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Brief Article

Screening autism spectrum disorder in adults with Down syndrome: preliminary findings

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SUMMARY

Objectives

In recent years, several studies have highlighted the presence of autism spectrum disorder in individuals with other neurodevelopmental disorders. The objective of this preliminary study is to detect the presence of autistic traits in a sample of adults with Down syndrome.

Methods

31 adults with Down syndrome participated in the study. An evaluation of the Intelligence Quotient and a screening of the presence of autistic traits through standardized instruments were done.

Results

The obtained Intelligent Quotient scores confirm the presence of intellectual disability.
The preliminary results show the presence of autistic traits in our sample. In addition, we collected additional evidence on those who show familiarity for psychiatric or neurological disorders.

Conclusions

It is necessary to proceed to a formal screening of the presence of autism in Down syndrome and in other neurodevelopmental disorders.

Key words: autism spectrum disorder, Down syndrome, intellectual disability, developmental disabilities

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Introduction

Neurodevelopmental disorders consist of a wide range of disorders, evident from early childhood that last for lifetime. Neurodevelopment disorders, which often occur in comorbidity, include communication, tic and motor disorders, specific learning disorder, Attention-Deficit Hyperactivity Disorder (ADHD) as well as Autism Spectrum Disorder (ASD) and Intellectual Disability (ID) [1].

Down Syndrome (DS) is a genetic disorder arising from a chromosome defect involving chromosome 21 (also called trisomy-21) and is one of the most common genetic causes of ID [2]. Individuals with DS often show a cognitive decline associated with ageing characterized by a deterioration in memory, language and cognitive functioning. Individuals with DS show typical organization of brain structures related to some cognitive abilities, such as reduced volume in frontal and prefrontal areas, which is related to poor executive and linguistic abilities. They also frequently show psychiatric disorders such as externalizing disorders as well as depression,

anxiety and obsessive-compulsive disorder. Nevertheless, as for other genetic syndrome with intellectual disability, there is a significant lack of research specifically focused on treatments of psychiatric and behavioural problems in DS [3].

ASD is a neurodevelopmental disorder with early onset in childhood diagnosed on the basis of persistent deficits in social communication and social interaction across multiple contexts (deficits in social-emotional reciprocity, in nonverbal communicative behaviors used for social interaction, and in developing, maintaining, and understand relationships) and restricted, repetitive patterns of behavior, interests, or activities and/or hyper- or hyporeactivity to sensory input or unusual interest in sensory aspects of the environment [1]. In a study population in ASD, ID was found in 53%, which was mild in 35.8% of cases, moderate in 34%, and severe in 30.2% [4].

In recent years, many studies have reported the presence of autistic traits in people with DS [5-7].

ASD prevalence in general population is 1.85% [8]. In people with DS, the prevalence of ASD varies from 1 to 19% depending on the studies [5,6].

All considered, the main purpose of the present study is to assess autistic traits and behaviors in a sample of DS adults.

Methods

Participants

Participants were recruited from Transitional Care Clinic of Città della Salute e della Scienza Hospital. Inclusion criteria were age greater than 18 years and diagnosis of DS. People with other genetic conditions were excluded from the sample. An informed consensus was collected from the patient or the legal guardian. The privacy rights have been observed.

31 people with DS were included in the study. Mean age was 29.77 (± 6.14) years, minimum age 19, maximum age 43. Mean years of schooling 11.70 (± 2.2; min 8-max 13,); 16 participants were females (51.61%) and 15 were males (48.39%).

Demographic characteristics are summarized in Table I.

38.7% (n = 12) of our sample had at least one person with a psychiatric or neurological disorder in their family. These include neurological conditions such as epilepsy, neurodegenerative diseases (e.g., multiple sclerosis, amyotrophic lateral sclerosis, prion disease, unspecified dementia, colorblindness) or psychiatric disorders (e.g., bipolar, depression).

Specifically, 6.45% (n = 2) had familiarity in uncles, 12.9% (n = 4) in parents, 12.9% in grandparents (n = 4), and 6.45% in both parents and grandparents (n = 2).

In contrast, 61.29% (n = 19) showed no familiarity for these disorders.

Instruments

We administered the Wechsler Adult Intelligence Scale - Fourth Edition; WAIS-IV scale [9] to evaluate the Full Scale IQ (FSIQ).

In addition, we calculated Verbal Comprehension (VCI), Perceptual Reasoning (PRI), Working Memory (WMI) and Processing Speed (PSI) Indices.

We used the Italian version of WAIS-IV [10] to calculate FSIQ score, VCI, PRI, WMI, PSI comparing our sample' scores to standardization samples'.

We also use the Italian version of Scale of Pervasive Developmental Disorder in Mentally Retarded Persons (PDD-MRS in Italian STA-DI: Scala di valutazione dei Tratti Autistici nelle persone con Disabilità Intellettiva) [11,12]: this is a screening instrument useful to guide clinician to identify autistic traits in people with intellectual disability. The interviews were administered directly to the subject or to the caregiver.

Both IQ and PDD-MRS/STA-DI scales have been administered by a psychologist with expertise in both ASD and cognitive and neuropsychological assessment in neurodevelopmental disorders.

Results

Full Scale IQ and indices

The administration of the WAIS- IV scale showed the following results. For the whole sample the minimum score on the Full IQ scale was 32, while the maximum was 74 with an average score of 37.58 (± 8.46).

VCI, PRI, MMI and PSI scores are summarized in the table below (Tab. I).

Autism

12.09% of the total sample obtain scores indicative of the presence of autistic traits. However, 9.67% of the sample obtain borderline scores that require an additional clinical and diagnostic study. Considering borderline and positive scores, therefore, 22.58% of our sample shows autistic traits. PDD-MRS/STA-DI scores for all participants and according to age and gender are summarized in Table I.

Further analysis

We first conducted a t-test to detect any gender and age differences in the WAIS- IV Full Scale IQ or Indices and in the score at PDD-MRS/STA-DI. The results show us that there are no gender or age differences.

Then, we conducted a t-test to see if there were any differences related to psychiatric and neurological familiarity. The results show us that there were no significant differences in the scores obtained on the Full Scale IQ and Indices on WAIS-IV.

The results, on the other hand, show a statistically significant difference regarding the scores obtained on the

TABLE I. *Demographic characteristics, Full Scale IQ (FSIQ), Verbal Comprehension (VCI), Perceptual Reasoning (PRI), Working Memory (WMI) and Processing Speed (PSI) Indices and PDD-MRS/STA-DI scores according to gender, to age groups and for all participants. For PDD-MRS/STA-DI scores percentages are also indicated.*

	AGE	Y. SCHOOLING	FSIQ	VCI	PRI
	Min-max Mean (± SD)	Min-max Mean (± SD)	Min-max Mean (± SD)	Min-max Mean (± SD)	Min-max Mean (± SD)
Females (n = 16)	20-41 31.06 (± 6.47)	8-13 11.43 (± 2.39)	32-58 37.31 (± 6.49)	47-78 51.689 (± 7.83)	44-65 50.18 (± 6.70)
Males (n = 15)	19-41 28.4 (± 5.66)	8-13 12 (± 2.07)	32-74 37.86 (± 10.39)	47-100 52.26 (± 13.36)	44-65 50.13 (± 6.05)
19-30 years old group (n = 16)	19-29 24.87 (± 2.96)	8-13 12.68 (± 1.25)	32-74 37.18 (± 10.41)	47-100 51.43 (± 13.10)	44-65 48.43 (± 5.85)
31-41 years old group (n = 15)	31-41 35 (± 3.85)	8-13 10.66 (± 2.8)	33-58 38 (± 6.08)	47-78 52.53 (± 7.73)	45-65 52 (± 6.41)
All partecipants (n = 31)	19-43 29.77 (± 6.14)	8-13 11.70 (± 2.22)	32-74 37.58 (± 8.46)	47-100 51.97 (± 10.68)	44-65 50.16 (± 6.29)

PDD-MRS/STA-DI. In particular, subjects with psychiatric or neurological familiarity (n = 12, mean = 8.17, SD = 4.01) show significantly higher scores than those without familiarity (n =19, mean = 4.21, SD = 1.75), t = -3.78, p < .001.
We conducted an ANOVA to detect any differences in the scores obtained at the various scales by comparing them by degree of familiarity. The results show us that there are significant differences in PDD-MRS/STA-DI. In particular, subjects whose parents showed psychiatric or neurological pathologies (n = 4, mean = 12.25, SD = 2.17) obtained significantly higher scores, F = 13.80, p < 001.
In addition, we wanted to investigate the relationship between Full Scale IQ and Indices of the WAIS-IV and the scores on the PDD-MRS/STA-DI scale: to do so, we created a dichotomous (0 *vs* 1) index that grouped scores below the PDD-MRS/STA-DI cut-off (<= 6) on one side and borderline or positive scores (>= 7) on the other.
T-test scores show us that there are significant differences exclusively for the Perceptual Reasoning (PRI) Index. In particular, subjects who expressed borderline or positive scores obtained significantly lower PRI scores (n = 7) (mean = 46, SD = 1.63) t = 2.09, p < 05.
Despite the absence of significance, we attempted to calculate the effect size to measure the strength of the relationship between the variables.
The results show us that for both the FSIQ, PRI and PSI subscale, Cohen's d expresses a large effect size.
Cohen's d for the VCI and WMI indices expresses a mean score (see Table II).

Discussion

Our study underlines the need of a multi-level analysis in DS and, generally, in the neurodevelopmental disorders. Neurodevelopmental disorder should be considered as an unique spectrum and co-occurrence with ASD, ID, ADHD, communication, tic and motor disorders and specific learning disorder should drive clinicians to look to this disorders with a wide range of investigations [13].
Medical diseases (such as epilepsy, thyroid and cardiac diseases) and psychological or psychiatric disorders (such as mood disorders and cognitive deficits similar to Alzheimer disease) have been shown in adults with DS [14-16]. However the life expectancy of people with DS has increased in the last decades [14].
Our study confirms the presence of intellectual disability in DS but, at the same time, highlights the presence of

TABLE II. *Cohen's d for WAIS-IV Full Scale QI and Indices. * dichotomous (0 vs 1) indicate scores below the PDD-MRS/STA-DI cut-off and borderline or positive scores.*

	PDD-MRS/ STA-DI *	N	Mean	SD	Cohen's d
FSIQ	0	24	38.79	9.25	**0.80**
	1	7	33.43	2.07	
VCI	0	24	53.25	11.87	0.66
	1	7	47.57	0.97	
PRI	0	24	51.38	6.64	**1**
	1	7	46	1.63	
WMI	0	24	52.13	6.68	0.66
	1	7	49	0	
PSI	0	24	53.92	6.36	**0.87**
	1	7	50	0	

WMI	PSI	PDD-MRS/STA-DI				
Min-max Mean (± SD)	Min-max Mean (±S D)	% Negative (n)	% Borderline (n)	% Positive (n)	% Borderline + positive (n)	Total Score Min-max Mean (± SD)
49-60 51.56 (± 3.52)	50-67 53.13 (± 5.79)	68.75% (11)	12.5% (2)	18.75% (3)	31.25% (5)	1-14 6.75 (± 3.69)
49-80 51.26 (± 7.98)	50-72 52.73 (± 6.02)	86.66% (13)	6.66% (1)	6.66% (1)	13.33% (2)	1-12 4.67 (± 2.79)
49-80 52.37 (± 7.97)	50-72 53.06 (± 6.23)	75% (12)	12.5% (2)	12.5% (2)	25% (4)	1-14 5.56(± 3.63)
49-57 50.4 (± 2.61)	50-67 53 (± 5.55)	80% (12)	6.66% (1)	13.33 (2)	20% (3)	1-13 5.93 (± 3.26)
49-80 51.42 (± 5.99)	50-72 53.03 (± 5.81)	77.4% (24)	9.7% (3)	12.9% (4)	22.58% (7)	1-14 5.74 (± 3.40)

autistic traits in DS population that deserve further analysis by specialized and multidisciplinary teams, as with multistep-multinetwork model [4]. Furthermore, it seems that the comorbidity between DS and ASD leads to the manifestation of a greater number of behavioral problems such as stereotypic, compulsive or self-injurious behaviors (see [17] for a summary) or aggressive behavior [15]. In adults, DS may show a cognitive deficit not only related to ID but also to cognitive impairment, that should be detected. We suggest that, in DS, ASD should be considered as a co-occurrence and detected, too.

Screening test, as PDD-MRS/STA-DI, should be used routinely in visiting DS people. If possible, a complete neuropsychological evaluation should be used in this population.

If screening of ASD is positive in DS, a complete evaluation for ASD should be used, with Autism diagnostic interview-revised- ADI-r [18] and Childhood Autism Rating Scale-CARS-st [19].

In clinical practice, clinicians should be careful to consider the high co-occurrence among neurodevelopmental disorder. This study underlies the high prevalence of ASD in DS. This result goes against the common thoughts that describe DS population as very friendly and not prone to ASD social deficits. A good clinical guideline for clinicians visiting a patient with a neurodevelopmental disorder should be to carefully explore all neurodevelopmental disorders to detect a possible co-occurrence using a systematized screening with testing.

Conclusions

Recognizing ASD traits in DS, or ASD with complete syndrome could be useful to better understand challenging behavior in DS linked to autism functioning, as hyper-sensoriality or sameness research or routinary behavior related to autism functioning.

If ASD is confirmed, a cognitive-behavioral treatments should be considered to improve ASD symptoms in DS.

The major limitation of the present study is the sample size; however, the data obtained can be useful for defining further research protocols and guiding clinical practice. Future research and clinical practice should detect ASD among other neurodevelopmental disorder as intellectual disability, ADHD, Specific learning disabilities and especially in genetic syndromes, as Down's syndrome, that could drive to ASD misdiagnosis; so a specific ASD screening should be used in clinical practice in all Neurodevelopmental disorder. On the base of high co-occurrence among neurodevelopmental disorder, in the specialized visit for ADHD, or ASD or ID, all neurodevelopmental disorder should be carefully screened.

Ethical consideration

None.

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Conflict of interest

The Authors declare no conflict of interest.

Author contributions

The Authors contributed equally to the work.

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