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Research Article

New Evolutionary Model on "Autistic Spectrum Disorder" (PEMA), the severity scale (PASS), and the clinical questionnaire for the late teenage years and adults (PTAS-Qa)

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Abstract

Starting from the concept of "Autism Spectrum Disorder", as framed in the DSM-V, the present research proposes a new etiological theory (PETA) focused on the evolutionary model of the neurobiological matrix, overcoming the modest or inconsistent psychodynamic or conspiracy theses present in the international scene. Based on this assumption, therefore, a new study model (PEMA) was suggested that can focus on the presence or absence of 9 dysfunctional traits (type-deficit and type-clinical features) centered on the areas of social, emotional, and cognitive-communication and interaction, grading the impairment according to a precise 5-level severity scale (PASS), underpinning the 40-item questionnaire (*Perrotta Dysfunctional Traits of the Autistic Spectrum Questionnaire for the late teenage years and Adults, PTAS-Qa*) on a binary Y/N scale and based on a total score of 11/11, thus offering a more structured view of the patient's cognitive and psychological organization, avoiding diagnostic fragmentations that might be inconsistent with the nosographic picture and therefore not perfectly framed. By exemplifying the category, it is, therefore, possible to more easily frame the patient, who would thus be analyzed from a strictly functional point of view, also facilitating the therapeutic interventions to be prepared, according to models already known in the literature (e.g. ABA).

Contents of the manuscript

Introduction. General profiles and definitions

The concept of "autism" (from Greek αὐτός, aütós - same) has evolved. If the precursors who had delved into certain symptoms such as John Langdon Down and Ludwig Binswanger in the last thirty years of the 19th century had focused on the dissociative manifestation of reality and fragmentation of the inner plane, later Emil Kraepelin and again Eugen Bleuler in 1911 began to speak of syndrome and disorders, linking them, however, to schizophrenia; it was not until Melanie Klein in the 1930s, Hans Asperger in 1938 and Leo Kanner in 1943 that childhood psychosis, pathological syndrome and early childhood autism were spoken of [1,2].

Psychoanalysis is also concerned with the topic, trying to make interpretative contributions, by Anna Freud, Margaret Mahler, and Bruno Bettelheim, albeit speculative and of dubious scientific validity [3,4], as well as the scientific fraud perpetrated by Andrew Wakefield who correlated autism with the use of certain vaccine therapies (trivalent type) [5], at the expense of an exact nosographic placement among neurobiological disorders; the current definition of "Autism Spectrum Disorders (ASD), encompasses a whole constellation of symptoms and syndromes at varying degrees or levels of

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intensity, thus fueling confusion between the neuropsychiatric nature (as a syndrome of neurological origin) and the purely psychiatric nature (classifying it only as a mental disorder). And it is precisely this need for better clinical framing that has prompted reviewers of the DSM psychiatric manual, in its fifth edition, to unify several syndromes under the broader category of autism spectrum disorders, without, however, clarifying the high variability of psychiatrically derived dysfunctional traits [6-13] and closely related neurological syndromes, such as epilepsy, macrocephaly, hydrocephalus, cerebral palsy, and other abnormalities of the nervous system [14-16].

Autism is a highly variable neurodevelopmental disorder that initially appears during childhood and generally follows a steady course with no remission: symptoms slowly begin to manifest from the age of six months, becoming more explicit from the age of two or three years and continuing to increase into adulthood, although often in a less obvious form. The condition, however, is distinguished not by a single symptom (which might instead be a representation of a specific characteristic) but by a triad of characteristic symptoms: deficits in social interaction, deficits in communication, and restricted and repetitive interests and behaviors. Still, other aspects, such as atypical feeding, are also common but are not essential for diagnosis; in fact, it is no coincidence that individual symptoms of autism can be found in the general population, but for one to be able to speak of pathology, it is necessary to distinguish the situation by severity. Epidemiological statistics are also biased in this direction, in that they speak of a ratio of 5-10 per 10,000 individuals, without taking into account the dysfunctional findings of individual pathological traits that impair certain cognitive and psychic functions but are not sufficient to diagnose autistic spectrum [7].

In DMS-V, *autism* has been framed according to a new diagnostic orientation (2013) which, in addition to replacing the expression "*Pervasive (or generalized) developmental disorders*" with the term "Autism spectrum disorders", also eliminates the presence of the different subtypes of the pathology or forms of autism, such as *Asperger's syndrome*, *Rett's syndrome*, *Disintegrative disorder* and *Pervasive disorder not otherwise specified* [7].

In DSM-V, on the other hand, the autistic disorder has an autonomous structure, absorbing almost all previous pathologies, becoming "*Autism Spectrum Disorder*", diagnosed according to these criteria (A, B, C, and D): [7].

A. Persistent deficit in social communication and social interaction in different contexts, which cannot be explained by a generalized delay in development and manifested by all three of the following points: 1) deficit in socio-emotional reciprocity that goes from an abnormal and unsuccessful social approach in the normal conversation (question and answer) through a reduced sharing of interests, emotions, mental perception and reaction up to the total lack of initiative in social interaction; 2) deficits in non-verbal communication behaviors used for social interaction, from a poor integration of verbal and non-verbal communication, through abnormalities in eye contact and in body language, or deficits in understanding and using non-verbal communication, up to the total lack of facial expressiveness and gestures; 3) deficit in the creation and maintenance of relationships appropriate to the level of development (not including those with parents and caregivers); ranging from difficulty in adapting behavior to different social contexts through difficulties in sharing imaginative play and making friends to the apparent lack of interest in people.

B. The pattern of restricted, repetitive behaviors, interests, or activities as expressed by at least 2 of the following points: 1) language, movements, or use of stereotyped or repetitive objects, such as simple motor stereotypes, echolalia, repetitive use of objects, or idiosyncratic phrases; 2) excessive fidelity to the routine, verbal or non-verbal behavior reused or excessive reluctance to change: motor rituals, insistence on doing the same road or eating the same food, incessant questions or extreme stress following minor changes;3) highly restricted and fixed interests, abnormal in intensity or arguments: strong attachment or interest in unusual objects, excessively persistent or detailed interests; 4) hyper/hyporeactivity to sensory stimuli or unusual interests towards sensory aspects of the environment: apparent indifference to heat/cold/pain, adverse response to specific sounds or consistencies, excessive sniffing or touching objects, attraction to lights or spinning objects.

C. Symptoms must be present in early childhood (but may not become completely manifest).

D. The set of symptoms must limit and compromise daily functioning. The 3 severity levels are 3 (requires very substantial support); 2 (requires substantial support); 1 (requires support).

To sum up, to simplify, the autistic spectrum according to DSM 5 is therefore mainly characterized by symptoms relating to the areas of communication and social interaction: [7].

A. difficulties in socio-emotional reciprocity: the presence of an abnormal social approach and difficulty in sustaining and maintaining a conversation with the other; reduced sharing of interests and emotions; inability or difficulty in developing interaction with peers appropriate to the level of development;

B. deficit of non-verbal communicative behaviors used for social interaction: anomalies of visual contact (lack of direct gaze), facial expressions, body postures, and understanding or use of the gestures that regulate interaction with the other;

C. difficulties in the development and maintenance of relationships appropriate to the level of development: difficulty in adapting behavior according to the various social contexts, difficulties in the development and sharing of a game of imagination, and lack of interest in peers.

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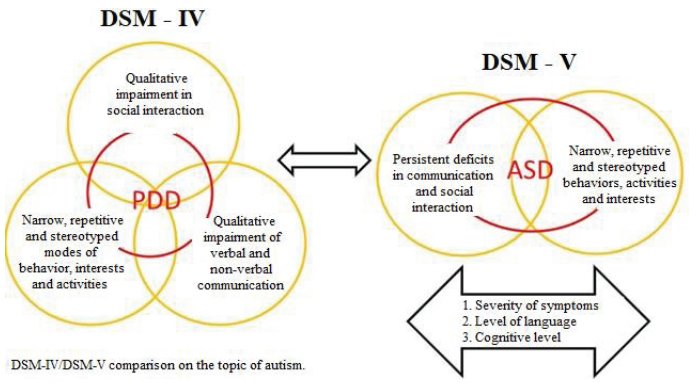

DSM-IV/DSM-V comparison on the topic of autism.

Thus, the need for this study is to identify a new developmental model of the autism spectrum, which would allow for better nosographic framing, at all developmental stages of the individual.

The autism spectrum has a high degree of heterogeneity that makes it difficult to understand, especially before 24 months of age, how to recognize whether a child has autism. The individual with autism can have features and symptoms ranging from very mild to very severe and marked and can occur in all ethnic, socioeconomic, and age groups. Males also are four times more likely to have autism than females. Some children with autism then appear homotypic, before the age of one or two, and then suddenly "regress" by going on to lose the language or social skills they had previously acquired. The heterogeneity of the symptoms and characteristics of the autism spectrum determines the fact that the signs and signals of autism that allow for early diagnosis are also variable among themselves, heterogeneous, and difficult to detect and interpret correctly. These aspects place at the center of the process toward early diagnosis the role of the parent in recognizing the signs of autism that will need to be reported to the clinical team, starting with the pediatrician and ending with the specialized services that will set up and implement individualized rehabilitation treatment. As a parent, you are in the best position to detect early signs and signals of autism. If autism is recognized during childhood, treatment can take full advantage of the remarkable plasticity of a child's brain and the effectiveness of the latest and most up-to-date behavioral intervention techniques. Although an autism spectrum disorder is difficult to recognize and diagnose before 24 months of age, signs of autism often emerge between 12 and 18 months of age [7].

Early signs of autism involve the absence of normal behaviors, not the presence of abnormal behaviors, and for this reason, they can be quite difficult to detect and interpret correctly; in some cases, early symptoms of autism can even be misinterpreted as signs of a "good child" or a child's particularly quiet temperament, as the child may appear calm, independent, and undemanding. However, alarm bells can be detected early if one knows what to look for, and that is the main focus of this informative article. Typical signs detectable early (by age two) by the parent, in interaction with their child, include the following: do not smile at you when you smile at him (poor emotional reciprocity); does not look at you while you feed him (limited or absent eye contact); does not respond to his name or the sound of a familiar voice; does not imitate your movements and facial expressions; does not share or present reciprocity of interest and enjoyment; does not visually follow objects or your gesture when you point things out (lack of shared attention); does not greet or use other gestures to communicate; does not make noises or vocalizations to get your attention; does not seek cuddles or seek to be picked up. Clinical signs, on the other hand, typical during outpatient visits with the pediatrician, always under 24 months, and unless otherwise indicated, are the following: by 6 months, no big smiles or other positive emotional expressions of amusement; by 9 months, no sharing of sounds, smiles or other facial expressions; by 12 months, no gestures of social reciprocity such as pointing or showing; by 12 months, no vocalizing; by 12 months, lack of response to name (no turning to voice); by 14 months, the child does not point to distant objects; by 16 months, no "simple words" spoken; by 24 months, no meaningful two-word phrases that do not involve mere imitation or repetition [7].

As children grow older, the signs of autism become more articulated and heterogeneous, and the criteria for recognizing autism revolve around impaired social skills, difficulty with nonverbal communication, rigidity of thought and behavior (which can cause major problem behaviors, and difficulty with language (in case it is present, even if in partial form). Concerning social and interaction skills the main signs of autism beyond the age of two years may be (and unless otherwise clinically indicated): difficulty in understanding feelings or talking about them; lack of participation in group play; difficulty in the ability to imitate the behavior of others; total or partial lack of attention when others talk to him; lack of sharing his interests and what arouses his attention with others; disinterest or unawareness of other people's behavior; difficulty in the ability to establish contact with age peers in playing or making friends; refusal or disinterest in physical contact; lack of imaginative play (e.g. making a plane with a fork, using games to simulate a story). Concerning nonverbal communication, the main signs of autism may be the failure to understand the meaning and message of facial expressions, tone of voice, and gestures of others; poor use of gestures (e.g., pointing with hands); avoidance of eye contact or, in any case, poor and elusive eye contact in many cases; poor or incorrect emotional expressiveness through facial expressions. Such communication impairment and difficulty further impact difficulties in the area of social skills. With respect to rigidity of thought and behavior, the main signs of autism may be: showing great interest in certain unusual areas (e.g., numbers, symbols, train schedules or sports statistics); spending long periods watching moving objects focusing on a specific part of that object (e.g., fan, washing machine, ...); frequently repeating the same actions or movements; reacting unusually or excessively to images, smells, textures and sounds (e.g., "I'm not sure if I'm sensitive to the same thing. may be particularly sensitive to loud noises and cover his ears when he hears them); unresponsive to people entering/leaving the room, as well as to others' efforts to get the child's attention; abnormal postures, clumsiness or eccentric ways

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of moving (e.g., walking exclusively on tiptoes); following a rigid routine and needs to do things the same way all the time (e.g., insists on taking a specific route to school); difficulty adapting to changes in the program or environment (e.g., shows annoyance if the arrangement of furniture, his toys, or the organization of the day is changed); showing great interest in unusual or strange objects such as keys, light switches, or rubber bands with which he interacts all the time; obsessive alignment of things or arrangement in a certain precise order. Concerning language difficulties (in cases where language is present, even if impaired), the main signs of autism may be: using language incorrectly (grammatical errors, misspelled words); referring to himself in the third person (pronominal inversion or confusion in the use of pronouns); taking what is said literally (literal interpretation of language); speaking in an abnormal tone of voice or rhythm; repeating the same words or phrases with high frequency, often without communicative intent; responding to a question by repeating it, instead of answering it. However, in some special cases, such as in Asperger's syndrome, language acquisition may be early and the child presents good vocabulary and sentence syntax while maintaining difficulties with prosody, intonation, and literal interpretation [7].

Many psychometric instruments in the literature can detect the diagnosis of the autism spectrum, such as in the case of the Repetitive Behaviour Questionnaire-2 (RBQ-2A), Autism Parent Screen for Infants (APSI), Battelle Development Inventory (BDI-2), Brief Infant-Toddler Social and Emotional Assessment (BITSEA), First Year Inventory (FYI), Infant-Toddler Checklist/ Communication and Symbolic Behavior Scales Developmental Profile (ITC/CSBS-DP), Program of Research and Studies on AUTISM (PREAUT-Grid), Checklist for Early Signs of Developmental Disorders (CESDD), Autism-Spectrum Quotient for Adult (AQa); Social Attention and Communication Study (SACS), Childhood Asperger Syndrome Test (CAST), and Checklist for Autism in Toddlers, Revised/ Revised with Follow-Up (M-CHAT-R/F) [17-25].

However, none of them identifies for the late teenage years and adult population the individual dysfunctional traits even in the hypotheses of the non-sufficiency presence of the conditions to diagnose the disorder, just as none of them identifies the dysfunctional hypotheses in a framework of substantial functionality. The purpose of this study is therefore to improve the nosographic approach to the disorder under consideration and to foster a different perspective that does not only take into account the rigid and structured descriptive framework found in the DSM manual but can also ensure the recognition of individual dysfunctional features that although not sufficient to describe the pathological phenomenon is nevertheless capable of drawing a profile that has certain specificities, focusing more not so much on the structural component but more on the functional.

The new evolutionary theory of the autistic spectrum and its application model

Based on what has been stated, a new theory on the autistic spectrum (Perrotta Evolutionary Theory on Autistic Spectrum, PETA) is suggested that takes into account its neurobiological matrix and explained by the related model (Perrotta Evolutionary Model on Autistic Spectrum, PEMA) that distinguishes the presence of individual dysfunctional traits (graded by the Perrotta Autistic Spectrum Severity Scale, PASS, on five levels) from autism spectrum disorder as defined by the DSM-V.

1. Specifically, the model identifies the following 9 dysfunctional traits (type-deficit and type-clinical features) for the late Teenage years and Adults:***Persistent deficits in verbal-emotional communication and social interaction*** **(not explained by generalized developmental delay):** Ranging from an abnormal and unsuccessful social approach in normal conversation (question-answer) to reduced sharing of interests, emotions, perception, and mental reactions, to a total lack of initiative in social interaction.

2. ***Persistent deficits in nonverbal-emotional communication and social interaction*** **(not explained by generalized developmental delay):** Poor paraverbal and nonverbal communication, abnormalities in eye contact and body language, deficits in understanding and use of nonverbal communication, to a total lack of facial expressiveness.

3. ***Persistent social-emotional communication and social interaction deficits*** **(not explained by generalized developmental delay):** Ranging from difficulty adapting behavior to different social contexts to difficulty sharing imaginative play and making friends to apparent lack of interest in people to reluctance to change and excessive loyalty to routine.

4. ***Persistent deficits in the area of language*****:** Echolalia and idiosyncratic sentences.

5. ***Persistent deficits in the area of movement and action*****:** Apraxia, impaired toe-walking, simple motor stereotypes, motor rituals, and repetitive use of objects.

6. ***Persistent deficit of the expressive-communicative area*****:** Excessive emotional-communicative response and excessively stupefied face concerning the external event.

7. ***Persistent deficit of the perceptual-sensory area*****:** Highly limited and fixed interests but abnormal in intensity or subject matter, strong attachment to or interest in unusual objects, excessively persistent or detailed interests, hyper/hyporesponsiveness to sensory stimuli or unusual interest in sensory and environmental aspects, apparent indifference/hyper-excitement to heat/cold/pain, abnormal response to specific sounds or textures, excessive need to sniff/smell/touch objects, attraction to lights or moving objects.

8. ***The persistent deficit in the intimate-sexual area*****:** Difficulty in establishing, and then maintaining, an effective, loving, and sexual relationship that is also lasting, pleasurable, and rewarding.

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9. *Persistent cognitive-psychiatric matrix characteristics*: IQ below 70 or above 120 and obsessive-compulsive tendencies, manic type hyperactivity, attention deficit, deep anxious states, paranoid and delusional slippage, phobias, aggression, schizoid (need for isolation/ closure/voluntary limitation concerning insistent or pervasive external stimuli) and schizotypy (overly imaginative ideas and mystical-magical beliefs), impaired empathic ability, self-injurious acting out, sleep disturbances and epilepsy.

Perrotta Dysfunctional Traits of the Autistic Spectrum Questionnaire for the late Teenage years and Adults (PTAS-Qa)

Based on the definition of "Autism Spectrum Disorder" (ASD) recognized by the DSM-V (APA), as an early-onset neurodevelopmental disorder characterized by difficulties in interaction, deficits in social communication, presence of narrow interests, and repetitive and stereotyped behaviors, a questionnaire was developed that could differentially grade the individual nine identified traits, based on a specific score for each survey area. The following scale grading and questionnaire construction are to be considered working proposals, which are in the process of validation to search for the population sample for a clinical study.

The questionnaire is suitable for individuals 16 years of age and older and is structured in two parts: the first part (Section A) is devoted to preliminary data about the patient; the second part (Section B), on the other hand, is devoted to the actual questionnaire and specifically, the questionnaire consists of 40 items (divided into nine subject areas related to the nine dysfunctional traits), on a binary yes/no scale. The sum of the relative scores, concerning yes answers only, determines the final score, and this also determines the severity of the autistic condition:

Perrotta Autism Severity Scale (PASS)

The minimum score is 0/11; the maximum score is 11/11, but the diagnosis of "*Autism Spectrum Disorder*" is determined only if all three of the following conditions will be met:

(a) The final score must be at least 4.1/11 or higher;

(b) The following sub-items must have been answered positively: 1.1, 1.5, 1.7, 1.8, 1.15, 1.18, 1.21, 1.27, 1.34 and one or more of 1.28, 1.29, 1.30 e 1.33;

(c) There must be no other diagnostic hypotheses that explain the symptoms differently.

If even one of these conditions was not met but the score was still equal to or above 4.1 and up to 6 one would speak of "modest functioning in the presence of autistic traits", between 6.1 and 8 one would speak of "poor functioning with marked autistic traits", and above 8 one would speak of "inadequate functioning in the presence of differently framed autistic spectrum". If one or more conditions were not met and the score was between 2.1 and 4 one would speak of "normal functioning in the presence of autistic traits", while equal to or below 2 one would speak of "no autism spectrum diagnosis".

Score	Presence of the 3 conditions	Diagnosis
0-2	NO	Absence of autism spectrum diagnosis
2.1-4	NO	Normal functioning, in the presence of autistic traits
4.1-6	NO	Modest functioning, in the presence of autistic traits
6.1-8	NO	Poor functioning, in the presence of autistic traits
8-11	NO	Inadequate functioning, in the presence of autism spectrum differently framed
<4	YES	Condition not feasible
4	YES	"Autism spectrum disorder (mild form)"
4.1-6	YES	"Autism spectrum disorder (moderate form)"
6.1-8	YES	"Autism spectrum disorder (severe form)"
8-11	YES	"Autism spectrum disorder (extreme form)"

Conclusion

The proposed model, accompanied by a graded scale and a questionnaire, provides the therapist with an accurate snapshot of both the presence of certain autistic traits and the possible diagnosis of "Autism Spectrum Disorder", while also offering a structured view of the patient's cognitive and psychological organization, avoiding diagnostic fragmentations that might not be consistent with the nosographic picture and therefore not perfectly framed. By exemplifying the category, it is, therefore, possible to more easily frame the patient, who would thus be analyzed from a strictly functional point of view, also facilitating the therapeutic interventions to be prepared, according to models already known in the literature (e.g. ABA).

(**Questionnaire**)

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