NASA : National Auronautic Space Administration
NMDA : N - Methy - D - Aspartate.
OCD : Obsessive Compulsive Disorder.
OXTR : Oxytocin Receptor.
Pb : Lead
PBMCs : Peripheral Blood Mononuclear Cells.
PCHE : Pseudocholinesterase
PDD : Pervasive Developmental Disorder.
PET : Positron Emission Tomography.
PUFAs : Polysaturated Fatty Acids.
RELP : Restriction Fragment Length Polymorphism.
SCQ : Social Communication Questionnaire.
SE : Selenium
SIB : Self Infurious Behaviour.
SLE : Systemic Lupus Erythrosis.
SWAT : Strength Weakness opportunity Threat.
TSC : Tuberous Sclerosis Complex.
U.S. : United States.
U/V : Ultraviolet
WHO : World Human Organization.
Zn : Zinc

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Introduction