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Evolutionary Trajectories in Children with Autism Spectrum Disorder

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Abstract: Autism spectrum disorders are generally regarded as lifelong conditions, affecting communication, relationships, and adaptive skills. Studies on the developmental trajectories of people out of autism have found adequate adaptive social-communication skills, effective experiences of inclusion in regular education classrooms, normal intellective functioning, and an absence of typical autism symptomatology. It therefore seems plausible to start reading the 'after autism' psychopathological conditions in a continuum that features several possible clinical and non-clinical phenotypes. The present retrospective research aimed to examine the different developmental trajectories of 17 children with an original diagnosis of autism, evaluated in a follow-up approximately 5 years after the end of the therapy. The stability of the optimal outcomes is evidenced by the absence of clinical diagnostic criteria for autism spectrum disorder. However, some difficulties persisted in adaptive functioning, especially in the social domain, consistent with the dysfunctional core that characterized the clinical features of autism in childhood. Furthermore, many of the participants showed residual relational atypia, such as alterations in pragmatic communication, or a psycho-affective disorder, or specific developmental disorders. The presence of some residual atypia provides important food for thought, not only in orienting any therapy with which continue to support older children, but also for a greater understanding of the pathological core towards which has evolved the original diagnosis of autism.

Keywords: Autism Spectrum Disorders, Developmental Trajectories, Follow up, Adaptive Functioning, Atypia

1. Introduction

Autism spectrum disorders (ASDs) are generally regarded as lifelong conditions, affecting communication, relationships, adaptive skills, academic and vocational attainment [1]. However, recent research indicates that a percentage ranging from 3% to 25%, depending on the study, several years after the original diagnosis no longer fulfil the diagnostic criteria of autism [2-6]. In a follow-up study, Kelley, Naigles and Fein [7] examined a sample of children with optimal outcomes aged 8-13 years and found that these were comparable to those with the typical development group on all language measures and showed psychiatric vulnerability only in attention regulation. However, the authors observed a residual lack in the theory of mind and in constructing narratives [8]. Also, other authors who have dealt with younger children (from 5 to 9 years old) with optimal outcomes, found residual pragmatic and semantic language deficits, while grammatical skills were intact [8-10]. Other studies on the developmental trajectories of people with an original diagnosis of autism have found adequate adaptive social-communication skills, effective experiences of inclusion in regular education classrooms [2], normal intellective functioning and an absence of typical autism symptomatology [5].

Dell' Osso & Lorenzi [11], starting from a dimensional approach of psychiatric disorder, outlined a framework that allows us to bring together 'before and after' autism, defining the concept of sub-threshold autistic traits (ATs) or broad autism phenotype. According to the authors, it is possible to hypothesize the existence of a vulnerability factor that

represents the starting point for the development of different psychopathological trajectories not preordained but shaped by interaction with the environment and lifetime events [12]. It therefore seems plausible to start reading the 'after autism' psychopathological conditions in a continuum that features several possible clinical and non-clinical phenotypes.

There are several authors who have dealt with the study of residual psychopathology of children with an original diagnosis of autism. A parent interview for psychiatric disorders of youth aged 8-21 years old [13] showed that those who no longer met autism criteria, as well as high functioning autism, had elevated attention problems with or without hyperactivity, social or specific phobias and tics. However, in the high functioning group, these symptoms did not significantly reduce over time, as was the case in patients with optimal outcomes.

Another line of research studied the persistence of deficits in adaptive functioning in people with an original diagnosis of autism. Some studies [14, 15] show the presence of deficit in the domain of socialization and communication and relative strength in the domain of daily living. Kanne et al. [16] states that the greatest discrepancy is observed between socialization skills and intellectual functioning, indicating that, despite having good cognitive abilities, these people had difficulty in using their strengths in a functional way in everyday contexts, particularly in adaptive-communicative and adaptive-social areas. However, in other studies [2, 17], the adaptive skills of children with a previous diagnosis of autism did not significantly differ from those of typically developing children, in terms of socialization or communication.

The results confirm the heterogeneous nature of developmental trajectories in autism. These trajectories and the possible psychopathology of 'after autism' are still much debated topics and require a clinical evaluation that can go beyond the categorical diagnosis to circumscribe the moments of transition from one state to another and place them in a new neuro- and psycho-developmental framework. In this sense, the present retrospective research aimed to examine the different developmental trajectories of children with an original diagnosis of autism, evaluated in a follow-up approximately 5 years after the end of the therapy.

The specific objectives of this study were 1) to verify the stability of the results achieved during therapy, 5 years after the end of the therapeutic path, through the administration of Module 3 of ADOS-2 and a clinical interview; 2) to check for psychopathological symptoms; 3) to verify the quality of socio-adaptive skills.

2. Methods

2.1. Participants

The present study involved 17 children and adolescents aged 9 to 17 (mean=13.57; sd=1.97), who received a diagnosis of autism spectrum disorder (ASD) between 2.5 and 6.8 years of life (mean=3.93; sd=1.09). All underwent a therapeutic path for at least four years at the Institute of Ortofonologia, at the end of which the diagnostic criteria for autism classification were no longer present. After about 5 years (mean=5.75; sd=1.17) from the end of the therapeutic path, a follow-up was proposed to verify the symptomatologic condition through clinical interviews and the administration of standardised tests.

The study group consists of 6 females (35%) and 11 males (65%); the mean age of the mother was 44.62 years (sd=5.03) and of the father was 50.40 years (sd=4.43); 23% of mothers and 6% of fathers had a university degree, while 77% of mothers and 82% of fathers had a secondary school diploma; finally, 12% of fathers had a first-degree secondary school license. All families were of Italian origin.

Children with significant medical problems, or with sensory or motor impairments, were excluded from the sample.

2.2. Procedures

All participants in the research were diagnosed from a team of highly experienced clinicians including psychologists/psychotherapists, child neuropsychiatrists and neurologists. For children evaluated between 2013 and 2016, the diagnosis of autism was based on the DSM-5 criteria [18], for which, in addition to clinical observations, the children were given the Autism Diagnostic Observation Schedule-second edition (ADOS-2) [19]. After receiving the diagnosis, all the children were included in a developmental therapy program for ASD [20, 21]. At the end of the therapeutic path, among the children who no longer fell within the diagnostic classification of spectrum disorder, 17 were monitored over time, and on average after 5 years a direct assessment was proposed to identify autism spectrum disorder (clinical interview, observation, and ADOS-2) and an indirect assessment, through a questionnaire filled in by parents for the measurement of social and adaptive skills (ABAS-II) [22].

Informed consent was obtained from all parents (Declaration of Helsinki). This research complied with the ethical guidelines and legal requirements of the country in which it was conducted. The study also adhered to the ethical standards of the American Psychiatric Association.

2.2.1. Measures

The Autism Diagnostic Observation Schedule-Generic (ADOS-G) [23] is a semi structured, standardized assessment of social interaction, communication, play and imaginative use of materials for individuals suspected of having autism spectrum disorders. The total score defines three diagnostic categories: Absence of autism (ADOS score between 0 and 6); Autism spectrum (ADOS score between 7 and 11); and Autism (ADOS score between 12 and 24). The reliability was assessed through the inter-rater agreement (0.92) and through the test-retest reliability (0.82).

ADOS-2 [19] allows for a standardized and semi-structured evaluation of the aspects of communication and social interaction (SA), restricted and repetitive behaviors (RRB)

and playful/imaginative use of material, involving a series of activities that directly elicit behaviors linked to the diagnosis of autism spectrum disorder.

It consists of several modules. Those used in this study were as follows:

The Toddler Module is used for children between 12 and 30 months of age who do not consistently use phrase speech. This module provides scores that describe different clinical risk ranges for autism (none or low risk: scores from 0 to 9; moderate risk: from 10 to 13; high risk: greater than 13) to allow the clinician to quantify and formalize a clinical impression and to avoid a formal classification that may not be appropriate in this age group.

Module 1 is administered to children aged 31 months and over who use little or no phrase speech. It consists of a series of structured activities aimed at investigating aspects related to the area of social affect and restricted and repetitive behaviors. Scores above 8 are indicative of autism spectrum disorder.

Module 2 is administered to children under 30 months of age who use phrase speech but are not verbally fluent. It consists of activities of imaginative play and joint interaction and conversation. Scores above 7 are indicative of autism spectrum disorder.

The Leiter International Performance Scale – Revised [24] was used to measure nonverbal IQ through nonverbal stimuli, which is useful in cases where subjects have verbal linguistic impairment. The IQ scores had a mean of 100 and a standard deviation of 15.

The Adaptive Behaviour Assessment System-II (ABAS-II) [22] assesses the level of adaptive functioning in children and adults. The parent report of the ABAS-II used in this study provided information in the skill areas of Communication, Community Use, Functional Academics, Home/School Living, Health and Safety, Leisure, Self-direction, Self-care, Social and Motor. Skill area scores are presented as norm-referenced scaled scores (M=10; sd=3) and are aggregated into three composite scores: Conceptual Adaptive Domain (CON; Communication, Functional Academics, Self-Direction), Social Adaptive Domain (SO; Leisure, Social) and Practical Adaptive Domain (PR; Community Use, Home/School Living, Health and Safety, Self-Care). A General Adaptive Composite (GAC) score is also calculated from all skill area scores. Composite scores are presented as norm-referenced standard scores (M=100; sd=15).

2.2.2. Statistics

To evaluate the changes in scores that children achieved in follow-up, analyses of variance for repeated measures were performed. The size of the effect was calculated using the partial eta squared. To analyze the changes over time of the measures based on categorical variables, a chi-squared analysis was conducted. Correlational analyses were carried out to evaluate the relationships between the scores obtained in various measures.

The significance level was set at $p<0.05$. All statistical analyses were performed using SPSS software version 21.0.

3. Results

3.1. Descriptive

Table 1 shows the scores obtained by the children in ADOS and in cognitive tests at the time of the original diagnosis (T0) and at the end of the therapeutic path (T1). From the beginning to the end of therapy, IQ scores improved significantly ($F1.16=18.94$; $p< .001$); in the same way, the transition from the ASD classification was also confirmed by a significant reduction in the ADOS scores that did not reach the clinical cut-off ($F1.16=171.86$; $p< .001$).

Table 1. *Descriptive characteristics of the participants.*

	T0	T1
IQ, mean (sd)	78.8±20.6	100.8±11.6
IQ, classification (%)		
Below average	41.2%	0%
Average	58.8%	84.1%
ADOS, mean (sd)	14.2±4.8	5.6±2.9
ADOS, classification		
Moderate	41%	
Severe	58%	100%
No-ASD	0%	

*Legend: T0=time of the original diagnosis; T1=End of the therapeutic path; IQ=Intelligence Quotient.

3.2. Developmental Trajectories

After about 5 years (mean=5.75; sd=1.17) from the end of the therapeutic path, the 17 participants were re-evaluated with Module 3 of ADOS-2, which includes an interview about social relationships, and through the ABAS-2 for adaptive skills.

Compared to the ADOS-2 Module 3 scores, the children/adolescents obtained average values below the clinical cut-off both in Social Affect (mean 3.71; sd=1.76) and in Restricted and Repetitive Behaviours (mean 0.24; sd=0.44), as well as in the Comparison Scores (mean 2.24; sd=0.83).

The scores concerning socio-adaptive behaviors were also analyzed, which showed that, even if the average scores of the adaptive areas investigated did not fall within the classification of 'extremely deficient' (composite and standard scores < 2 standard deviations from the norm), there were still some issues (see Table 2): the average GAC score indicated the presence of a borderline adaptive global functioning (-1.5 sd from average). Specifically, the Conceptual domain scores (CON) were at the low limit of the mean (between 1 and 1.5 ds from the mean); within this area, in about 17% of subjects, communication skills (CO) were severely impaired (question examples: Talk about their favorite activities; Speak clearly and distinctly, etc.). Home/School living skills (SCO) were still severely impaired in more than 35% of subjects (question examples: He/she reads and respects common symbols, for example 'Do not enter', ' Exit' or 'Stop' etc.). Finally, those deficient in the greatest number of children/teenagers (41%) were those about self-regulation behaviors (AC) (question examples: He/she suspends an activity, without complaining, when he/she is told that he/she must stop; Avoid lying because

he/she knows he/she will be punished etc.).

The scores of the Practical Adaptive Domain (PR) are the most adequate (±1 sd from the norm); within this area, only 6% of cases show severely impaired home living autonomy (VC) skills (question examples: Tidies up clothes; Helps with housework etc.).

About 29%, on the other hand, had serious impairments both in autonomy for extra-familiar environment (AM) (question examples: Looks both ways before crossing the street; Manages money for small purchases etc.) and in self-care autonomies (CUR) (question examples: Eats independently; Washes hands with soap etc.). Finally, about 23% had severe impairments in their ability to engage in personal protective behaviors (SS) (question examples: Respects basic rules of safety at home or outdoors; Tastes hot foods before eating them, etc.).

The Social domain scores (SO) appeared to be on average less adequate; in particular, the skills that were still severely impaired in more than 40% of the participants were those related to socialization (SOC) (for example, Has friends; Has good relationships with parents and other adults; Seeks the friendship of his/her peers), while those related to self-organization skills in free time and play (Leisure) are less severely impaired (for example, Reads during free time; Goes to play at another child/teenager's house, etc.).

Table 2. *Means (±sd) of the composite and standard ABAS-II scores and % of scores below the average.*

ABAS-II Domains/Subscales	Scaled Scores	Impairment Level Mild*	Severe**
GAC	78.6±19.2	35.3%	29.4%
CON	81.1±19.5	23.5%	29.4%
CO-scaled score	7.1±3.5	35.3%	17.6%
SCO-scaled score	7.1±4.6	17.6%	35.3%
AC-scaled score	5.9±3.9	17.6%	41.2%
DAP-composite	84.2±15.2	29.4%	17.6%
VC-scaled score	8.4±2.7	29.4%	5.9%
AM-scaled score	6.5±3.5	47.1%	29.4%
SS-scaled score	7.6±3.4	11.8%	23.5%
CUR-scaled score	7.4±3.2	17.6%	29.4%
DAS-composite	76.5±20.6	0%	52.9%
SO-scaled score	5.3±3.4	29.4%	41.2%
TL-scaled score	6.2±4.5	58.8%	5.9%

Legend: GAC: General Adaptive Composite; CON: Conceptual Adaptive Domain; CO=communication; SCO=functional academics; AC: self-control; PR: Practical Adaptive Domain; VC: home living; AM: community use; SS: Health and Safety; CUR=Self-care; DAS: Social Adaptive Domain; SO=socialization; TL=leisure.

Composite score: mean 100±15; Scaled score: 10±3.

*Score between 1 and 2 standard deviations from the mean: **score < 2 standard deviations from the mean.

From the analysis of the clinical data, it also emerged that, at follow-up, 11 participants (out of 17) had residual relational atypia, characterized by slight alterations in pragmatic communication or in the area of mentalization; of the other 6, one had a psycho-affective disorder, characterized by low mood; three children had specific developmental disorders, such as specific speech or motor coordination disorders; one child had a residual diagnosis of intellectual disability and one child had no specific disorder or atypia.

4. Discussion

Autism spectrum disorders represent a clinical and diagnostic domain characterized by extreme variability and complexity; in the face of the presumed impossibility of a cure related to function that is neuro-atypical, some studies [2, 25] report the possibility of optimal outcomes up to the absence of diagnostic criteria years after the first diagnosis. The stability of the optimal outcomes is confirmed in the sample of this study, evidenced by the absence of clinical diagnostic criteria for autism spectrum disorder and measured on average 5 years after the end of a therapeutic course of at least 4 years. However, we observed that some difficulties persisted in adaptive functioning, especially in the social domain, consistent with the dysfunctional core that characterized the clinical features of autism in childhood. However, it is important to underline that all the children evaluated at follow-up regularly attended school, without the support of an additional teacher, achieved a sufficient level of autonomy and, except for one child, showed an average intellectual level. None of them was using drugs or specific interventions for psychiatric comorbidities.

The therapeutic path that the participants followed (DERBBI) [21] is based on a model that works on the processes of attunement and communication, using the body-relational dimension. As therapy progressed, interventions were increasingly individualized based on the specific needs of the individual and the areas that still appeared dysfunctional. In this way, it was possible to redefine the unexpressed individual potential, which can emerge if the therapeutic intervention reflects the complexity of the systems and processes that characterize the developmental age, actively involving parents and especially in the initial stages, working on the sensory-perceptive motor organization, on intersubjectivity and on emotional regulation, intended as precursors of cognitive, linguistic and behavioral communication development.

In all cases, the starting ADOS scores were indicative of a

level of impairment from moderate to severe, but this did not prevent the achievement of a good outcome. The presence of some residual atypia, observed at follow up, provides important food for thought, not only in orienting any therapy with which continue to support older children, but also for a greater understanding of the pathological core towards which has evolved the original diagnosis of autism. From a therapeutic point of view, in fact, an intervention aimed at a child with alterations in pragmatic communication is, for example, very different from the one necessary for a child with a low mood. In one case, in fact, it is still necessary to work on metacognitive aspects of communication to achieve an expansion in the social-relational sphere, while in the second it is necessary an emotional re-signification for an improvement in social relationships.

A follow-up assessment, even after several years, allows the clinician to monitor the presence of generalized skills, which persist even when children and their family are no longer supported by a therapeutic intervention. This is because a therapeutic intervention should favor the internalization of the coping mechanisms necessary for the adaptation to environment and the integration of sensory aspects that the autistic pathology often 'dismantles', as Meltzer [26] argued, referring to that mechanism (dismantling) related to the difficulty to integrate, in a common image, the experience and construction of an emotional meaning (factors underlying both cognitive, emotional and relational development). For this to happen, it is necessary to work not only on dysfunctional behaviors, but on the pathological nuclei that keep them alive.

Thus, the main residual difficulties that emerged at the follow-up in the behavioral self-regulation, in self-care and in socialization, confirm the importance of planning interventions focused on autonomy and social adaptation, even before performance; these interventions represent the central fulcrum of a life project that contemplates and supports, in a perspective view, the child, the family and the school in different stages of development and of the disorder.

5. Conclusion

The long-term monitoring of developmental trajectories of subjects with an original diagnosis of autism spectrum disorder allows the clinician to verify the stability of results obtained during the therapeutic path. Even in the presence of severe impairments in social-cognitive, communicative and behavioral functioning, it is possible to achieve optimal outcomes. The intellectual and language levels at the time of diagnosis are not the only positive prognostic indicators, as one of the 17 children taken into consideration still had an IQ below the norm. This consideration assumes a central value in psychodiagnostics because it underlines the need to define the personological characteristics of each one; considering only the categorical diagnosis, we risk 'crushing' the individual in a disease trajectory [12], ignoring the potential that can give rise to alternative pathways. The real risk, in our opinion, is to consider in the same way the oppositional conducts, the mentalization deficits, the self or hetero-aggressive behaviors and the low mood as consequences of the same original autistic nucleus, thus not allowing for diversified and personalized interventions.

Such a small number of subjects obviously does not allow too many generalizations but favors a qualitative understanding of the phenomenology of the disorder in the 'post-autism' period. It's important that the outcomes must be evaluated in this perspective and it's hoped that this study will bring new data to these considerations, in order to follow both the 'illness trajectory' and the developmental path.

Ethics Approval and Consent to Participate

The research complied with the ethical guidelines and legal requirements of the country in which it was conducted and the ethical standards of the American Psychiatric Association (APA). The study was approved by the Internal Review Board (IRB) of the Institute of Ortofonologia in Rome. No IRB's reference number is available.

Conflict of Interest

The authors declare that they have no competing interests.

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