شبكة المعلومات الجامعية

# بسم اللّه الرحمن الرحيم



# شبكة المعلومات الجامعية

# التوثيق الالكتروني والميكروفيلم

شبكة المعلومات الجامعية

# 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

# يجب أن

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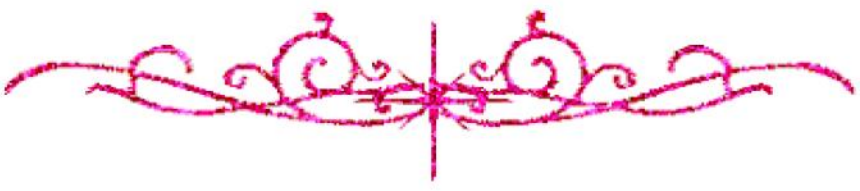

# بالرسالة صفحات
# لم ترد بالأصل

# Clinical and Genetic Study of Children with White Matter Diseases

Thesis

***Submitted for fulfillment of PhD in Childhood Studies***
***(Child Health and Nutrition)***
***Medical Studies Department for Children***

By

**Karima Raafat Mahmoud Youssef**

Assistant Researcher at Clinical Genetics Department, Human Genetics
& Genome Research Division, National Research Centre (NRC)
M.B. B.Ch, MSc. Pediatrics

Under supervision of

**Dr. Omar El-Sayed Elshourbagy**

Professor of Public Health and Family Medicine
Department of Medical Studies for Children
Faculty of Postgraduate Childhood Studies

**Dr. Randa Kamal Abdel Raouf**

Professor of Pediatrics
Department of Medical Studies for Children
Faculty of Postgraduate Childhood Studies

**Dr. Maha Saad Zaki**

Professor of Clinical Genetics
Clinical Genetics Department
National Research Centre

**Dr. Ekram Maher Fateen**

Prof. of Biochemical Genetics
Biochemical Genetics Department
National Research Centre

**Dr. Mohamed Abdel Hamid Elsayed**

Researcher of Molecular Genetics
Medical Molecular Genetics Department
National Research Centre

***Faculty of Postgraduate Childhood Studies***
***Ain Shams University***
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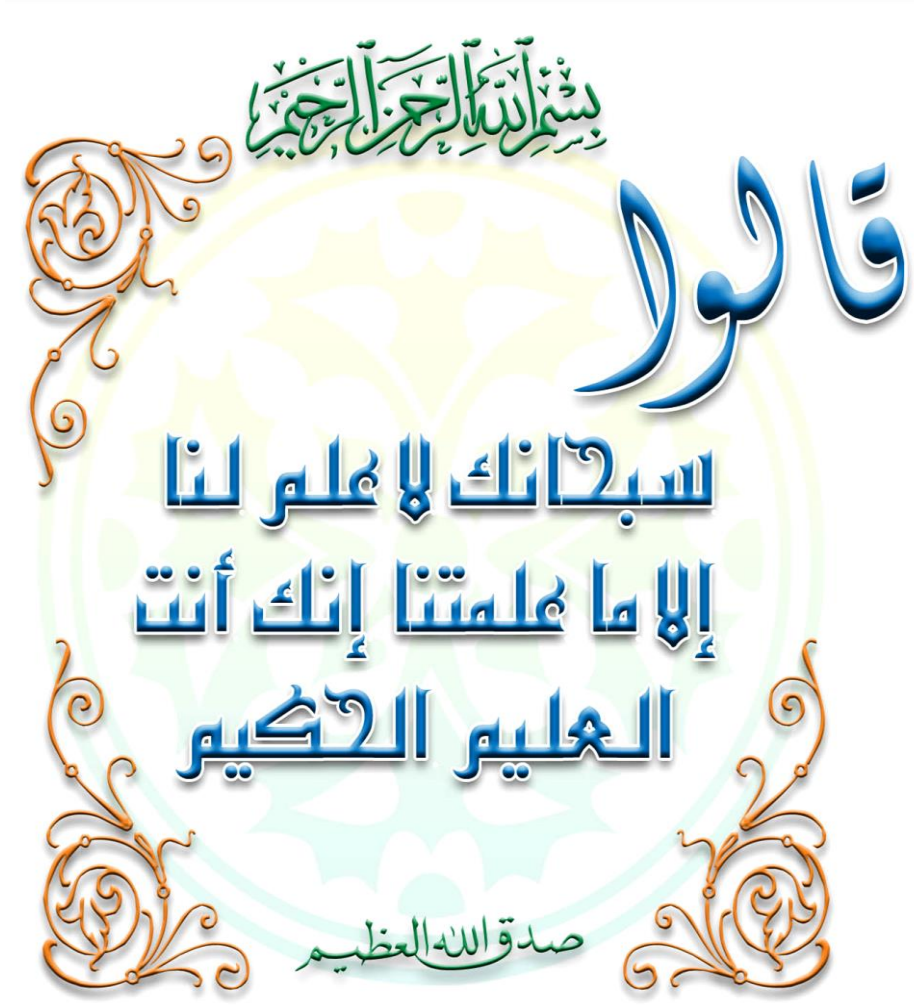


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# List of Abbreviations

| Abbr. | Full term |
|---|---|
| **ADD** | Attention deficit disorder |
| **ADEM** | Acute disseminated encephalomyelitis |
| **AGS** | Aicardi – Goutieres syndrome |
| **AIMP1** | Aminoacyl-tRNA synthetase complex- interacting multifactorial protein 1 |
| **ALSP** | Axonal spheroids and pigmented glia |
| **AMN** | Adrenomyeloneuropathy |
| **AR** | Autosomal recessive |
| **ASA** | Arylsulfatase A |
| **BH4** | Tetrahydrobiopterin |
| **CD** | Canavan disease |
| **CIS** | Clinically isolated syndrome |
| **CMD** | Congenital muscle dystrophies |
| **CNS** | Central nervous system |
| **CSF** | Cerebrospinal fluid |
| **CT** | Computed tomography |
| **DHPR** | Dihydropteridine reductase |
| **ECM** | Extracellular matrix |
| **EEG** | Electroencephalogram |
| **eIF2B** | Eukaryotic translation initiation factor 2B |
| **EMG** | Electromyography |
| **FGE** | Formylglycine-generating enzyme |
| **FLAIR** | Fluid-attenuated inversion recovery |

| | |
|---|---|
| **GA type I** | Glutaric aciduria type I |
| **GALC gene** | Galactosylceramidase |
| **GALNS** | Galactosamine (N-acetyl)-6-sulfate sulfatase |
| **GFAP gene** | Glial fibrillary acidic protein gene |
| **GLIA** | Global Leukodystrophy Initiative |
| **GMI & II** | Gangliosidosis type I & II |
| **GTPCH** | Guanosine triphosphate cyclohydrolase |
| **HCC**: | Hypomyelination with congenital cataract |
| **HDLS** | Hereditary diffuse leukoencephalopathy with axonal spheroids |
| **HSCT** | Hematopoietic stem cell transplantation |
| **IDS** | Iduronate 2-sulfatase |
| **LAMA2**: | Laminin subunit alpha -2 |
| **LDs** | Leukodystrophies |
| **MLC** | Megalencephalic leukoencephalopathy with subcortical cysts |
| **MLD** | Metachromatic leukodystrophy |
| **MRI** | Magnetic resonance imaging |
| **MS** | Multiple Sclerosis |
| **NAA** | N-Acetyl Aspartic Acid |
| **NCL** | Neuronal ceroid lipofuscinoses |
| **NCV** | Nerve Conduction Velocity |
| **NRC** | National Research Centre |
| **NRG-**1 | Neuregulin-1 |
| **PAH** | Phenylalanine hydroxylase |
| **Phe** | Phenylalanine |

| | |
|---|---|
| **PIND** | Progressive intellectual and degenerative diseases |
| **PKU** | Phenylketonuria |
| **PLP1** | Proteolipid protein 1 |
| **PMD** | Pelizaeus – Merzbacher Disease |
| **PMDL** | Pelizaeus – Merzbacher Disease Like |
| **PNS** | Peripheral nervous system |
| **PTPS** | 6-pyruvoyl-tetrahydropterin synthase. |
| **PYCR2** | Pyrroline -5 – Carboxylate Reductase |
| **SULF1** | sulfatase 1 |
| **SULF2** | Sulfatase 2 |
| **SUMF1** | Sulfatase Modifying Factor 1 |
| **T1-W** | T1-weighted sequence |
| **T2-W** | T2- weighted sequence |
| **VLCFA** | Very long chain fatty acids |
| **VWM** | Vanishing white matter disease |
| **VWMD** | Vanishing white matter disease |
| **WGS** | Whole-genome sequencing |
| **WM** | White matter |
| **XL-ALD** | X-linked adrenoleukodystrophy |