9Medical Studies
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Faculty of Postgraduate
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Neurobehavioral Assessment and Neuroimaging Correlation in Children with Cerebellar Malformations

Thesis Submitted for the Fulfillment of Ph.D. Degree in Childhood Studies

Presented by

Hasnaa Mohamed Elbendary *(M.Sc. Pediatrics)*

Research Assistant
Clinical Genetics Department
Human Genetics and Genome Research Division
The National Research Center

Supervised by

Prof. Dr. Ehab M. Eid
Professor of Public Health and Family Medicine
Department of Medical Studies for Children
Institute of Postgraduate Childhood Studies

Prof. Dr. Yasser E. Nassef
Professor of Child Health
National Research Centre

Dr. Ahmed F. Ahmed
Assistant Professor of Child Health
National Research Centre

Dr. Rasha T. Khattab
Lecturer of Radiology
Faculty of Medicine
Ain Shams University

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Cognitive Impairment in Cerebellar Malformations: A Logit Model Based on Cognitive Testing

Hasnaa Mohamed Elbendary *, Yasser Nassef **, Ahmed Fathay *** Rasha Khattab ****Walaa Amin*** and wafaa Osaman**** Ehab Mohamed Eid *****

* Clinical Genetic Department, National Research Centre, Egypt.
** Professor of Preventive Medicine and Epidemiology, Medical Studies Department for Children, Faculty of Postgraduate Childhood Studies, Ain Shams University, Egypt.
*** Child Health department, National Research Centre, Egypt.
**** Lecturer of Radiology, Faculty of Medicine, Ain Shams University, Egypt.
***** Lecturer of Pediatric, Faculty of Medicine, Ain Shams University, Egypt.
E-mail: Hasnaa_mohamed24@yahoo.com

Abstract

Background: The complex development of the cerebellum together with the high number of involved genes and the long embryonic development of the cerebellum make the cerebellum vulnerable to a broad spectrum of malformations and disruption. Neuroimaging plays a key role in the diagnosis of posterior fossa malformations.

Objective: Our scope was to determine to what extent cerebellar malformations affect behaviour, cognition, motor functions and quality of life.

Methodology: A total of 60 patients with proven cerebellar malformations were enrolled in our cross-section study and rated into five groups according to MRI finding. Patients were undergoing behavioural, cognitive, motor and quality of life assessment by CBCL, Wechsler IQ, SARA and quality of life scoring respectively.

Results: Our findings highlighted that children with cerebellar malformations experience high prevalence of Cogntive as IQ ranges among our studied population between 25-99, mean was 57.7, cases with intellectual disability represented 90% of our studied population , in addition to behavioral and motor impairment as well as poor quality of life due to functional disabilities, except patients with DWM although they have favourable neurodevelopmental outcome al all patients with DWM scored FSIQ range between 91-99 ,but this does not prevent them from experiencing poor quality of life, as 40.0 of patients showing very poor QOL, 20.0% poor QOL, 28.3% neither poor nor good QOL, 11.7 good QOL, NO one has very good QOL

ranking 38.3 are very dissatisfied, 12% dissatisfied, 28.3 % neither satisfied nor dissatisfied, 13.3 are satisfied, no one in our study showing very satisified way of living ranking.

Conclusion: Cerebellar malformation is heavily connected to cognition and behaviour as well as motor function.

Recommendations: Further studies need to be done in order to confirm role of cerebellum in higher orders functions like behavior, emotion processing and cognition.

Keywords: CBCL, SARA, WHOQOL BERF, Wechsler, Cerebellar malformations.

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List of Abbreviations

Abbreviation	Full Term
ADHD	Attention deficit hyperactivity disorder
AMPD2	Adenosine Monophosphate Deaminase 2
ANOVA	Analysis of variance
ASD	Autism spectrum disorders
CA	Cerebellar agenesis
CASK	Calcium/Calmodulin Dependent Serine Protein Kinase
CBA	Cerebellar atrophy
CBCL	Child Behavior Check list
CBH	Cerebellar hypoplasia
CC	Corpus callosum
CCAS	Cerebellar cognitive affective syndrome
CDG	Congenital Disorders of Glycosylation
CDG1A	Congenital disorder of Glycosylation type Ia
CH	Cerebellar hypoplasia
CHMP1A	Charged Multivesicular Body Protein 1A
CLP1	Cleavage and polyadenylation factor I subunit 1
CMS	Chudley-McCullough syndrome
CNS	Central nervous system
COASY	Coenzyme A Synthase
CVH	Cerebellar vermian hypoplasia
DSD	Disorder of Sex Development
DTI	Diffusion tensor imaging
DWC	Dandy–Walker complex
DWM	Dandly Walker Malformations
DWV	Dandly Walker variant
EXOSC8	Exosome complex component 8
FSIQ	Full Scale Intelligent Quotient
GCH	Global cerebellar hypoplasia
HS	Highly significant
ICARS	International Cooperative Ataxia Rating Scale
IQ	Intelligent quotient

Abbreviation	Full Term
IVH	Inferior vermian hypoplasia
JS	Joubert Syndrome
MCAP	Megalencephaly–capillary malformation
MCM	Mega-cisterna magna
MEB	Muscle-eye-brain disease
MPPH	Megalencephaly–polydactyly–polymicrogyria–hydrocephalus
MHM	Midbrain–hindbrain malformations (MHM)
MRI	Magnetic resonance imaging
MRS	Magnetic Resonance Spectroscopy
MTI	Molar tooth Image
OFC	Occipito-frontal head circumference
OMIM	Online Mendelian Inheritance in Man
P. value	Probability value
PCH	Ponto Cerebellar hypoplasia
PCLO	Piccolo presynaptic cytomatrix protein
PET	positron emission tomography
PTCD	Pontine Tegmental Cap Dysplasia
PTF1A	Pancreas Associated Transcription Factor 1a
PWI	perfusion-weighted imaging
QOL	Quality of life(QOL)
RARS2	Arginyl-tRNA synthetase 2,
RES	Rhomboencephalosynapthies
SARA	Scale for assessment and rating of Ataxia
SCA	Spinocerebellar ataxia
SCPs	superior cerebellar peduncles
SD	Standard deviation
SEPSECS	Sep (O-phosphoserine) tRNA Sec (selenocysteine) synthase
SMA	Spinal muscle atrophy
SPSS	Statistical Package for the Social Sciences
SWI	Susceptibility-weighted imaging

Abbreviation	Full Term
TBC1	**Domain Family Member 23**
TBC1D23	TBC1 Domain Family Member 23
TOE1	Target Of EGR1, Exonuclease
TSEN2	tRNA-splicing endonuclease subunit Sen2
TSEN54	tRNA splicing endonuclease complex.
UCH	Unilateral cerebellar hypoplasia
US	Ultrasound
VACTERL	Vertebral defects, Anal atresia, Cardiac defects, Tracheo-Esophageal fistula, Renal defects and Limb defects
VRK	Vaccinia-related kinase
WAIS-IV	Wechsler Adult Intelligence Scale 4th edition
WHO	World health organization
WHOQOL	World health organization quality of life BREF form
WISC	The Wechsler Intelligence Scale for Children
WISC-IV	The Wechsler Intelligence Scale for Children 4th edition
WISC-V	The Wechsler Intelligence Scale for Children 5th edition
WPPSI	The Wechsler Preschool and Primary Scale of Intelligence-IV
WPPSI-R	Wechsler Preschool & Primary Scale of Intelligence Revised
WWS	Walker Warburg syndrome

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