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Assessment of Intelligence Quotient in Children with Mucopolysaccharidoses

Tarek Hamed Attia, Mohamed Refaat Beshir, Ahmed Hosni Mohamed Abdel Fattah, Rania Emad Abd-elsamad Mohamed*

Department of Pediatrics, Faculty of Medicine, Zagazig University, Egypt

***Corresponding author:** Rania Emad Abd-elsamad Mohamed, **Mobile**: (+20)01093543650, **Email:** dr.rania.emad@gmail.com

ABSTRACT

Background: The category of lysosomal storage diseases known as mucopolysaccharidoses (MPS) is characterized by the buildup of glycosaminoglycans (GAGs) in tissues and organs as a result of the lack of certain enzymes needed for the breakdown of cellular GAGs. For all individuals, the Wechsler and Primary Scale of Intelligence (WPPSI) was used to calculate their intelligence quotient (IQ) and evaluate their cognitive abilities. **Objective:** The aim of the present study was to assess intelligence quotient in MPS children. **Patients and Methods:** This study was conducted in 72 children in Pediatrics Department in Zagazig University Hospital, over 6 months. They were divided into 2 groups; A case group included 24 children diagnosed with MPS in Pediatrics Department in Zagazig University Hospital, and a control group included 48 children matched for age and sex. **Results:** This study showed that age of onset of disease ranged between 1 and 6 with mean 1.96 (SD 1.30) years. This study showed that the mean age of diagnosis was 5.25 (SD 2.83) years and ranged between 1 and 13. The present study showed that all cases with MPS had positive coarse facies. The mean value of full IQ was statistically lower among the case group than the control group. **Conclusion:** Among children with MPS, there was a measurable decrease in IQ associated with positive family history. IQ was low among children with MPS compared with controls.

Keywords: Intelligence quotient, Children, Mucopolysaccharidoses, case control study, Zagazig University.

INTRODUCTION

A series of lysosomal storage diseases known as mucopolysaccharidoses (MPS) are characterised by the buildup of glycosaminoglycans (GAGs) in tissues and organs as a result of a lack of the enzymes necessary for the breakdown of cellular GAGs [(1)].

MPS are uncommon hereditary metabolic illnesses brought on by lysosomal enzyme deficiency or dysfunction. Although MPS patients may be intelligent, cognitive deficits are frequently seen in this population [(2)].

Patients with MPS often present at birth as healthy, and as they mature, the clinical symptoms get progressively worse. Within each of the several MPS subtypes as well as within the illnesses themselves, there are many varied clinical manifestations and rates of development. Several of these conditions cause significant morbidity and early mortality [(3)].

The course of morbidity and death for various MPS illnesses has been changed by treatments including hematopoietic stem cell transplantation (HSCT) and enzyme replacement therapy (ERT) [(4)]. The mental processes involved in learning and the integration of these processes into behaviors like learning, attention, memory, intelligence (intelligence quotient; IQ), and consciousness are collectively referred to as cognition [(5)].

Special intelligence test: The Wechsler and Primary Scale of Intelligence (WPPSI) was used to measure each participant's IQ and evaluate their cognitive abilities. School systems and psychologists give children between the ages of 6 and 16 the WISC Test (Wechsler Intelligence Scale for Children), an IQ test. The goal of the test is to identify a student's cognitive strengths and limitations as well as whether or not they are gifted [(6)].

The aim of the present study was to assess IQ in MPS children.

PATIENTS AND METHODS

Type of study: Case control study.

Study Setting: This study was conducted in the Pediatrics Department at Zagazig University Hospital, over 6 months.

Study population: This study included 72 children who were divided into 2 groups; the case group included 24 children diagnosed with MPS, and the control group included 48 children matched for age and sex.

Inclusion criteria: Children aged <18 years old, and children diagnosed as MPS who receive ERT.

Exclusion criteria: Cases with CNS disorders as brain tumors and congintal anomalies, and cases with lost follow up.

Sample size: Assuming the total number of cases that met inclusion and exclusion criteria was 4 cases/ month during the study period (6 months); all MPS cases (24 cases) were included as a comprehensive sample and 48 children as a control group (ratio 1:2).

Methods:

All the participating patients and controls were subjected to the following:

Full history taking: personal history, consanguinity, family history, age of onset of the disease, age of diagnosis of the disease, and age of treatment.

Examination:

General examination: General examination included coarse facies, weight, length, head circumferences,

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heart and chest examination, abdominal examination and special focus on MPS examination [motor system and sensory system and reflexes]. We found in the cases abnormal general examination (examples: enlarged forehead, enlarged eyebrow, microcephaly and large protruated tongue).

Investigation:

Wechsler Intelligence Scale-Revised for Children (WISC). WISC reflects the intellectual performance through verbal (VIQ), performance (PIQ), and full scale IQ. It is used for assessment of different cognitive functions, including executive functions, and provides an overview of the integrity of cognitive abilities [(7)] and its Arabic version has been validated [(8)].

The WISC comprises ten subtests numbered in order of their administration, in which verbal and performance tests are alternated. The Wechsler Intelligence Scale for Children assesses a panel of cognitive function parameters. Five verbal linguistic subtests underlie the verbal IQ; information (factual knowledge), similarities (verbal concept formation), arithmetic (mental arithmetic), vocabulary (word definitions), and comprehension (social understanding). Five visuospatial subtests underlie the performance IQ: picture completion (perception of visual detail), picture arrangement (logical reasoning), block design (visual analysis), object assembly (part/whole construction), and coding (symbol manipulation).

The test also measures the total IQ. This test is also a practical tests of the working memory. Scoring is as follows: >90 is normal, 90-70 borderline, and <70 is borderline. VIQ and PIQ were interpreted as normal if >7 in each subscale, abnormal if <7 [(6)].

Ethical consent:

An approval of the study was obtained from Zagazig University Academic and Ethical Committee. Every legal guardian of the patient signed an informed written consent for acceptance of participation in the study. This work has been carried out in accordance with The Code of Ethics of the World Medical Association (Declaration of Helsinki) for studies involving humans.

Statistical analysis

The collected data were coded, processed and analyzed using the SPSS (Statistical Package for Social Sciences) version 22 for Windows® (IBM SPSS Inc, Chicago, IL, USA). Data were tested for normal distribution using the Shapiro Walk test. Qualitative data were represented as numbers and relative percentages. Chi square test ($\chi 2$) to calculate difference between two or more groups of qualitative variables. Quantitative data were expressed as mean ± SD (Standard deviation). Independent samples t-test was used to compare between two independent groups of normally distributed variables (parametric data). P value ≤ 0.05 was considered significant.

RESULTS

Table 1 shows that there were no statistically significant difference between the case group and the control group regarding the demographic data.

Table (1): Comparison between the case group and the control group regarding the demographic data.

Variable			Case group No. (24)	Control roup No. (48)	t test	P value
Age (years)	**Mean ± SD**		8.23± 2.65	8.04± 1.31	0.415	.679
Gender	**Female**	**No. (%)**	7 (29.2%)	24(50.0%)	X^2 2.832	.092
	Male	**No. (%)**	17 (70.8%)	24 (50.0%)		

Table 2 shows that there were statistically significant difference between Cases group and Controls group regarding consanguinity **Table (2)**.

Table (2): Comparison between the case group and the control group regarding consanguinity.

Variable			Case group	Control group	X^2	P value
Consanguinity	**No**	**No. (%)**	8(33.3%)	32(66.7%)	2.832	.042
	Yes	**No. (%)**	16(66.7%)	16(33.3%)		

Table 3 shows that 8 (33.3%) children of the case group had a family history of MPS.

Table (3): Family history of MPS among the case group.

Variable		No.	%
Family history	**No**	16	66.7
	Yes	8	33.3

Table 4 shows age of onset of disease ranged between 1 and 6 with mean 1.96 (SD 1.30) years, Age of diagnosis ranged between 1 and 13 with mean 5.25 (SD 2.83) years, and age of starting treatment ranged between 4 and 14 with mean 7.12 (SD 2.62) years.

Table (4): Age of onset of disease, age of diagnosis, and age of starting treatment among MPS cases.

Variable		Cases (No. 24)
Age of onset of disease (years)	**Range**	1.00-6.00
	Mean ± SD	1.96±1.30
Age of diagnosis (years)	**Range**	1.00-13.00
	Mean ± SD	5.25±2.83
Age of starting treatment (years)	**Range**	4.00-14.00
	Mean ± SD	7.12±2.62

Mean value of full IQ was statistically higher among the case group than the control group. There were no statistically significant differences between sex and full IQ or between consanguinity and full IQ **Table (5).**

Table (5): Comparison between Cases group and Controls group regarding full IQ.

Variable		Case group	Control group	t test	P value
Full IQ	**Mean ± SD**	77.38± 17.26	103.88± 10.99	-7.920	0.000

Mean value of full IQ was statistically lower among MPS children with positive family history than those negative family history **Table (6)**.

Table (6): Relation between full IQ and sex, consanguinity and family history among cases group.

Variable		Full IQ Mean ± SD	t test	P value
Sex	**Female**	82.86 ± 16.66	0.997	0.329
	Male	75.12 ± 17.48		
Consanguinity	**No**	71 ± 15.61	1.684	0.208
	Yes	80.56 ± 17.63		
Family history	**No**	83.06 ± 15.33	6.444	0.019
	Yes	66 ± 15.92		

DISCUSSION

This present study showed that the mean age of diagnosis of MPS was 5.25 (SD 2.83) years and ranged between 1 and 13.

Lin *et al.* [9] in their retrospective analysis included 129 Taiwanese patients with MPS (age range 0.7 to 19.5 years; median age 7.9 years) from eight medical centers in Taiwan from January 1996 through December 2018. The mean age at the diagnosis of M6+PS III in their patients was 4.6 years (n = 27). Consistently, **Truxal *et al.*** [10] reported a mean age of the diagnosis of MPS III of 3.4 years (n = 25). According to **Colmenares-Bonilla *et al.*** [11] the mean age for diagnosis of MPS was 4.9 years.

The present study showed that all cases with MPS had positive coarse facies. **Zelei et al.** [12] reported that the general manifestations of MPS include coarse facies and thick eyebrows.

In the current study mean value of full IQ was statistically lower among the case group than the control group. This agrees with **Izumi *et al.*** [13] who aimed to delineate the psychological status of 10 patients with the attenuated phenotype of MPS type II and their parents (six fathers and five mothers) for the improvement of clinical management. Their results indicated that the mean IQ scores were slightly lower than the general population.

CONCLUSION

Among children with MPS, there was a measurable decrease in IQ associated with positive family history. IQ was low among MPS children compared with the controls.

Conflict of interest: The authors declare no conflict of interest.

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Author contribution: Authors contributed equally in the study.

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