

# **GENETIC STUDIES OF EPILEPSY IN SOME EGYPTIAN CHILDREN AND THEIR FAMILIES**

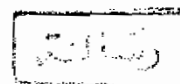
**THESIS**

Submitted in partial fulfillment of Master Degree  
in **Paediatrics**

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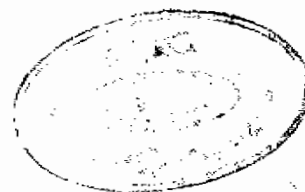
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## Acknowledgement

*First and foremost, thanks to God*

*I would like to express my deepest appreciation and gratitude to Professor Dr. Hamed Shatla, professor of Paediatrics, Ain Shams University, for his continuous support and encouragement throughout the work.*

*I would like also to express my deep gratitude and thanks to Dr. Moustafa Abdel Aziz El-Sayed El-Hodhod, Lecturer of Paediatrics, Ain Shams University for his sincere supervision, encouragement and energetic help in the details of this work.*

*I am very grateful to Dr. Ibtesam Mohamed Ramzy Hussein, Lecturer of Human Genetics, Human Genetics Department, NRC, for her great effort and continuous guidance and encouragement through out the preparation of this work.*

*I would like also to express my thanks and gratitude to Dr. Hanan Hosny Afifi, Lecturer of human genetics, Human genetics Department, NRC, for her valuable advice, kind supervision and continuous encouragement.*

*At last but not the least, I would like to acknowledge my appreciation to my parents and my husband for their continuous support and understanding.*



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## LIST OF ABBREVIATIONS

<b>BFNC</b>	: Benign familial neonatal convulsions
<b>Ca<sup>2+</sup></b>	: Calcium ions
<b>cDNA</b>	: Complementary DNA
<b>CMV</b>	: Cytomegalovirus
<b>CNS</b>	: Central nervous system
<b>COFS</b>	: Cerebro-oculo-facio-skeletal syndrome
<b>CP</b>	: Cerebral palsy
<b>CSF</b>	: Cerebro spinal fluid
<b>CT</b>	: Computed tomographic scan.
<b>di</b>	: dicentric chromosome
<b>DNA</b>	: Deoxyribonucleic acid
<b>DNPH</b>	: Dinitrophenyl hydrazine
<b>DZ</b>	: Dizygous twins
<b>EEG</b>	: Electroencephalography
<b>GABA</b>	: Gamma aminobutyric acid
<b>GTC</b>	: Generalized tonic-clonic seizures
<b>HIM</b>	: Human immunodeficiency virus
<b>HLA</b>	: Human leucocytic antigens
<b>Hz</b>	: Hertz
<b>IM</b>	: Intramuscular
<b>IQ</b>	: Intelligence quotient
<b>IV</b>	: Intravenous
<b>JME</b>	: Juvenile myoclonic epilepsy
<b>K<sup>+</sup></b>	: Potassium ions
<b>M.S</b>	: Metabolic screening
<b>ML</b>	: Mucopolidosis
<b>MR</b>	: Mental retardation
<b>MRI</b>	: Magnetic resonance imaging
<b>MSUD</b>	: Maple syrup urine disease
<b>MZ</b>	: Monozygous twins
<b>NCPP</b>	: National Collaborative Perinatal project.
<b>NF</b>	: Neurofibromatosis
<b>PCR</b>	: Polymerase chain reaction
<b>PKU</b>	: Phenylketonuria
<b>PVH</b>	: Periventricular haemorrhage
<b>RFLP</b>	: Restriction fragment length polymorphism
<b>RNA</b>	: Ribonucleic acid
<b>SQ</b>	: Social quotient
<b>T<sub>3</sub></b>	: Triiodothyronine
<b>T<sub>4</sub></b>	: Thyroxine
<b>TORCH</b>	: Toxoplasmosis, Rubella, cytomegalovirus, Herpes simplex virus
<b>TS</b>	: Tuberous sclerosis



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# INTRODUCTION

## INTRODUCTION

Epilepsy is the tendency to develop recurrent seizures. It has a worldwide prevalence with no geographical, cultural, or sex predelection.

Incidence of epilepsy was estimated in U.S.A. as 0.331/1,000/year with a prevalence of 3.7/1,000/year (*Kotagal 1990*).

The incidence of epilepsy is highest in the first decade, with a small secondary peak after age 60 (*Crombie et al, 1960; Pond et al, 1960*).

Seizures are by far the most common manifestation of neurological disease in the newborn (*Kotagal, 1990*).

Seizures are classified clinically into: (Dreifuss, 1983):

- 1- Partial seizures.
- 2- Generalized seizures.
- 3- Unclassified (neonatal seizures).
- 4- Status epilepticus.

The most common causes of neonatal and childhood seizures are: (Wallace 1992):

**\* Metabolic Derangement such as:** Hypoxic-ischemic encephalopathy, hypocalcaemia, hypomagnesaemia, hypoglycaemia, hyponatraemia, and inborn errors of metabolism.

- \* **Intracranial haemorrhage.**
- \* **Infections:** Such as congenital intrauterine infections (e.g. toxoplasmosis and herpes simplex) and bacterial meningitis.
- \* **Drug Induced:** Withdrawal or toxic.
- \* **Vascular causes:** Such as malformations of intracerebral blood vessels, vasculopathies and embolic or thrombotic phenomena.
- \* **The genetic role in epilepsy was suspected in different studies.**

Seizures were described in different syndromes caused by chromosomal aberration such as in trisomy 18 (*Kotagal 1990*), trisomy 12 p syndrome (*Guerrini et al, 1990*), deletion (2)(q31 q33) (*Ramer et al, 1990*), Angelman syndrome with deletion of 15q (*Fryburg et al, 1991*), ring chromosome 20 (*Halal et al, 1992*), ring chromosome 6 (*Paz-y-Mino et al, 1990*) and ring chromosome 17 in a case of Miller-Dieker syndrome (*Sharief et al, 1991*).

Some cases with epilepsy were reported to be inherited as autosomal dominant conditions as in benign familial neonatal convulsions and Neurofibromatosis I (*Wallace 1992*).

Epilepsy was also described in autosomal recessive disorders such as some inborn errors of metabolism as in phenylketonuria, maple syrup urine disease, non-ketotic

hyperglycinaemia, urea cycle disorders and galactosaemia (*Kotagal 1990*).

The genetic mechanism has also been identified in benign partial epilepsies (Doose & Baier 1991; Pedley 1991; Sillanpaa et al, 1991).

Little studies have been done for evaluation of genetic causes of epilepsy in Egypt.

# AIM OF THE WORK

## **AIM OF THE STUDY**

1. It is an updating review on epilepsy emphasizing a diagnostic, prognostic and therapeutic overview as well as knowledge about the genetic basis of epilepsy.
2. To study the genetic role in cases of neonatal and childhood epilepsy among some Egyptian families.