JOURNAL OF PSYCHOPATHOLOGY 2026;32:135-145
DOI: 10.36148/2284-0249-2352

Review

Diagnostic Expansion & Retrospective Autism Diagnosis in Adults: Balancing Recognition and Risk

Giuseppe Maina[1,2],Gabriele Di Salvo[1,2], Camilla Perotti[1], Gianluca Rosso[1,2]

[1] Department of Neurosciences "Rita Levi Montalcini", University of Turin, Turin, Italy; [2] Psychiatric Unit, San Luigi Gonzaga University Hospital, Orbassano, Turin, Italy

SUMMARY

Psychiatric diagnosis is simultaneously one of the most indispensable and conceptually fragile instruments in clinical medicine. It is indispensable because treatment planning, clinical communication, service provision and research all depend on a shared diagnostic language. At the same time, it remains fragile, as psychiatric categories are interpretative constructs rather than fixed biological disease entities. Over the past two decades, autism spectrum disorder has become increasingly visible in adult psychiatry, with a rise in first diagnoses made in adulthood. This shift has corrected important blind spots, particularly for women, cognitively able individuals and those without classic childhood presentations. However, the expanding clinical and cultural prominence of autism has also introduced new risks. In adults, autism is increasingly invoked as a retrospective explanatory framework for longstanding psychiatric histories previously conceptualized as obsessive-compulsive disorder, social anxiety disorder, schizoid or avoidant traits, trauma-related dysregulation or schizophrenia-spectrum conditions. While some late diagnoses are valid and clinically useful, others may reflect psychometric over-phenotyping, uncritical use of screening tools or a conceptual flattening of complex psychopathology into a more salient neurodevelopmental narrative. These developments also have medico-legal implications, as later autism diagnoses may be reframed as claims that earlier clinicians should have identified the condition. This paper examines the expanding boundaries of retrospective autism diagnosis in adults through diagnostic expansion, over-phenotyping, conceptual reductionism and historical diagnostic humility. It argues for a more disciplined approach that preserves improved recognition while limiting overconfident reinterpretation, unwarranted litigation and distortion of clinical and judicial resources

Key words: autism spectrum disorder (ASD), adult diagnosis, retrospective diagnosis, diagnostic expansion, over-phenotyping

Giuseppe Maina
E-mail: giuseppe.maina@unito.it

How to cite this article: Maina G, Di Salvo G, Perotti, et al. Diagnostic Expansion & Retrospective Autism Diagnosis in Adults: Balancing Recognition and Risk. Journal of Psychopathology 2026 Jul 28 Online First. https://doi.org/10.36148/2284-0249-2352

© Copyright by Pacini Editore Srl

OPEN ACCESS

This is an open access article distributed in accordance with the CC-BY-NC-ND (Creative Commons Attribution-NonCommercial-NoDerivatives 4.0 International) license. The article can be used by giving appropriate credit and mentioning the license, but only for non-commercial purposes and only in the original version. For further information: https://creativecommons.org/licenses/by-nc-nd/4.0/deed.en

Introduction

Psychiatric diagnosis occupies a peculiar position within medicine. In many areas of somatic pathology, diagnosis aspires to refer to a relatively stable lesion, pathogen, physiological defect or biological process. Psychiatry proceeds differently. Its disorders are usually inferred from clusters of symptoms, developmental trajectories, patterns of impairment and longitudinal course. That does not make psychiatric diagnosis illegitimate, but it does make it especially vulnerable to historical change, conceptual drift and overinterpretation.

This vulnerability has long been recognized. Hyman described the problem of diagnostic reification, namely the tendency to treat practical diagnostic constructs as if they were fully natural kinds rather than historically evolving tools of clinical reasoning[1]. Kendler likewise emphasized that the relationship between current psychiatric criteria and underlying disease reality remains incomplete and provisional[2]. These observations are not merely philosophical. They become clinically urgent whenever a diagnostic category rapidly expands its explanatory reach.

Adult autism diagnosis is now one of the clearest examples of this phenomenon. The rise of first diagnoses in adulthood has been driven by multiple convergent developments: broader spectrum concepts, increased public awareness, recognition of female and less prototypical presentations, wider use of screening tools, and the availability of neurodevelopmental narratives that help many adults reinterpret difficult life histories. Much of this has been beneficial. Yet precisely because the autism framework has become more available, more resonant and more clinically prominent, the risk of overextension has increased. A diagnosis can be both valid and vulnerable to inflation. These are not mutually exclusive propositions.

The present paper focuses on that tension. It does not deny the reality of missed autism, nor does it dispute that many adults were historically overlooked. Its concern is different. It is that autism, once established as an interpretative master category in adult psychiatry, may begin to absorb heterogeneous psychiatric phenomena that resemble it superficially but differ in developmental structure, phenomenology and treatment implications. The resulting problem is not simply overdiagnosis in the crude sense. It is something more nuanced: a drift toward psychometric over-phenotyping and conceptual reductionism, often reinforced by retrospective certainty that exceeds what the clinical evidence can reasonably sustain.

Materials and methods

The present study was conducted as a structured narrative review with systematic search elements, aimed at examining the expansion of retrospective autism diagnoses in adult psychiatry and its implications for diagnostic validity, differential diagnosis, psychometric assessment, and medico-legal reasoning.

A literature search was performed in PubMed/MEDLINE, Scopus, and Google Scholar for articles published in English between January 2000 and January 2026. Additional sources were identified through manual screening of reference lists and inclusion of key conceptual and classificatory works. Search terms included combinations of “autism spectrum disorder”, “adult diagnosis”, “late diagnosis”, “retrospective diagnosis”, “camouflaging”, “female autism”, “AQ”, “ADOS-2”, “diagnostic validity”, “overdiagnosis”, “diagnostic inflation”, “obsessive-compulsive disorder”, “social anxiety disorder”, “schizophrenia”, “psychosis”, “bipolar disorder”, “differential diagnosis”, and “medico-legal”.

Studies were included if they addressed adult or retrospective autism diagnosis, diagnostic classification, camouflaging or female presentation, diagnostic instruments, differential diagnosis, conceptual issues in psychiatric nosology, or medico-legal implications. Empirical studies, reviews, and major classificatory texts were included. Editorials without substantive contribution and studies not relevant to adult diagnosis were excluded.

Titles and abstracts were screened for relevance, followed by full-text assessment where necessary. Reference lists were manually searched for additional relevant studies. Given the heterogeneity of the literature, findings were synthesized narratively and organized into thematic domains.

Results

The structured review identified 46 relevant sources spanning empirical studies, epidemiological analyses, diagnostic instrument validation studies, classificatory frameworks, and conceptual contributions. The selection process is summarized in Figure 1.

Table I summarizes representative studies included in the review, with particular emphasis on adult autism diagnosis, delayed recognition, screening validity, and key differential diagnostic domains.

Table I summarizes representative studies included in the review, with particular emphasis on adult autism diagnosis, delayed recognition, screening validity, and key differential diagnostic domains.

Across the included literature, findings converged on six major domains: diagnostic expansion and evolving classification, rising adult diagnosis, female presentation and camouflaging, differential diagnosis with other psychiatric conditions, psychometric limitations of assessment tools, and medico-legal implications of retrospective diagnosis.

The evidence consistently supports a dual conclusion. First, the increase in adult autism diagnosis reflects a meaningful correction of historical underrecognition, particularly among cognitively able individuals and women. Second, the expansion of diagnostic frameworks, increased reliance on screening instruments, and broader cultural availability of autism narratives introduce a measurable risk of diagnostic overextension in complex adult psychiatric populations.

Diagnostic expansion and the sociology of psychiatric attention

The idea of diagnostic expansion should not be caricatured as a claim that the diagnosis itself is false. It refers instead to the process by which a valid category gradually comes to encompass a broader and more heterogeneous range of presentations. Batstra and Frances argued that this is often how inflation occurs in psychiatry: not by fraud or bad faith, but by the accumulation of well-intentioned changes that lower interpretative thresholds and widen boundaries[3]. Frances later developed the broader critique that modern psychiatry often

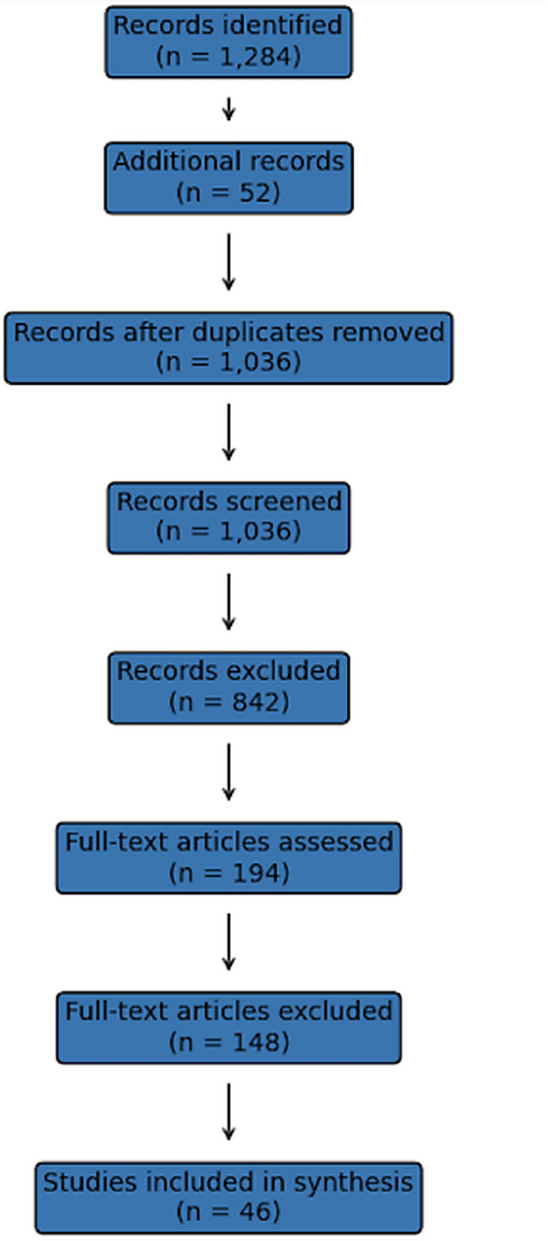


FIGURE 1. *PRISMA-style flow diagram illustrating the identification, screening, eligibility assessment, and inclusion of studies in this structured narrative review.*

medicalizes increasingly ordinary or nonspecific forms of distress under the pressure of expanding diagnostic sensibilities[4].

Autism illustrates a more complex version of the same dynamic. Here, the pressure is not to medicalize normality in any simple sense. It is to reinterpret longstanding psychiatric, interpersonal and developmental difficulties through an autism framework that has gained substantial explanatory and cultural authority. Lai and Baron-Cohen famously described a 'lost generation' of autistic adults who were excluded from earlier recognition by narrower concepts and underdeveloped services[5]. That formulation captured something important and true. But as the diagnosis becomes more culturally legible and more clinically available, the threshold for suspecting it may decrease, and the clinician may begin to see autistic structure in cases where the fit is less secure than the narrative appeal suggests.

Diagnostic evolution from DSM-IV to DSM-5 and DSM-5-TR

Changes in classification are central to this story. King and colleagues reviewed the major diagnostic reorganization introduced with DSM-5, which replaced autistic disorder, Asperger's disorder and pervasive developmental disorder not otherwise specified with the single umbrella diagnosis of autism spectrum disorder[6]. This was not a trivial revision. It changed both the language and the clinical imagination of autism, shifting attention toward a broader spectrum with more heterogeneous and more compensated presentations.

DSM-5-TR did not overturn this framework. Rather, it retained the DSM-5 spectrum model while refining and updating text across the manual. In practical terms, the most consequential change for adult psychiatry remains the DSM-5 move to a single spectrum construct. ICD-11 has also consolidated autism as a neurodevelopmental spectrum diagnosis, further reinforcing a broader international conceptualization[7-9]. This is one reason why current adult diagnoses cannot be straightforwardly mapped backward onto earlier diagnostic expectations. They arise within a significantly altered nosological environment.

Rising adult diagnosis: correction of underrecognition and risk of inflation

The growth of adult autism diagnosis is empirically well documented. Grosvenor and colleagues, using U.S. health system and claims data, found that autism diagnosis rates increased substantially between 2011 and 2022 among both children and adults, with especially marked increases among young adults and females[10]. Zeidan and colleagues, in a systematic review update of global prevalence, concluded that increases in measured prevalence likely reflect a combination of broadened definitions, greater awareness, improved case identification and expanded service capacity rather than a simple increase in underlying incidence[11].

These findings support two propositions at once. First, underrecognition was real. Second, prevalence growth alone cannot prove that every new diagnosis represents a previously obvious but missed case. Once a category

TABLE I. *Representative studies included in the structured narrative review, covering diagnostic expansion, adult autism diagnosis, screening instrument validity, differential diagnosis, and conceptual frameworks relevant to retrospective assessment.*

Study	Design / source type	Main population or focus	Main contribution	Relevance to present review
Baron-Cohen et al., 2001	Instrument development	Adults with Asperger syndrome/high-functioning autism and controls	Developed the Autism-Spectrum Quotient (AQ) as a trait measure	Foundational for discussion of screening tools and their intended scope
Booth et al., 2013	Brief report / validation	Adults screened with AQ-10	Evaluated AQ-10 as a brief ASD screener in adults	Supports argument that screening tools are triage instruments, not stand-alone diagnostic proof
Ashwood et al., 2016	Diagnostic prediction study	Adults referred for autism assessment	AQ predicted clinician diagnosis, but not with sufficient specificity to replace expert assessment	Supports limits of psychometric inference in adult diagnosis
Lehnhardt et al., 2013	Clinical review	Adults assessed for Asperger syndrome	Emphasized broad differential diagnosis and limits of simple screening approaches	Supports need for developmental and phenomenological integration
Conner et al., 2019	Outpatient clinic validity study	Adult psychiatric outpatient sample	Examined diagnostic validity of autism measures in real-world clinic population	Supports imperfect correspondence between measures and clinical diagnosis
Maddox et al., 2017	Diagnostic accuracy study	Adults with complex psychiatric conditions	ADOS-2 identified many autistic adults but showed high false-positive rates in psychosis	Central support for concerns about false positives in complex adult psychiatry
Grosvenor et al., 2024	Epidemiological trend study	US children and adults, 2011-2022	Showed marked increase in autism diagnosis rates, especially in young adults and females	Supports rise in adult diagnosis while not resolving validity of each new diagnosis
Zeidan et al., 2022	Systematic review	Global prevalence data	Found prevalence increases reflect broader definitions, awareness, and ascertainment changes	Supports distinction between rising identification and true incidence
Loomes et al., 2017	Systematic review and meta-analysis	Male-to-female ratio in ASD	Demonstrated lower male:female ratio than traditionally assumed	Supports historical underrecognition of females
Harrop et al., 2024	Trend analysis	State-wide female autism diagnostic rates	Documented increasing diagnostic rates among autistic females	Supports corrective dimension of recent diagnostic expansion
Hull et al., 2019	Questionnaire development/validation	Autistic adults	Developed CAT-Q to assess camouflaging autistic traits	Supports role of camouflaging in delayed recognition
Hull et al., 2021	Empirical association study	Autistic adults without intellectual disability	Camouflaging associated with anxiety and depression	Supports clinical significance of camouflaging, but also need for careful interpretation
Milner et al., 2024	Observational study	Autistic men and women	Camouflaging predicted later age at diagnosis, especially in females	Supports delayed diagnosis while highlighting interpretative complexity
Cath et al., 2008	Preliminary case-controlled study	Adults with ASD, generalized social anxiety disorder, or OCD	Showed symptom overlap across ASD, social anxiety, and OCD	Key support for superficial resemblance across diagnostic groups
Paula-Pérez, 2013	Differential diagnostic review	OCD versus repetitive/restrictive autistic phenomena	Distinguished obsessive-compulsive symptoms from autistic repetitive behaviors	Supports OCD-autism differential framework

TABLE I. *Follows from the previous page.*

Study	Design / source type	Main population or focus	Main contribution	Relevance to present review
Postorino et al., 2017	Review	Individuals with ASD and anxiety/OCD	Reviewed anxiety and OCD in ASD and associated diagnostic challenges	Supports overlap and comorbidity without collapsing boundaries
Stein et al., 2019	Primer/review	OCD	Clarified centrality of obsessions and ego-dystonic compulsions	Supports phenomenological distinction from autistic sameness/routines
Heimberg et al., 2014	Review	Social anxiety disorder in DSM-5	Emphasized fear of negative evaluation as core mechanism	Supports differential diagnosis between social anxiety and autism
Jutla et al., 2022	Conceptual review	Autism spectrum disorder and schizophrenia	Reviewed distinctions and overlaps between ASD and schizophrenia	Supports schizophrenia-spectrum differential diagnosis
Trevisan et al., 2020	Comparative empirical study	ASD versus schizophrenia	Found positive symptoms differentiate better than negative symptoms	Supports risk of confusion when only social withdrawal/oddity are considered
Joshi et al., 2013	Controlled clinical analysis	Youth with bipolar I disorder with/without ASD	Examined comorbidity and correlates of bipolar disorder and ASD	Supports need to distinguish trait autism from mood pathology
Borue et al., 2016	Longitudinal study	Youth with bipolar disorder and high-functioning ASD	Reported mixed affective presentations and functional impairment	Supports overlap and complexity in bipolar-autism presentations
Vannucchi et al., 2014	Systematic review	Adults with Asperger's syndrome and bipolar disorder	Found clinically meaningful comorbidity and diagnostic difficulty	Supports bipolar-spectrum differential section
Matsuo et al., 2015	Cross-sectional empirical study	Adults with mood disorders and schizophrenia	Found elevated autistic-like traits in mood and psychotic disorders	Supports concept of autistic-like traits as non-specific in adult psychiatry
Murphy et al., 2016	Clinical service review	Adults with ASD	Emphasized collateral history, specialist assessment, and careful differential diagnosis	Supports section on retrospective diagnosis and assessment standards
Lord et al., 2020	Primer	Autism spectrum disorder	Broad overview of autism diagnosis, course, and clinical features	Background reference for diagnostic framework
King et al., 2014	Classification review	DSM-5 autism diagnostic reorganization	Reviewed shift to single ASD spectrum diagnosis	Supports discussion of nosological broadening
Lai and Baron-Cohen, 2015	Commentary/review	Adults with previously missed autism	Proposed concept of the "lost generation"	Supports underrecognition argument while setting up risk of overextension

Abbreviations: AQ, Autism-Spectrum Quotient; AQ-10, 10-item Autism-Spectrum Quotient; ADOS-2, Autism Diagnostic Observation Schedule, Second Edition; CAT-Q, Camouflaging Autistic Traits Questionnaire.

expands and awareness rises, both valid recognition and conceptual inflation may occur simultaneously. Psychiatry has seen this before. Diagnostic growth is never self-interpreting. It must be understood against changing definitions, changing service pathways and changing expectations about what the diagnosis explains.

Women, diagnostic bias and camouflaging

One of the strongest arguments in favor of improved adult recognition concerns women. Loomes and colleagues showed in a landmark meta-analysis that the male-to-female ratio in autism is lower than the traditional 4:1 figure, suggesting that girls and women are disproportionately at risk of underdiagnosis[12]. Harrop

and colleagues later documented increasing diagnostic rates among autistic females over time, again supporting the conclusion that part of the recent expansion is corrective rather than merely inflationary[13].

The concept of camouflaging has sharpened this issue further. Hull and colleagues developed the Camouflaging Autistic Traits Questionnaire and demonstrated that masking and compensation are measurable, clinically meaningful and common in autistic adults[14]. In a later study, the same group found that higher camouflaging was associated with anxiety and depression in autistic adults without intellectual disability[15]. Milner and colleagues then reported that camouflaging predicted later age at diagnosis more strongly in females than in males, reinforcing the idea that delayed diagnosis can arise from compensatory adaptation rather than absent symptoms[16].

These studies are important. They make it impossible to dismiss all adult diagnosis as fashionable excess. Yet they also introduce a new interpretative hazard. Once camouflaging becomes a widely available explanatory device, the absence of overt developmental signs may itself be transformed into evidence for the diagnosis. At that point, the diagnosis risks becoming difficult to disconfirm. Camouflaging is real, but it cannot function as a universal escape hatch from the demand for developmentally coherent evidence.

Phenomenology and the danger of conceptual flattening

At a deeper level, the problem is phenomenological. Classical psychopathology, particularly in the tradition of Jaspers, insisted that symptoms are not reducible to their outward form. They acquire diagnostic meaning only within the broader structure of subjective experience and developmental context. More recent phenomenological work, including that of Parnas and Sass, has similarly warned against the loss of interpretative depth when psychiatry leans too heavily on checklist logic and surface description[17,18].

This matters because many of the features that now trigger autism suspicion in adults are not specific to autism. Social withdrawal, rigid routines, interpersonal awkwardness, repetitive thought and sensory discomfort can all occur in autism. But they can also arise from obsessive-compulsive disorder, social anxiety, trauma, chronic shame, depression, schizoidity or schizophrenia-spectrum conditions. If the clinician focuses only on observable behavior, distinct experiential structures can appear deceptively similar. What results is conceptual flattening: a reduction of heterogeneous psychopathology into a smaller number of more fashionable categories.

Autism and obsessive-compulsive disorder

The autism-OCD differential remains one of the most clinically important examples. Paula-Pérez examined the distinction between obsessive-compulsive disorder and restrictive and repetitive autistic behaviors, emphasizing differences in emotional valence, psychological function and underlying mechanism[19]. Stein and colleagues, in their review of obsessive-compulsive disorder, reiterated the centrality of intrusive obsessions and ego-dystonic compulsions as a threat-reduction strategy[20]. In autism, by contrast, routines and repetitive behaviors more often function as self-regulation, predictability or focused engagement and may be experienced as comforting or identity-congruent rather than alien and unwanted[21].

Cath and colleagues demonstrated symptomatic overlap among autism spectrum disorder, generalized social anxiety disorder and obsessive-compulsive disorder in adults, highlighting the risk of superficial diagnostic resemblance in clinically complex populations[22]. Postorino and colleagues further reviewed the high rates and assessment challenges of anxiety disorders and OCD in autistic individuals, underscoring how common true overlap can be[23]. The diagnostic task, then, is not merely to note repetition and rigidity, but to determine whether they are driven by intrusive fear, by preferred sameness, by sensory regulation, or by something else entirely.

Autism and social anxiety disorder

The differential diagnosis with social anxiety is no less important. In social anxiety disorder, avoidance is fundamentally organized around fear of scrutiny, humiliation and negative evaluation. In autism, social difficulty is more likely to arise from problems decoding reciprocity, implicit rules and conversational timing, or from exhaustion and overload in social environments. Heimberg and colleagues described social anxiety as rooted in anticipatory fear and self-focused evaluative concern, a structure quite different from the more basic social-cognitive mismatch typical of autism[24].

Yet the outward picture may overlap. Both groups may avoid social situations, appear awkward or quiet, or report feeling out of place. This is why longstanding social anxiety histories are particularly vulnerable to retrospective autistic reinterpretation. Sometimes such reinterpretation is clinically justified. Sometimes it obscures precisely the fear-based mechanism most relevant for treatment. The key question is not simply whether the person avoids social situations, but why.

Autism and schizophrenia-spectrum conditions

The autism-schizophrenia differential raises different

but equally important issues. Jutla and colleagues emphasized that autism and schizophrenia are distinct disorders with different natural histories and core features: autism is an early developmental condition with a relatively stable course, whereas schizophrenia usually emerges later and is characterized by hallucinations, delusions and declining function[25]. Trevisan and colleagues showed that autism and schizophrenia are better differentiated by positive symptoms than by negative symptoms, precisely because social withdrawal, flatness and communication oddity may appear in both[26].

This is crucial in adult retrospective diagnosis. Social detachment, reduced eye contact and unusual communication do not by themselves distinguish autism from schizophrenia-spectrum conditions. Psychosis cannot be reduced to autistic difference, and a later autism diagnosis should not erase the qualitative specificity of hallucinations, delusional conviction or formal thought disorder. Here again, the risk is not merely diagnostic error in the narrow sense. It is a broader conceptual substitution that smooths over psychopathological distinctions under the pressure of a more available narrative.

The differential diagnosis between autism spectrum disorder and bipolar-spectrum conditions deserves explicit consideration, because several affective and behavioral manifestations of bipolarity may be misread as autistic traits, especially in adults with complex or longstanding psychiatric histories. This problem is amplified by the fact that both conditions can involve social withdrawal, irritability, agitation, mood lability, aggressive outbursts, sleep disturbance and fluctuating functioning. While the developmental architecture of autism differs fundamentally from the episodic nature of bipolar disorder, the surface phenomenology may, at times, appear deceptively similar. A failure to distinguish longitudinal trait-like autistic features from state-dependent affective dysregulation may therefore contribute to retrospective overdiagnosis of autism, or conversely to the underrecognition of bipolar-spectrum psychopathology in autistic individuals.

A first area of overlap concerns **irritability, agitation and aggression**. These are often discussed in autism, particularly when patients present with behavioral dysregulation, sensory overload, frustration intolerance or abrupt emotional escalation. Yet they are also prominent in bipolar-spectrum conditions, especially in mixed states, dysphoric mania, rapid cycling presentations and bipolar depression with activation. Oakley and colleagues noted that mood problems in autism may present with atypical features such as severe irritability, making recognition of affective disorders more difficult[27]. In parallel, the literature on bipolar disorder in autistic populations has repeatedly stressed that irritability, impulsive aggression and behavioral activation may be mistakenly attributed to autism itself rather than recognized as part of a comorbid mood episode. This is clinically important, because the treatment implications differ substantially.

A second area of overlap involves **depression and social withdrawal**. In autism, social detachment or reduced social engagement may reflect lifelong differences in social reciprocity, sensory fatigue, difficulty navigating implicit social rules or a preference for predictability over social complexity. In bipolar depression, by contrast, withdrawal is more likely to be state-dependent, associated with lowered drive, hopelessness, anergia, diminished reactivity and depressive inhibition. However, when the clinical history is reconstructed retrospectively, depressive withdrawal may be redescribed as evidence of "autistic sociality," particularly if the patient also reports long-standing interpersonal discomfort or rigid coping styles. The opposite error is also possible: genuine autistic social communication differences may be mistaken for depressive retreat. What distinguishes the two is not outward social reduction alone, but developmental continuity, temporality, and the phenomenological context in which the withdrawal occurs.

A third source of confusion concerns **mixed, unstable or atypical affective states**. Bipolar-spectrum conditions do not always present with classic euphoric mania. Especially in psychiatrically complex patients, one may see combinations of depressed mood, racing thoughts, agitation, irritability, impulsive outbursts and interpersonal volatility. Borue and colleagues, in their longitudinal study of youth with comorbid bipolar disorder and autism spectrum disorder, found that the bipolar-autism group more often showed distractibility, racing thoughts, depressed mood, social withdrawal and mixed symptom presentations, together with greater functional impairment[28]. Although that study was conducted in youth, its phenomenological implications remain highly relevant for adult psychiatry: bipolarity in autistic individuals may not look prototypically manic, and conversely, affective activation in nonautistic adults may be misperceived as part of a broad autistic phenotype.

The literature also suggests that **autistic traits and bipolar-spectrum psychopathology may be genuinely comorbid** rather than merely confounded. Vannucchi and colleagues, in a systematic review of bipolar disorder in adults with Asperger's syndrome, concluded that the co-occurrence is clinically meaningful and likely underrecognized, while also emphasizing how difficult the differential diagnosis can be when impulsivity, aggression, mood instability and atypical affective presentations are present[29]. Similarly, Matsuo and colleagues found that a substantial proportion of adults with bipolar

disorder showed high autistic-like traits, independently of current symptom severity, suggesting either shared vulnerability, phenotypic overlap, or both[30]. These findings reinforce the need for caution: elevated autistic-like traits in bipolar patients do not automatically establish autism spectrum disorder, but they do indicate that the boundary between the two conditions can become clinically blurred if developmental evidence is weak or if state-trait distinctions are not rigorously maintained.

For these reasons, the clinician should resist the temptation to interpret **agitation, aggression, irritability, social withdrawal or depressive flattening** as autistic by default in adults with possible bipolar-spectrum pathology. These symptoms may be similar, overlapping or comorbid, but they are not diagnostically interchangeable. The decisive questions remain developmental and longitudinal. Was the social communication pattern present from early life, across contexts and independent of mood episodes? Are rigidity and routine-seeking trait-like and organizing, or do they intensify mainly during affective destabilization? Is aggression linked to sensory overload and frustration around sameness, or to mixed-state irritability, dysphoric activation and affective impulsivity? Without such distinctions, retrospective diagnostic formulations risk collapsing episodic mood psychopathology into a trait-based neurodevelopmental narrative.

In this sense, bipolar-spectrum conditions represent an important test case for the broader argument of the present paper. They show how psychometric over-phenotyping and conceptual reductionism may arise when broad behavioral descriptors are detached from temporality, state dependence and phenomenological structure. They also show why adult autism diagnosis, particularly when retrospective, must remain anchored not only to trait capture but to developmental evidence, episodic course and differential psychopathology.

Psychometric over-phenotyping and the limits of adult autism instruments

The increasing use of screening and structured assessment tools has added another layer of complexity. These instruments are clinically useful, but their role is often misunderstood. Baron-Cohen and colleagues developed the AQ as a trait measure, not a stand-alone diagnostic instrument[31]. Booth and colleagues evaluated the AQ-10 as a brief adult screener and confirmed its possible usefulness in triage contexts, not as definitive proof of diagnosis[32]. Ashwood and colleagues showed that AQ scores can predict clinician diagnosis in adult referrals, but not with the degree of specificity required to replace expert assessment[33]. Lehnhardt and colleagues described the AQ as a simple but nonspecific screening instrument within a much broader diagnostic process[34]. Conner and colleagues examined several autism measures in an adult outpatient clinic and found that their correspondence with clinician diagnosis was imperfect in real-world settings[35]. Maddox and colleagues demonstrated that the ADOS-2 correctly identified autistic adults in their sample but also produced a high false-positive rate among adults with psychosis, reminding clinicians that the social-communication difficulties measured by ADOS-2 are not autism-specific in complex psychiatric populations[36].

This is where the concept of psychometric over-phenotyping becomes useful. Screening tools capture broad dimensions of behavior and experience. In highly comorbid adults, especially those with anxiety, obsessionality, trauma or personality pathology, several different instruments may simultaneously yield elevated scores. The patient then begins to accumulate diagnostic identities because the measures detect overlapping dimensions rather than discrete disease entities. Psychometric density is mistaken for diagnostic certainty. The more the clinician relies on scores without phenomenological and developmental integration, the greater the risk of over-phenotyping.

Retrospective diagnosis and historical humility

Adult autism diagnosis is frequently retrospective by necessity. The problem is not that retrospective diagnosis is illegitimate, but that it is methodologically vulnerable. A persuasive adult formulation may emerge only after detailed interviewing, richer collateral information, newer concepts and the patient's own reconstructed narrative have become available. That reconstruction may be valid. It may also be shaped by hindsight.

Murphy and colleagues stressed the importance of collateral developmental history, specialist assessment and careful differential diagnosis in adult autism services[37]. Lai and Baron-Cohen highlighted that first diagnosis in adulthood occurs in a context of increasing awareness and broadened criteria, which itself changes what becomes clinically thinkable[5]. The crucial lesson is that retrospective coherence is not the same as prospective recognizability. A later diagnosis can make a life history feel newly intelligible without proving that prior clinicians should necessarily have reached the same conclusion using older frameworks and less complete information.

Medico-Legal Risks of Retrospective Autism Diagnoses: diagnostic error, unwarranted litigation and the burden on justice systems

The medico-legal implications of this issue deserve much more attention than they usually receive. Once a

late diagnosis is made, the patient, family or legal representative may conclude that previous clinicians failed to recognize an underlying neurodevelopmental disorder that had been present all along. In some cases that conclusion may be justified. In others it is methodologically fragile. A diagnosis established years later under broader criteria, greater public awareness, richer retrospective narratives and more autism-literate services does not by itself demonstrate that the condition was clearly recognizable, much less diagnostically mandatory, at an earlier time.

This distinction matters because it separates present plausibility from past obligation. The relevant medico-legal question is not simply whether the patient meets current criteria now. It is whether a competent clinician, using the definitions, service expectations and differential diagnostic standards available then, should necessarily have made the diagnosis earlier. These are not the same question.

The confusion between them creates several risks. It can foster unwarranted or opportunistic litigation in which a late autism diagnosis is implicitly converted into proof of past negligence. It can distort professional judgment by encouraging clinicians to read older cases backward through contemporary concepts. It can also consume significant judicial and clinical resources. Weak or overextended claims still require records review, expert reports, institutional response and legal adjudication. When evolving diagnostic frameworks are retrospectively reified into standards of certainty that did not exist at the time, the result is not merely unfairness to individual clinicians. It is also a needless burden on already stretched health and justice systems. Historical humility is not a defensive reflex. It is an epistemic necessity in a field whose categories are themselves evolving. A later diagnosis should not be construed as retrospective proof of prior diagnostic error; in most cases, it is not.

Toward a balanced diagnostic framework

A balanced response requires that psychiatry hold two truths together. The first is that adult autism is real, clinically important and historically underrecognized in many people. The second is that improved recognition does not abolish the need for restraint. Diagnostic assessment in adults should remain anchored in developmental history, persistent cross-context impairment, careful phenomenological differentiation and rigorous exclusion of alternative explanations. Screening instruments should support assessment, not replace it. Female presentations and camouflaging should broaden clinical awareness, but they should not become universal explanations that remove the need for evidence.

In practical terms, this means that obsessive-compulsive disorder, social anxiety disorder and schizophrenia-spectrum conditions must be treated as central, not peripheral, differential diagnoses in adult autism assessment. It also means that the explanatory relief often produced by a late autism diagnosis should be respected without being mistaken for proof that no other formulation was ever reasonable.

Conclusions

The rise of retrospective autism diagnoses in adult psychiatry reflects genuine progress, but also genuine risk. It has corrected serious underrecognition, especially among women and cognitively able adults. It has also offered many people a coherent and helpful framework for understanding longstanding difficulties. These are major gains.

Yet gains in recognition do not cancel the danger of expansion beyond coherent boundaries. When autism becomes a dominant interpretative narrative, heterogeneous psychiatric histories may be reorganized too quickly under a neurodevelopmental template. Obsessionality may become autistic rigidity, social fear may become autistic withdrawal, and longstanding psychiatric complexity may be flattened into a more culturally available story than the evidence can fully support.

The concepts of diagnostic expansion, psychometric over-phenotyping and conceptual reductionism help clarify what is at stake. The challenge is not to oppose adult autism diagnosis, but to keep it disciplined. Psychiatry should become more sensitive without becoming conceptually lax, more inclusive without becoming diagnostically indiscriminate, and more historically informed without becoming retrospectively absolutist. That balance is difficult. It is also essential.

Conflict of interest statement

All authors declare that the research was conducted in the absence of any commercial or financial relationships that could be construed as a potential conflict of interest. Giuseppe Maina has been a consultant/speaker and/or has received research grants from Angelini, Boheringer, Innovapharma, Janssen, Fb Health, Otsuka, Lundbeck, Italfarmaco and Sanofi. Gabriele Di Salvo has been a speaker for Lundbeck and Angelini. Gianluca Rosso has been a speaker and/or consultant from Angelini, Lundbeck, Janssen and Otsuka outside of the current work. Nevertheless, all authors report that this did not result in conflicts of interest for the present study.

Funding

This research received no specific grant from any funding agency, commercial or not-for-profit sectors.

Authors' contributions

All authors have read and approved the final version

submitted and take public responsibility for all aspects of the work. Giuseppe Maina designed the study. Gabriele Di Salvo, and Camilla Perotti, managed literature search and Gianluca Rosso and Gabriele Di Salvo analyzed the data. Giuseppe Maina wrote the draft. Gianluca Rosso provided substantial comments and helped drafting the manuscript in its final form.

Ethical consideration

The manuscript presents a review of the existing literature and does not report any original research involving human participants or animals.

References

1 Hyman SE. The diagnosis of mental disorders: the problem of reification. Annu Rev Clin Psychol. 2010;6:155-179 https://doi.org/10.1146/annurev.clinpsy.3.022806.091532.

2 Kendler KS. The phenomenology of major depression and the representativeness and nature of DSM criteria. Am J Psychiatry. 2016;173(8):771-780 https://doi.org/10.1176/appi.ajp.2016.15121509

3 Batstra L, Frances A. Diagnostic inflation: causes and a suggested cure. J Nerv Ment Dis. 2012;200(6):474-479 https://doi.org/10.1097/NMD.0b013e318257c4a2.

4 Frances A. Saving normal: an insider's revolt against out-of-control psychiatric diagnosis, DSM-5, big pharma, and the medicalization of ordinary life. New York (NY): HarperCollins; 2013.

5 Lai MC, Baron-Cohen S. Identifying the lost generation of adults with autism spectrum conditions. Lancet Psychiatry. 2015;2(11):1013-1027 https://doi.org/10.1016/S2215-0366(15)00277-1.

6 King BH, Navot N, Bernier R, Webb SJ. Update on diagnostic classification in autism. Curr Opin Psychiatry. 2014;27(2):105-109 https://doi.org/10.1097/YCO.0000000000000040.

7 American Psychiatric Association. Diagnostic and statistical manual of mental disorders. 5th ed. Arlington (VA): American Psychiatric Publishing; 2013.

8 American Psychiatric Association. Diagnostic and statistical manual of mental disorders, Fifth Edition, Text Revision (DSM-5-TR). Washington (DC): American Psychiatric Publishing; 2022.

9 World Health Organization. Clinical descriptions and diagnostic requirements for ICD-11 mental, behavioural and neurodevelopmental disorders (CDDR). Geneva: WHO; 2024.

10 Grosvenor LP, Croen LA, Lynch FL, et al. Autism diagnosis among US children and adults, 2011-2022. JAMA Netw Open. 2024;7(10):e2442218. doi:10.1001/jamanetworkopen.2024.42218.

11 Zeidan J, Fombonne E, Scorah J, et al. Global prevalence of autism: a systematic review update. Autism Res. 2022;15(5):778-790 https://doi.org/10.1002/aur.2696.

12 Loomes R, Hull L, Mandy WPL. What is the male-to-female ratio in autism spectrum disorder? A systematic review and meta-analysis. J Am Acad Child Adolesc Psychiatry. 2017;56(6):466-474 https://doi.org/10.1016/j.jaac.2017.03.013.

13 Harrop C, Tomaszewski B, Putnam O, et al. Are the diagnostic rates of autistic females increasing? An examination of state-wide trends. J Child Psychol Psychiatry. 2024;65(7):973-983. doi:10.1111/jcpp.13939.

14 Hull L, Mandy W, Lai MC, et al. Development and validation of the Camouflaging Autistic Traits Questionnaire (CAT-Q). J Autism Dev Disord. 2019;49(3):819-833 https://doi.org/10.1007/s10803-018-3792-6.

15 Hull L, Lai MC, Baron-Cohen S, et al. Is social camouflaging associated with anxiety and depression in autistic adults? Mol Autism. 2021;12(1):13 https://doi.org/10.1186/s13229-021-00421-1.

16 Milner V, Colvert E, Hull L, et al. Does camouflaging predict age at autism diagnosis? Autism Res. 2024;17(3):626-636 https://doi.org/10.1002/aur.3059.

17 Jaspers K. General psychopathology. Manchester: Manchester University Press; 1963.

18 Parnas J, Sass LA. Self, solipsism, and schizophrenic delusions. Philos Psychiatry Psychol. 2001;8(2-3):101-120.

19 Paula-Pérez I. Differential diagnosis between obsessive compulsive disorder and restrictive and repetitive behavioural patterns in autism spectrum disorders. Rev Psiquiatr Salud Ment. 2013;6(4):178-186 https://doi.org/10.1016/j.rpsm.2012.07.005.

20 Stein DJ, Costa DLC, Lochner C, et al. Obsessive-compulsive disorder. Nat Rev Dis Primers. 2019;5(1):52 https://doi.org/10.1038/s41572-019-0102-3.

21 Lord C, Brugha TS, Charman T, et al. Autism spectrum disorder. Nat Rev Dis Primers. 2020;6(1):5 https://doi.org/10.1038/s41572-019-0138-4.

22 Cath DC, Ran N, Smit JH, van Balkom AJLM, Comijs HC. Symptom overlap between autism spectrum disorder, generalized social anxiety disorder and obsessive-compulsive disorder in adults: a preliminary case-controlled study. Psychopathology. 2008;41(2):101-110 https://doi.org/10.1159/000111555.

23 Postorino V, Kerns CM, Vivanti G, et al. Anxiety disorders and obsessive-compulsive disorder in individuals with autism spectrum disorder. Curr Psychiatry Rep. 2017;19(12):92 https://doi.org/10.1007/s11920-017-0846-y.

24 Heimberg RG, Hofmann SG, Liebowitz MR, et al. Social anxiety disorder in DSM-5. Depress Anxiety. 2014;31(6):472-479 https://doi.org/10.1002/da.22231.

25 Jutla A, Foss-Feig J, Veenstra-VanderWeele J. Autism spectrum disorder and schizophrenia: an updated conceptual review. Autism Res. 2022;15(3):384-412 https://doi.org/10.1002/aur.2659.

26 Trevisan DA, Foss-Feig JH, Naples AJ, et al. Autism spectrum disorder and schizophrenia are better differentiated by positive symptoms than negative symptoms. Front Psychiatry. 2020;11:548 https://doi.org/10.3389/fpsyt.2020.00548.

27 Oakley B, Loth E, Murphy DG. Autism and mood disorders. Int Rev Psychiatry. 2021;33(3):280-299. doi:10.1080/09540261.2021.1872506.

28 Borue X, Mazefsky C, Rooks B, et al. Longitudinal course of bipolar disorder in youth with high-functioning autism spectrum disorder. J Am Acad Child Adolesc Psychiatry. 2016;55(12):1064-1072.e6. doi:10.1016/j.jaac.2016.09.491.

29 Vannucchi G, Masi G, Toni C, et al. Bipolar disorder in adults with Asperger's syndrome: a systematic review. J Affect Disord. 2014;168:151-160. doi:10.1016/j.jad.2014.06.042.

30 Matsuo J, Kamio Y, Takahashi H, et al. Autistic-like traits in adult patients with mood disorders and schizophrenia. PLoS One. 2015;10(4):e0122711. doi:10.1371/journal.pone.0122711.

31 Baron-Cohen S, Wheelwright S, Skinner R, et al. The Autism-Spectrum Quotient (AQ): evidence from Asperger syndrome/high-functioning autism, males and females, scientists and mathematicians. J Autism Dev Disord. 2001;31(1):5-17 https://doi.org/10.1023/a:1005653411471.

[32] Booth T, Murray AL, McKenzie K, et al. Brief report: an evaluation of the AQ-10 as a brief screening instrument for ASD in adults. J Autism Dev Disord. 2013;43(12):2997-3000 https://doi.org/10.1007/s10803-013-1844-5.

[33] Ashwood KL, Gillan N, Horder J, et al. Predicting the diagnosis of autism in adults using the Autism-Spectrum Quotient (AQ) questionnaire. Psychol Med. 2016;46(12):2595-2604 https://doi.org/10.1017/S0033291716001082.

[34] Lehnhardt FG, Gawronski A, Pfeiffer K, et al. The investigation and differential diagnosis of Asperger syndrome in adults. Dtsch Arztebl Int. 2013;110(45):755-763 https://doi.org/10.3238/arztebl.2013.0755.

[35] Conner CM, Cramer RD, McGonigle JJ. Examining the diagnostic validity of autism measures among adults in an outpatient clinic sample. Autism Adulthood. 2019;1(1):60-68 https://doi.org/10.1089/aut.2018.0023.

[36] Maddox BB, Brodkin ES, Calkins ME, et al. The accuracy of the ADOS-2 in identifying autism among adults with complex psychiatric conditions. J Autism Dev Disord. 2017;47(9):2703-2709 https://doi.org/10.1007/s10803-017-3188-z.

[37] Murphy CM, Wilson CE, Robertson DM, et al. Autism spectrum disorder in adults: diagnosis, management, and health services development. Neuropsychiatr Dis Treat. 2016;12:1669-1686. https://doi.org/10.2147/NDT.S65455.