Psychiatry; 73:20-30.

10. Eijk- Van Os PG, Schouten JP. (2011) Multiplex Ligation- dependent Probe Amplification (MLPA®) for the detection of copy number variation in genomic sequences. **Methods Mol Biol**; 688:97- 126.
11. El- Sady S., El- Shoubary A., Hafez N., Abd El- Halim I., Ewis A., Abu- Hasseba A. (2011). A Standardization, translation and modification of the preschool language scale- 4. **Doctoral Thesis** of Phoniatrics presented by A. Abu- Haseeba, faculty of medicine, Ain Shams University. Egypt.
12. Emberti Gialloreti L., Mazzone L., Benvenuto A., Fasano A., Alcon AG, Kraneveld A., Moavero R., Raz R., Riccio MP, Siracusano M., Zachor DA, Marini M., Curatolo P. (2019). Risk and Protective Environmental Factors Associated with Autism Spectrum Disorder: Evidence- Based Principles and Recommendations. **J. Clin Med**. 8; 8(2): 217.
13. Fu J. M., Satterstrom F. K., Peng M. et.al. (2022). Rare coding variation provides insight into the genetic architecture and phenotypic context of autism. **Nat Genet** 54, 1320- 1331.
14. Genovese A., Butler M. G. (2020). Clinical Assessment, Genetics, and Treatment Approaches in Autism Spectrum Disorder (ASD). **Int. J. Mol. Sci.**, 21, 4726.
15. Howard J., Herold B., Major S., Leahy C., Ramseur K., Franz L., Deaver M., Vermeer S., Carpenter K. L., Murias M., Huang W. A.& Dawson G. (2023). Associations between executive function and attention abilities and language and social communication skills in young autistic children. **Autism**, 0(0).
16. Hyman S. L., Levy S. E., Myers S. M., Kuo D. Z., et.al. (2020). Identification, Evaluation, and Management of Children with Autism Spectrum Disorder. **Pediatrics**; 145: e20193447.
17. Iyer J., Singh M. D., Jensen M., Patel P., Pizzo L., Huber E., et.al. (2018). Pervasive genetic interactions modulate neurodevelopmental defects of the autism- associated 16p11.2 deletion in Drosophila melanogaster. **Nat. Commun.** 9:2548.
18. Kim H., Ahn J., Lee H., Ha S. & Cheon K. A. (2020). Differences in Language Ability and Emotional- Behavioral Problems according to Symptom Severity in Children with Autism Spectrum Disorder. **Yonsei Medical Journal**, 61, 880- 890.
19. Maenner M. J., Shaw K. A., Baio J., Washington A., Patrick M., DiRienzo M., Dietz P. M. (2021). Prevalence of autism spectrum disorder among children aged 8 years—Autism and developmental disabilities monitoring network, 11 Sites, United States, 2016. **MMWR Surveillance Summaries**, 70, 1-16.
20. Major S., Isaev D., Grapel J., et.al, (2022). Shorter average look durations to dynamic social stimuli are associated with higher levels of autism symptoms in young autistic children. **Autism: The International Journal of Research and Practice**, 26(6), 1451-1459.
21. Montagut- Asunción M., Crespo- Martín S., Pastor- Cerezuela G., D'Ocon- Giménez A. (2022). Joint Attention and Its Relationship with Autism Risk Markers at 18 Months of Age. Children (Basel). 13;9(4): 556.
22. Morsanyi K., Stamenković D. (2021). Idiom and proverb processing in autism: a systematic review and meta- analysis, **Journal of Cultural Cognitive Science**, 5, pp. 367- 387.
23. Nevill R., Hedley D., Uljarević M., Sahin E., Zadek J., Butter E., Mulick J. A. (2019). Language profiles in young children with autism spectrum disorder: A community sample using multiple assessment instruments. **Autism**, 23, 141- 153.
24. Özaltun, M. F., S. Geyik, and Ş. G. Yılmaz. (2021). Screening for Copy Number Variations of the 15q13.3 Hotspot in CHRNA7 Gene and Expression in Patients with Migraines. **Current Issues in Molecular Biology**, 43(2): p. 1090- 1113.
25. Ridderinkhof A., de Bruin EI, van den Driesschen S., Bögels SM. (2020). Attention in Children With Autism Spectrum Disorder and the Effects of a Mindfulness- Based Program. **J Atten Disord**. ; 24(5): 681-692.
26. Schouten JP, McElgunn CJ, Waaijer R., Zwijnenburg D., Diepvens F., Pals G. (2002). Relative quantification of 40 nucleic acid sequences by multiplex ligation- dependent probe amplification. **Nucleic Acids Res.**; 15; 30(12): e57.
27. Tafaroji YM, Kamari E. (2020). Investigating mean length of utterance (MLU) in monolingual Persian speaking children with autism spectrum disorder (ASD). **Int J Health Stud**. 6(2): 15-23.
28. Yoon SH, Choi J., Lee WJ, Do JT (2020). Genetic and Epigenetic Etiology Underlying Autism Spectrum Disorder. **J Clin Med**. 31;9(4): 966.
29. Zhuo Fu, Yue- Xin Jia, Jun- Xian Fu, et.al. (2021). A case of 15q11-q13 duplication syndrome and literature review. **Brain Behav**; 11:e2219.

ASD and its heterogeneity. (Genovese& Butler, 2020). Another explanation of our results is attributed to the epigenetic regulation of gene expression which plays an important role in pathogenesis of ASD. Numerous mechanisms, including non- coding RNA, antisense RNA, and DNA methylation, are involved in the intricate regulation of gene expression. (Yoon et.al, 2020). Consistent with our results regarding 16p11.2 CNV, a study conducted to detect the genetic impact of 16p11 deletion in autism revealed that no single gene in the 16p11.2 region was shown to be substantially related with autism. (Fu et.al, 2022).

Assessment of language among the studied cases revealed that both the receptive, expressive and total language age were markedly affected in the autistic children compared to neurotypical children and the receptive language was more affected than the expressive language.

It is well known that the child's linguistic communication is impacted by ASD. Even more children with ASD exhibit variability in their language expression, including deficiencies in figurative speech, joint attention, and social pragmatic abilities. (Chan& Leung, 2022). Consistent with our results, a study that investigated the receptive and expressive language in ASD males and females showed that both groups displayed marked impairment of receptive skills with marked discrepancy between expressive and receptive language. (Barsotti et.al, 2023). On the other hand, another research that assessed the expressive and receptive language abilities of ASD cases found a significant difference between the two domains revealing that the receptive was better than expressive pattern in children with ASD. (Arutiunian et.al, 2021).

Comparison between the degree of social interaction and communication deficits, as well as restricted and repetitive behaviors, with receptive and expressive language revealed that the receptive language was mostly affected by the repetitive impairments among the cases while the expressive language was affected by all the domains equally.

Numerous research investigated the relationship between the degree of social communication impairments (SCI), restrictive and repetitive behaviors (RRB) with the language skills (receptive and expressive language), and emotional- behavioral issues. When compared to the severity of RRB, a study by Kim et.al., 2020, found no variations in language capacity; however, children with severe SCI had considerably reduced language skills. Another study by Nevill et.al., 2019 investigated factors associated with language in autistic children showed that language levels were weakly correlated with ASD severity and found no evidence of a substantial impact from restricted and repeated behaviors on these children's linguistic skills. (Nevill et.al, 2019).

Our study showed that the attention level in the autistic cases is greatly affected where most of our cases had moderate to severe attention deficits.

It was found that ASD is associated with diminished capacity to maintain focus on intricate, dynamic stimuli. Research has indicated that children with autism tend to look at stimuli for shorter periods of time and pay less attention to them when compared to neurotypical children. (Major et.al, 2022). Additionally, it was discovered that kids who were more adept at keeping their attention on the stimuli displayed lower levels of social withdrawal, improved social communication skills, and less symptoms of autism as well as higher levels of expressive language ability. (Howard et.al., 2023). In an investigation on the predictive power of joint attention on ASD, joint attention changes at 8 and 12 months of age were linked to early symptoms of ASD at 18 months of age. (Montagut et.al, 2022).

Conclusion:

There is no evidence based single pathway that results in symptoms of ASD. Multi- gene sequencing or epigenetic alternations hold promise in solving this molecular puzzle. Language skills plays a major role in the outcomes of ASD as development of language abilities predict social functioning, academic achievement, and psychiatric outcome in childhood. Therefore, it is critical that researchers learn more about the factors behind the disparate levels of language development in children with ASD.

References:

1. American Psychiatric Association (2022). **Diagnostic and statistical manual of mental disorders** (5th ed., text rev.) American Psychiatric Publishing.
2. Arutiunian V., Lopukhina A., Minnigulova A., Shlyakhova A., Davydova E., Pereverzeva D., Sorokin A., Tyushkevich S., Mamokhina U., Danilina K., Dragoy O. (2021) Expressive and Receptive Language in Russian Primary- School- Aged Children with Autism Spectrum Disorder. **Res Dev Disabil**. ; 117:104042.
3. Barsotti J., Mangani G., Nencioli R., Narzisi A., Pfanner L., Chilosi AM., Cipriani P., Mancini A., Cosenza A., Tancredi R., Calderoni S. (2023). Sex/ Gender Differences in the Language Profiles of Italian Children with Autism Spectrum Disorder: A Retrospective Study. **J Clin Med**. 26;12(15): 4923.
4. Bhandari R., Paliwal J. K., Kuhad A (2020). Neuropsychopathology of Autism Spectrum Disorder: Complex Interplay of Genetic, Epigenetic, and Environmental Factors. **Adv. Neurobiol**; 24:97-141.
5. Chan JKY, Leung PWL (2022). Common outcome, different pathways: Social information- processing deficits in autism spectrum disorder and attention- deficit/hyperactivity disorder. **World Journal of Psychiatry**; 12(2): 286-297.
6. Chung WK, Roberts TPL, Sherr EH, Green Snyder LA, Spiro JE (2021). 16p11.2 deletion syndrome. **Curr Opin Genet Dev**. 68:49-56.
7. Craig F., Margari F., Legrottaglie A. R., Palumbi R., De Giambattista C.& Margari L. (2016). A review of executive function deficits in autism spectrum disorder and attention- deficit/hyperactivity disorder. **Neuropsychiatric Disease and Treatment**; 12, 1191.
8. D'Abate L., et.al. (2019). Predictive impact of rare genomic copy number variations in siblings of individuals with autism spectrum disorders. **Nat Commun**. 10(1): 1-9.
9. D'Angelo D., et.al. (2016). Defining the effect of 16p11.2 duplications on cognition, behavior, and medical comorbidities. **JAMA**

Table (1) Age and gender of the studied group

Variables	Autistic Cases (no= 30)	
Age (Years) Range	4- 7	
Mean ± Sd	5.5 ± 1.2	
Sex	Number	Percentage
Male	26	86.7%
Female	4	13.3%

According to CARS scores, the autistic cases were classified as mild autism, moderate autism and severe autism.

Table (2) Classification of the autistic children according to CARS scores

Degree Of Autism	Range Of Scores	Number (30)	Percentage
Mild Cases	30- 33	5	16.7%
Moderate Cases	34- 36	12	40%
Severe Cases	>36	13	43.3%

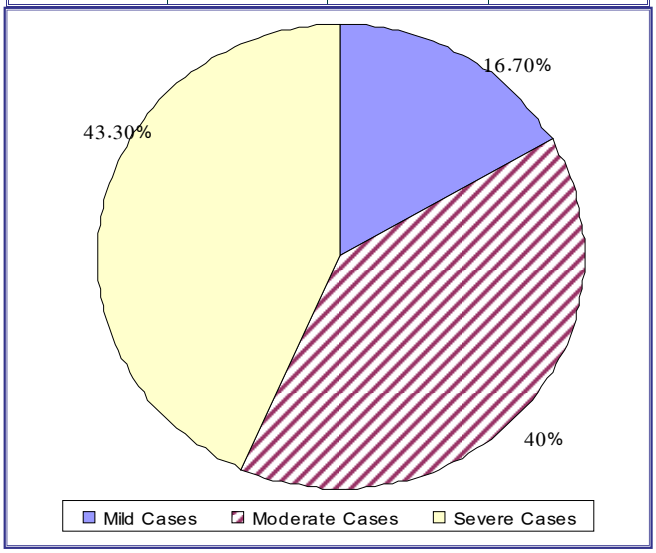

Figure (1) Classification of the autistic children according to CARS scores.

By comparing the domains of the ADI- R according to (CARS), it was found that social domain was more affected in the severe cases with significantly statistical difference. While the non verbal domain was more affected in the moderate cases. And the verbal domain was more affected in the mild cases with no significant statistical difference. Finally the repetitive behavior was more affected in the severe cases of autism.

Table (3) Comparison between ADI- R domains& the severity of autism

CARS		Mean	SD	F	P- Value	Sig.
Social Domain	Mild	12.6	2.5	5.43	0.028	S
	Moderate	16.7	4.3			
	Severe	17.9	4.8			
Non Verbal Domain	Mild	8.2	3.1	0.78	0.470	NS
	Moderate	9.6	2.6			
	Severe	8.2	3.1			
Verbal Domain	Mild	8.5	0.7	0.33	0.729	NS
	Moderate	7.5	0.7			
	Severe	8.2	1.5			
Repititive Domain	Mild	5.8	1.9	1.35	0.277	NS
	Moderate	6.8	1.9			
	Severe	7.5	2.3			

Comparison between expressive and receptive language age among the autistic cases showed that the receptive language age was more affected than the expressive language with no significant statistical difference.

Table (4) Comparison between the receptive& expressive language age of the studied group

		Mean	SD	Range		t	P Value	Sig.
				Min.	Max.			
Language	Receptive	2.1	1.0	1.0	5.5	0.66	0.513	NS
	Expressive	2.0	0.8	0.9	4.6			

By comparing the receptive& expressive language age and ADI- R domains, it was found that the receptive language had the highest mean value in the repititive domain while the expressive language had equal mean values in all domains.

Table (5) Comparison between language parameters and ADI- R domains

	ADI- R	Mean	SD	t	P- Value
Social Domain	Receptive	2.0	1.0	0.57	0.574
	Expressive	1.9	0.8	0.23	0.817
Non Verbal Comm. Domain	Receptive	2.0	1.04	0.75	0.461
	Expressive	1.9	0.8	0.18	0.857
Verbal Comm. Domain	Receptive	2.0	0.9	0.19	0.852
	Expressive	1.9	0.7	0.01	0.995
Repititive Domain	Receptive	2.1	1.0	0.21	0.884
	Expressive	1.9	0.8	0.20	0.732

Attention levels were assessed in the autistic cases by using (Developmental Programme for Children with Early Learning Handicaps, Cooper et.al). Our results showed the attention in most of the autistic cases was moderately to severely affected.

Table (6) Prevalence of attention grades among the studied group

Attention	Frequency	Percentage
Grade I	2	6.7%
Grade II	15	50.0%
Grade III	11	36.7%
Grade IV	1	3.3%
Grade V	1	3.3%

Comparison between attention levels and CARS scores showed that there was no significant statistical difference between severity of autism scores and levels of attention.

Table (7) Comparison between attention levels and CARS scores among the studied cases

Attention		Mean	SD	Median	Range		F	PValue	Sig.
					Min.	Max.			
CARS	Gr. 1	36.3	0.4	1.21	36.0	36.5	0.29	0.884	NS
	Gr. 2	35.9	3.3	1.54	30	42			
	Gr. 3	35.5	2.2	2.35	31.5	39.5			
	Gr. 4	38.0	-	3.40	34	38			
	Gr. 5	34.0	-	5.13	32.5	34			

Discussion:

This study's objective was to assess 15q11- 13& 16p11.2 CNV in autistic children in order to explore the possible influence of these variations on the phenotype of ASD and its severity.

According to the results of our study, no CNV were detected at 15q11- 13& 16p11.2 regions among the cases. A possible explanation for our results is that currently, it is believed that hundreds of loci are associated with ASD and this complexity makes it difficult to identify singular potential causative pathway and consequently a therapeutic approach. Additionally, it might have to do with the hundreds of genes that are involved in the complex interplay between heredity and environmental factors influenced by epigenetics, as well as the multifactorial cause of

Introduction:

Autism spectrum disorder ASD) is a neurodevelopmental condition marked by limitations in social communication and interaction as well as confined repetitive activities and behaviors in which one in every 54 children is affected by it. (Maenner et.al, 2021). The causative factors and the pathogenesis of autism are complex and involve the interaction between genetic, epigenetic, and environmental factors (Bhandari et.al, 2020). This is demonstrated by the heterogeneity in the biochemical and clinical phenotypes associated with ASD, which show that environmental risk factors can play a role in causing long- term dysfunction in those who are susceptible Emberti Gialloreti et.al 2019). According to Hyman et.al. (2020), microarray studies have demonstrated that a subset of persons with ASD have an excess of extremely large copy number variations (CNVs), which are pathogenic in 5.4% to 14% of cases. CNVs at many chromosomal regions such as 15q11-13, 16p11 have been identified as genetic risk factors for ASD and have demonstrated to have predictive value for clinical phenotype of ASD (D'Abate et.al, 2019). About 1% of people with ASD had microdeletions and microduplications at the 16p11.2 region, which is the most prevalent recurrent CNVs linked to ASD (Chung et.al, 2021). 71% of 16p11.2 deletions happen de novo, whereas 70% of duplications are familial. (D'Angelo et.al, 2016). The 16p11.2 gene locus contains 27-29 genes which are critical for neurodevelopment and may contribute to susceptibility of ASD (Iyer et.al, 2018). The architecturally complicated 15q11- q13 regions and their abnormalities are linked to a number of neuropsychiatric diseases, including ASD (Zhuo Fu et.al, 2021). The 15q11-13 region contains many significant genes, such as GABRA5, GABRB3, CHRNA7 and UBE3A which are essential for neurodevelopment and whose functions can be abnormally altered to cause autism and autism related NDDs. (Özaltun et.al, 2021). Most of children with ASD manifest with language deficits. They can present with difficulties in both receptive and expressive language skills. (Tafaroji& Kamari, 2020). These children have impairments in conversational skills, difficulties in initiating dialogues and in responding during communication, and interaction. (Morsanyi& Stamenković, 2021). Defects in attention is one of the prominent deficits of ASD and attention impairment is thought to be a primary contributing factor to the core symptoms in children with ASD (Ridderinkhof et.al, 2020). Disengagement, difficulties maintaining attention, and deficits in executive functions are among the attention problems frequently observed in ASD. (Craig et.al, 2016)

This study aimed to detect copy number variations in 15q11- 13 and 16p11.2 regions and how they could likely play a role in modifying and affecting language development and attention in these children. This will lead to appropriate early screening, medical and neuropsychiatric support for the parents and their children.

Methodology

Subjects:

This study was carried out in the outpatient clinic for "Autistic Disorders", Medical Research Center of Excellence, at the National Research Center in Egypt. The study was carried out on 30 autistic children who were diagnosed using the Autism Diagnostic Interview Revised (ADIR) and DSM- 5 criteria. The severity of the cases was assessed by (CARS) and their language was also assessed by using The Arabic Preschool Language Scale. (El- Sady et.al., 2011)

Procedures

1. Evaluation of 15q11- 13& 16p11.2 CNV using MLPA (Multiplex Ligation- dependent Probe Amplification): (Eijk- Van& Schouten, 2011).
2. DNA extraction was done using a Paxgene DNA extraction kit in accordance with the manufacturer's instructions from 3 ml of peripheral blood lymphocytes that had been collected on a Paxgene tube from the cases and the reference samples.
3. The NanoDrop spectrophotometer was used to assess the DNA samples' quality and quantity.
4. As directed by the manufacturer (MRC- Holland), the SALSA MLPA probemix P343- C3 Autism- 1 was used to evaluate the copy number variations of 15q11- 13 and 16p11.2.
5. On the first day, the MLPA probemix was denaturated by DNA and hybridized overnight. The following day, probe ligation and amplification were performed (Schouten et.al, 2002).
6. ABI 3500 Genetic Analyzer (USA) was used to separate amplified products. The MRC- Holland software Coffalyser. Net was used to interpret the data.
7. Ratios between 0.75 and 1.30, less than 0.75, were regarded as deletions.

Ethical Aspect

Ethical consideration in accordance with guidelines provided by the National Research Center's Ethical Committee and the University of Postgraduate Childhood Studies' Research Ethics Committee (IPGCS, 2014). Parents gave their informed consent after being made aware of the study's purpose and the confidentiality of all patient data.

Statistical Analysis

Version 12 of the statistical software for social science (SPSS) was used to examine the data that had been gathered. For all significant tests, the cut- off point was set at the level of significance at ($p < 0.05$).

Results:

Evaluation of 15q11- 13& 16p11.2 CNV by using MLPA (Multiplex Ligation- dependent Probe Amplification) revealed that no copy number variation (deletion or duplication) was detected in the 15q11- 13 and 16p11.2 regions among the studied cases.

The age range of the autistic cases was between 4- 7 years old. Their mean value of age was 5.5 years ± 1.2. They were 26 males (86.7%) and 4 females (13.3%).

Copy number variants in chromosomes 15& 16 and its contribution to phenotypic aspects in autistic children.

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Summary

Introduction: Autism Spectrum Disorder (ASD) is a neurodevelopmental disorder characterized by deficits in: social interaction; communication abilities, and limited, repetitive, and stereotyped patterns of behavior and interests. ASD results from complex interaction between genetic and non- genetic influences that mostly act through epigenetic regulation either alone or together and contribute to its development. Evidence provides strong support for the genetic contribution to ASD with many genomic loci as 15q11- 13 and 16p11 and the genes implicated in them. Copy number variation (CNV) is a type of structural variation that refers to variation in the number of copies of particular DNA sequences that can be either duplicated or deleted. Copy number variations (CNV) in the 16p11.2 and 15q11- 13 regions are associated with ASD. Our study aimed to assess copy number variation in 15q11- 13& 16p11 and exploration of their influence on language and attention in these children.

Methodology: Thirty autistic children were diagnosed with autism according to the Diagnostic& Statistical Manual of Mental Disorders, 5th edition (DSM- 5) criteria and the Autism diagnostic interview revised (ADIR) and the severity was assessed by using (CARS). Also their language was assessed using the Arabic Preschool Language Scale. Evaluation of 15q11- 13& 16p11.2 CNV was done using MLPA (Multiplex Ligation-dependent Probe Amplification).

Results: Our results revealed that there was no copy number variation CNV (deletion or duplication) detected in the 15q11- 13 and 16p11 regions among the studied cases. Assessment of the language parameters among the autistic children showed that the receptive language was more affected than the expressive language and the attention levels were severely affected.

Conclusion: ASD has multifactorial causation that is influenced by interaction between genetics and environmental factors together with other susceptibility variants that affect gene expression. Detection of such effects will lead to appropriate medical and neuropsychiatric support for such children.

اختلاف عدد النسخ فى كروموسومات ١٥ و١٦ ومدى تأثيرها على الجوانب المظهرية فى الاطفال المصابين بالتوحد

المقدمة: يعد اضطراب طيف التوحد من اضطرابات النمو العصبي الذى يتميز بضعف في: التفاعل الاجتماعي؛ مهارات التواصل؛ وأنماط السلوك والاهتمامات المقيدة المتكررة والنمطية. ينتج اضطراب طيف التوحد عن تفاعل معقد بين علم الوراثة والبيئة من خلال التنظيم اللاجيني الذى يساهم فى تطوره. توفر الأدلة دعما قويا للمساهمة الجينية في مرض التوحد من خلال العديد من المواقع الجينية بما في ذلك ١١p١٦ و١١q١٥– ١٣ والجينات المحمولة على هذه المواقع. يعد تباين عدد النسخ نوع من المتغيرات الهيكلية التي تتضمن تغييرات في عدد نسخ تسلسلات الحمض النووي المحددة التي يمكن حذفها و/أو تكرارها. يرتبط عدد تباين النسخ فى المنطقة ١١p١٦ و١١q١٥– ١٣ ارتباطا وثيقا بمرض التوحد. الهدف من هذه الدراسة هو تقييم تباين عدد النسخ فى ١١q١٥– ١٣ و١١p١٦ لدى الأطفال المصابين بالتوحد لتحديد مدى تأثير هذه التغيرات على اللغة والانتباة لدي هؤلاء المصابين. شملت الدراسة ٣٠ طفلا مصابا بالتوحد تم تشخيصهم وفقا لمعايير التوحد المحددة في الدليل التشخيصي والإحصائي للاضطرابات النفسية الإصدار الخامس (DSM5) واجراء المقابلة التشخيصية المنقحة لمرض التوحد (ADIR) وتم تقييم شدة المرض باستخدام مقياس تقييم التوحد في مرحلة الطفولة CARS. وتم تقييم لغتهم باستخدام مقياس اللغة العربية لمرحلة ما قبل المدرسة. تم تقييم تباين عدد النسخ فى ١١p١٦ و١١q١٥– ١٣ باستخدام تقنية تضخيم المجسات التعددى المعتمد على الربط (MLPA).

النتائج: كشفت نتائجنا أنه لم يتم اكتشاف أي اختلاف فى تباين عدد النسخ (حذف او تكرار) فى المنطقتين ١١q١٥– ١٣ و١١p١٦ لدى اطفال التوحد. وأظهر التقييم اللغوى أن اللغة الاستقبالية كانت أكثر تأثرا من اللغة التعبيرية بين الحالات.

الاستنتاج: ينتج اضطراب طيف التوحد عن اسباب متعددة العوامل والتى تتأثر بالتفاعل بين العوامل الوراثية والعوامل البيئية بالإضافة إلى المتغيرات القابلية الأخرى التي تؤثر على التعبير الجيني. لذلك فان اكتشاف مثل هذه التأثيرات سوف يؤدى إلى توفير الدعم الطبي والنفسي العصبي المناسب لهؤلاء الأطفال.

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