

STUDY OF SERUM LEVEL OF L-CARNITINE IN CHILDREN WITH AUTISM SPECTRUM DISORDER

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Introduction

Autism Spectrum Disorder (ASD) is a disorder characterized by language, social and behavioral impairments. Its prevalence is increasing and researches to detect etiology are growing.

It is defined by the American psychiatric association's diagnosis and statistical manual of mental disorders (DSM-5) as a single disorder that includes disorders that were previously considered separate-autism, Asperger's syndrome, childhood disintegrative disorder and pervasive developmental disorder not otherwise specified.

Early diagnosis means early treatment and better prognosis and outcome. The search for ASD biomarkers for early diagnosis is growing. Several studies have detected mitochondrial dysfunction as a cause of ASD and L-carnitine level as a marker to the mitochondrial function.

Aim of the work

We aimed to study the role of serum level of L-carnitine in the pathogenesis of autism manifestations and using it as a biomarker for diagnosis of ASD and assess if there is a relation between it and ASD severity.

Subjects and Methods

This case -control study was conducted on (60) children aged from 2 to 6 years (30 children diagnosed with ASD based on DSM-5 diagnostic criteria and were recruited from Outpatient Neurobehavioral Clinic of Alexandria University Children's Hospital (AUCH) (group I) and 30 age and sex matched sibling of group 1 who are not fulfilling the DSM-5 diagnostic criteria of ASD as a controls (group II) Children who had specific syndromes as fragile x syndrome and tuberous sclerosis and children who had chronic diseases as epilepsy and IEM were excluded from our study.

All cases were subjected to full history taking including medical history, perinatal history, developmental milestones, onset of symptoms, family history. Clinical examination with thorough neurological examination was done. Severity of ASD symptoms was assessed by using Childhood autism rating scale (CARS). And subjected to laboratory investigations using enzyme linked immune sorbent assay ELISA kits to detect serum level OF L-carnitine.

Results

Table 1: Comparison between the two studied groups according to demographic data

	Cases (n = 30)		Control (n = 30)		Test of sig.	P
	No.	%	No.	%		
Sex						
Male	25	83.3	18	60.0	$\chi^2=$ 4.022*	0.045*
Female	5	16.7	12	40.0		
Residence						
Rural	9	30.0	2	6.7	$\chi^2=$ 5.455*	0.020*
Urban	21	70.0	28	93.3		
Education						
Illiterate	3	10.0	2	6.7	$\chi^2=$ 0.218	^{FE} p= 1.000
Educated	27	90.0	28	93.3		
Age (years)						
Min. – Max.	26.0 – 62.0		30.0 – 55.0		t= 0.873	0.387
Mean ± SD.	43.50 ± 10.43		41.53 ± 6.59			
Median (IQR)	42.50 (35.25 – 49.25)		40.0 (36.50 – 47.25)			

Table 2: Distribution of the studied cases according to CARS and severity (n = 30) according to the autism severity measured by CARS

	No.	%
CARS		
Mild-ASD (15-29)	3	10.0
Moderate (30 - 35)	3	10.0
Severe (36 – 60)	24	80.0
Min. – Max.	15.0 – 49.0	
Mean ± SD.	39.93 ± 7.93	
Median (IQR)	41.50 (38.75 – 44.25)	
Severity		
Mild	3	10.0
Moderate	3	10.0
Severe	24	80.0

Table 3: Relation between sex and L- carnitine level in cases group

L-Carnitine level	Sex		t	p
	Males (n = 25)	Females (n = 5)		
Min. – Max.	16.0 – 40.0	25.0 – 32.0	1.077	0.296
Mean ± SD.	25.92 ± 7.47	28.0 ± 2.74		
Median (IQR)	24.0 (19.50 – 32.0)	28.0 (25.5 – 30.5)		

Conclusion

The finding of the current study suggest that

- ASD cases have significantly low serum level of L-Carnitine than healthy controls.
- L-carnitine was lower in males than females.
- There is a negative correlation between L-carnitine level and CARS as when the level of the CARS increases, the level of L-carnitine decrease and it was lower in severe group.