

**9Medical Studies
Department for Children**



**Faculty of Postgraduate
Childhood Studies**

Neurobehavioral Assessment and Neuroimaging Correlation in Children with Cerebellar Malformations

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

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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 Cognitive 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 satisfied 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.

List of Contents

Title	Page
Acknowledgment	i
Abstract	iii
Contents	iv
List of Abbreviations	v
List of Figures	viii
List of Tables	xiii
Introduction	1
Aim of the Study	6
Review of literature	7
• Chapter 1: Cerebellum Embryology and Malformations	7-34
• Chapter 2: Role of Neuroimaging in the Diagnosis of Prenatal Cerebellar Disruption	35-40
• Chapter 3: Cerebellar and cognition	41-54
• Chapter 4: Assessment of the IQ	55-69
Methodology	70
Results	80
Discussion	128
Conclusion & Recommendations	156
Summary	160
References	163
Appendices	218
Arabic summary	-

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	Dandy Walker Malformations
DWV	Dandy Walker variant
EXOSC8	Exosome complex component 8
FSIQ	Full Scale Intelligent Quotient
GCH	Global cerebellar <u>hypoplasia</u>
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 4 th 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 4 th edition
WISC-V	The Wechsler Intelligence Scale for Children 5 th edition
WPPSI	The Wechsler Preschool and Primary Scale of Intelligence-IV
WPPSI-R	Wechsler Preschool & Primary Scale of Intelligence Revised
WWS	Walker Warburg syndrome

List of Figures

Figure	Title	Page
Figure (1) :	Dandy walker malformation	13
Figure (2) :	MRI showing complete cerebellar agenesis and vermal agenesis	15
Figure (3) :	Rhombencephalosynapsis demonstrates continuity of the cerebellar hemispheres	17
Figures (4):	Total aplasia of the right cerebellar hemisphere	20
Figures (5):	Image of Joubert syndrome shows (molar tooth sign)	23
Figures (6):	MR images in different PCH subtypes	32
Figure (7):	Simplified representation of main characteristic symptoms in PCH	33
Figure (8):	Midsagittal T1-weighted image of pontine tegmental cap dysplasia	34
Figure (9):	Parcellation scheme used to divide the cerebellum into seven regions	37
Figure (10):	Examples of cerebellar parcellations	38
Figure (11):	Depicting cerebello-cerebral connectivity network subserving cognitive and affective processes	44
Figure (12):	The microstructural organization of the cerebellar cortex	45
Figure (13):	Flow chart detailing the study population.	77
Figure (14):	Pie Chart showing sex distribution of the studied patients	82
Figure (15):	Pie Chart showing consanguinity distribution among our studied patients	82
Figure (16):	Pie Chart showing oculomotor apraxia distribution among our studied patients	83
Figure (17):	Percentage of nystagmus among our studied patients	83

Figure	Title	Page
Figure (18):	Tone distribution of the studied patients	84
Figure (19):	Percentage of each catogrey of the studied patients according MRI findings	85
Figure (20):	Percentage of the Full Scale Intelligent Quotient among our studied patients	87
Figure (21):	Percentage of the Verbal Scale Intelligent Quotient among our studied patients	87
Figure (22):	: Illustration for the percentage of the Performance Scale Intelligent Quotient among our studied patients	88
Figure (23):	Percentage of the studied patients as regards the Mean Anxious T score	90
Figure (24):	Percentage of the studied patients as regards the Mean Withdrawal T score	91
Figure (25):	Percentage of the studied patients as regards the Mean Somatic T score	91
Figure (26):	Percentage of the studied patients as regards the Mean Social T score	92
Figure (27):	Percentage of the studied patients as regards the Mean Thought T score	92
Figure (28):	Percentage of the studied patients as regards the Mean Attention T score	93
Figure (29):	Percentage of the studied patients as regards the Mean Rule breaking behavior T score	93
Figure (30):	Percentage of the studied patients as regards the Aggression T score	94
Figure (31):	The studied patients QOL scores, the total score ranges from 8 to 20	98
Figure (32):	The studied patients QOL ranking scores	98
Figure (33):	OFC among different groups,	100
Figure (34):	Length distribution among our studied patients	100

Figure	Title	Page
Figure (35):	Percentage of each group as regards age of 1st walk and 1st spoke	101
Figure (36):	Oculomotor distribution among all groups in correlation to MRI findings	101
Figure (37):	Nystagmus distribution among our studied patients	102
Figure (38):	Tone distribution among our different groups	102
Figure (39):	FSIQ among different groups in correlation to MRI findings	104
Figure (40):	IQ range among different groups	104
Figure (41):	Verbal IQ range distribution among different groups in correlation to MRI	105
Figure (42):	Verbal IQ range among different groups	105
Figure (43):	Performance IQ distribution among different groups in correlation to MRI findings	106
Figure (44):	Performance IQ range among different groups	106
Figure (45):	CBCL scores among different groups	109
Figure (46):	Percentage of CBCL scores among MTI group	109
Figure (47):	Percentage of CBCL scores among CBH group	110
Figure (48):	Percentage of CBCL scores among CVH group	110
Figure (49):	Percentage of CBCL scores among PCH group	111
Figure (50):	Percentage of CBCL scores among DWM group	111
Figure (51):	SARA scale among different groups	113
Figure (52):	SARA total scores among different groups	113
Figure (53):	WHOQOL different parameters among different groups	115

Figure	Title	Page
Figure (54):	Negative correlation between Verbal IQ and MRI severity Score	118
Figure (55):	Negative correlation between Verbal IQ and MRI severity Score	118
Figure (56):	Positive correlation between somatic T score of CBCL and MRI severity Score	119
Figure (57):	Positive correlation between stance item of SARA scale and MRI severity Score	119
Figure (58):	Positive correlation between sitting item of SARA scale and MRI severity Score	120
Figure (59):	Positive correlation between speech item of SARA scale and MRI severity Score	120
Figure (60):	Positive correlation between finger chase item of SARA Scale and MRI severity Score	121
Figure (61):	Positive correlation between nose finger item of SARA Scale and MRI severity Score	121
Figure (62):	Positive correlation between Fast alternating movements item of SARA scale and MRI severity Score	122
Figure (63):	Positive correlation between Heel shin item of SARA Scale and MRI severity Score	122
Figure (64):	: Positive correlation between of SARA total score and MRI severity Score	123
Figure (65):	Positive correlation between physical parameter of WHOQOL and MRI severity Score	123
Figure (66):	Positive correlation between Psychological parameter of QOL and MRI severity Score	124

Figure	Title	Page
Figure (67):	Positive correlation between Enviromental parameter of WHOQOL and MRI severit109y Score	124
Figure (68):	Midline Sagittal view demonstrates long, thick, SCP in Joubert syndrome patients (MTI)	125
Figure (69):	Axial view demonstrates thin MBHB junction with deep interpeduncular fossa representing molar tooth sign in Joubert syndrome patients	125
Figure (70):	Sagittal and axial view showing hypoplasia of both cerebellar hemispheres with vermian affection Cerebellar Hypoplasia (CBH) Patients	126
Figure (71):	Sagittal and axial view demonstrate inferior vermian hypoplasia of the cerebellum in Cerebellar Vermian Hypoplasia (CVH) patients	126
Figure (72):	Sagittal and axial view demonstrates hypolpasia of the Pons, thin brain stem with hypoplasia of cerebellum in Ponto Cerebellar Hypoplasia (PCH) patients	127
Figure (73):	Scale for the assessment and rating of ataxia (SARA)	218
Figure (74):	The World Health Organization Quality of Life (WHOQOL)-BREF	221
Figure (75):	Child Child Behavior Check list	224
Figure (76):	Hand-scored profile from the CBCL 2-3 years	228
Figure (77):	Hand-scored profile from the CBCL 6-16 years	229

List of Tables

Table	Title	Page
Table (1):	Intelligence scoring	62
Table (2):	The studied patients clinical info and demographic data	81
Table (3):	Percentage of each catogrey of the studied patients according to MRI findings	85
Table (4):	Illustration of the Whole studied patients IQ.	86
Table (5):	Illustration of CBCL scores among whole studied sample	89
Table (6):	The studied patients SARA scores	95
Table (7):	WHOQOL scores among our studied patients	96
Table (8):	The studied patients WHOQOL ranking scores	97
Table (9):	The studied patients clinical info. and demographic data in correlation to MRI findings	99
Table (10):	FS1Q among different groups in correlation to MRI findings	103
Table (11):	Comparsion between CBCL different scores among whole studied patients	107
Table (12):	SARA scale among different groups	112
Table (13):	Comparison between WHOQOL parameters among different groups	114
Table (14):	Comparison between WHOQOL ranking scores among different groups	116
Table (15):	Correlations between MRI severity Scoring as regards age, OFC, length, age of 1st walk and 1st spoke	117