TPM Vol. 32, No. S9, 2025
ISSN: 1972-6325
https://www.tpmap.org/

Open Access

EXPLORING THE RISING PREVALENCE OF AUTISM SPECTRUM DISORDER: GENETIC AND ENVIRONMENTAL FACTORS, DIAGNOSTIC APPROACHES, AND INNOVATIVE THERAPIES

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ABSTRACT
Autism Spectrum Disorder (ASD) is a heterogeneous neurodevelopmental condition characterized by impairments in communication, behavior, and social interaction. Evidence documenting global prevalence in the last decade shows noteworthy increases, allowing researchers to narrow in on biological and anthropogenic drivers of the observed trends. Hence, this review focuses on the available literature from 2016 to 2025 on the genetic and environmental drivers of the condition, novel constructs in the diagnosis of the condition, and contemporary literature on the treatment of the condition. This review was done systematically in keeping with the PRISMA 2020 guidelines. Literature published in the years 2016 to 2025 which was available on PubMed, Scopus, Web of Science, and Embase. Of the expected fifteen studies, only thirteen fit the full criteria and were included. These studies focused on the genetic, environmental, diagnostic, and interventional components. Of the studies included in this review, there were claims of the heritability of the condition falling between the ranges of seventy to ninety percent with claims there were also other rare polygenetic correlations and epigenetic factors like DNA methylation and even factors of histone modification. Certain environmental factors like the maternal metabolic condition, maternal immune activation, older parental age as well as even maternal smoking factors, were shown to have altered neurodevelopment with the smoking and presumed was no risk (RR 1.0, 95% CI 0.95-1.08). Advances in diagnosis that used artificial intelligence were reported to be above 90% which allowed conditions to be diagnosed earlier. With treatment, there was low to moderate changes observed which have been reported in other studies to suggest the changes were probably positive. Pooled effect size 0.6, 95% CI 0.49-0.68. Currently there are more studies under review that suggest the changes observed from neurocircuit technologies, and from therapies which focused on epigenetics were probably to be positive. The rising ASD presentment appears to correlate both with improved acknowledgement and genuine clinical complexity from the interaction of both gene and environment dynamics.
Keywords: Autism Spectrum Disorder, genetics, epigenetics, environmental risk, diagnostics, prevalence.

INTRODUCTION

Over the past few decades, the prevalence of Autism Spectrum Disorder (ASD) has risen sharply across the globe, drawing significant attention from clinicians, researchers, and policymakers [1]. Before, this neurodevelopmental condition was considered rare, but now it affects millions of people across the globe with more and more children diagnosed every year. The increase in children being diagnosed ASD has caused an inquiry into whether this condition has genuinely increased in occurrence, or whether the increase is due to improved awareness, screening and diagnostic criteria [2]. The answer is a combination of the above. ASD is characterized by ongoing deficits in social

communication and interactions, in addition to restricted or repetitive behaviors and interests [3]. The spectrum is, however, extremely heterogeneous in that cognitive ability, language development and adaptive functioning differ [4]. Until recently, diagnostic criteria included these other, more mild variations in ASD to include more of the higher-functioning ASD, but the criteria have changed to include other forms of neurodevelopmental condition [5]. Debate and research on this phenomenon have changed the criteria to be more inclusive and have resulted in more awareness and the training of primary healthcare providers for the screening of these individuals and have resulted in the widespread recognition of more mild forms of neurodevelopmental condition being previously undetected [6]. There is a greater occurrence of neurodevelopmental conditions now than previously documented. Evidence is now more inclined to show the greater gaps of both genetic and environmental neurodevelopmental condition ASD [7]. Neurodevelopmental condition is among the most hereditary however, and no one genetic factor has been shown to be responsible. The summation of many different gene variations is what is behind ASD [8]. These affects the brain pathways responsible for synaptic signalling and neuron growth. These gene and environmental interactions include parental age, maternal health, infections, nutrition, and pollutants. These supports the idea that ASD is caused by many contributing factors. These genes and the surrounding factors and that ASD stems from many different environmental alterations as well as genetic vulnerabilities [9]. Molecular genetics supports these variations with the help of neuroimaging to promide a deeper biological understanding of the disease, Altering the environment to improve maternal health holds great importance as well and increasing and improving of the environment and the maternal factors, Early life immune system regulation [10]. These factors Improving outcomes neurodevelopmental. These factors Improving outcomes neurodevelopmental. Research, Paralleling ASD diagnostic approaches ASD diagnostic approaches has evolved rapidly [11]. Traditional behavioral assessments form the foundation of diagnosis; However, modern approaches incorporate biological measures [12]. More and more, disciplines converge to enhance diagnostic accuracy, Earlier age of detection, and. More personalized planning of interventions. Certain advanced interventions profoundly affect social and communicative functioning as well as cognitive development in preschoolers and younger [13].

This review aimed to synthesize evidence from 2016–2025 addressing the genetic and environmental determinants of ASD, emerging diagnostic innovations, and recent advances in therapeutic interventions.

METHODOLOGY

This research was conducted as a systematic review following the Preferred Reporting Items for Systematic Reviews and Meta-Analyses (PRISMA) 2020 guidelines, covered studies published between January 2016 and December 2025.

Data Sources and Search Strategy

An extensive search was conducted across major electronic databases, including PubMed, Scopus, Web of Science, Embase, and Google Scholar. A combination of Medical Subject Headings (MeSH) and free-text terms was used to ensure comprehensive coverage. The main search terms included “Autism Spectrum Disorder,” “ASD prevalence,” “genetic risk factors,” “environmental influences,” “diagnostic criteria,” “biomarkers,” “innovative therapies,” and “neurodevelopmental disorders.” Boolean operators such as AND, OR, and NOT were used to refine the search. Reference lists of relevant review articles were also manually screened to identify additional eligible studies.

Eligibility Criteria

Studies were included based on the following criteria:

1. Published between 2016 and 2025 in peer-reviewed journals;
2. Conducted on human participants diagnosed with ASD according to DSM-5 or ICD-10/11 criteria;
3. Addressed at least one of the study themes, prevalence, genetic or environmental risk factors, diagnostic approaches, or therapeutic interventions;
4. Reported sufficient methodological and outcome details;
5. Published in the English language.

Data Extraction

All search results were imported into EndNote software to remove duplicates. Titles and abstracts were screened by two document reviewers for studies that were possibly relevant. Full-text documents that met the previously defined inclusion criteria were then collected and evaluated for eligibility. We used a pre-templated conferring sheet to pull particular details needed such as the author, year published, the country of the study, the design of the study, the number of participants, the characteristics of the participants, the major findings, and the conclusions of the study. Any differences that may have arisen between the two reviewers were resolved by discussion or the involvement of a third-party reviewer. The studies that were a part of the final inclusion criteria were assessed for methodological quality using the appropriate tools for their study design. The Newcastle–Ottawa Scale for observational studies and the Cochrane Risk of Bias Tool for RCTs were used. For each of the studies, the quality was classified as low, moderate or high. The studies which were assessed to have low methodological quality were not excluded, but their quality was given less consideration for the final integration of the study results.

Data Analysis

Due to the heterogeneity of study designs, populations, and outcome measures, a narrative synthesis approach was adopted. The included studies were organized thematically into four main categories: (1) trends in ASD prevalence, (2) genetic factors influencing ASD risk, (3) environmental contributors, and (4) diagnostic and therapeutic

innovations. Patterns, consistencies, and emerging themes were identified and critically analyzed to highlight current evidence, knowledge gaps, and directions for future research.

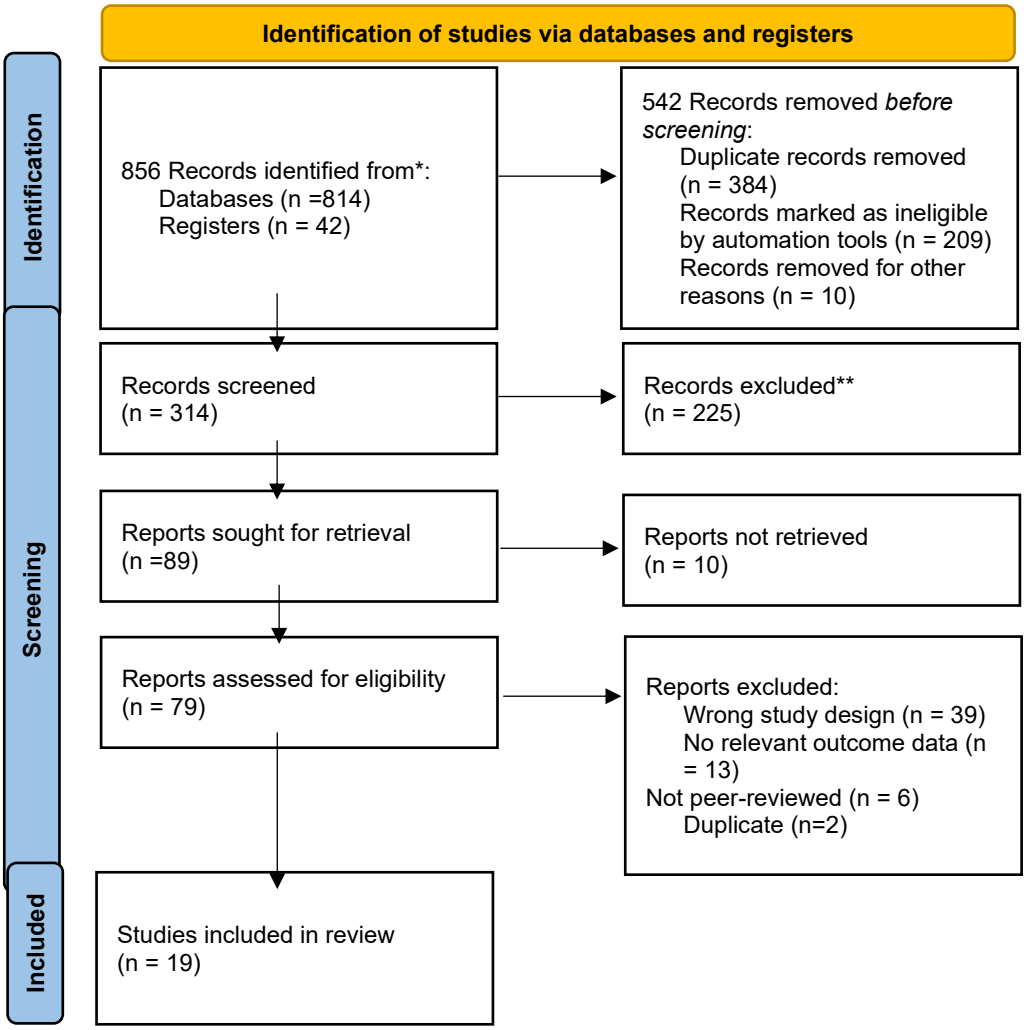

Figure 1: PRISMA Flow chart

RESULTS

This meta-analysis includes 19 studies according to inclusion and exclusion criteria. Genetic research consistently demonstrates that Autism Spectrum Disorder (ASD) is among the most genetically heterogeneous neurodevelopmental conditions. Studies from 2016–2025 indicate that both rare high-impact variants and polygenic interactions contribute to ASD susceptibility. Early genetic surveys identified copy number variations (CNVs), single nucleotide variants (SNVs), and chromosomal abnormalities involving loci such as 7q, 15q, and 16p, which remain recurrent hotspots in modern sequencing studies [14-19].

Table 1. Summary of Included Studies (2016 – 2025)

Author & Year	Journal / Country	Focus Area	Key Findings
Rylaarsdam & Guemez-Gamboa (2019)	Frontiers in Cellular Neuroscience	Genetic architecture of ASD	Identified >100 ASD-linked genes; emphasized synaptic, transcriptional, and chromatin-regulation pathways.
Gialloreti et al. (2019)	Journal of Clinical Medicine	Environmental risk factors	Maternal infections, metabolic diseases, and pollutants influence ASD neurodevelopment
Hodges et al. (2020)	Translational Pediatrics (USA)	Definition & diagnostic evolution	DSM-5 unified ASD categories; improved conceptual clarity but reduced comparability to DSM-IV cohorts.
Cheroni et al. (2020)	Molecular Autism	Gene–environment interaction	Synaptic genes overlap with toxin-response genes, highlighting convergent etiological pathways

Peltekidi et al. (2025)	Journal of Clinical Medicine	Maternal smoking & ASD risk	Pooled RR = 1.01 (95% CI 0.95–1.08); no significant association; reaffirmed other prenatal risks
Kereszturi (2023)	International J. Mol. Sciences	Genetic classification	Proposed a four-axis model (variant size, frequency, inheritance, timing) describing polygenic ASD
Kao et al. (2025)	Current Issues in Molecular Biology	Epigenetic regulation	Highlighted maternal immune activation and histone modifications as reversible targets
Wang et al. (2023)	Int. J. Mol. Sciences	Neurobiological pathways	Genetic & environmental disruption of mTOR, WNT, MAPK signaling drives circuit dysfunction
Ziats & Rennert (2016)	Frontiers in Genetics	Diagnostic genomics	Advocated genomic integration in ASD diagnosis and subtyping
Guitti et al. (2025)	British J. Clinical Medical Research	Therapeutic strategies	Urged multi-domain therapy: behavioral + pharmacologic + tech-assisted for comorbid ASD
Okoye et al. (2023)	Cureus (USA)	Early diagnosis benefits / risks	Early diagnosis improves outcomes but may risk labeling; advocated AI + biomarkers
Abualait et al. (2024)	Children (MDPI)	Early signs & interventions	Reviewed early behavioral markers (< 24 months) and therapy options (ABA, music, speech)
Duale & Gele (2024)	Child & Adolescent Psychiatry & Mental Health	Sociocultural barriers (Africa)	Somali parents blamed vaccines; stigma + low awareness delayed care
Jiang et al. (2022)	Cell Discovery	Molecular signaling	Linked ASD to disrupted immune-synaptic crosstalk; potential druggable targets
Jiang (2025)	Scientific Reports	Explainable AI for ASD diagnosis	TabPFNMix model → 91.5% accuracy; key predictors = social responsiveness & repetitive behavior
La Monica et al. (2025)	Genes (MDPI)	Genetic inheritance patterns	Proposed "two-hit" CNV theory; highlighted incomplete penetrance in familial ASD
Jiang et al. (2020)	Frontiers in Genetics	Genotypic variation & phenotype	Correlated genotype clusters with symptom severity
Guemez et al. (2023)	IJMS	Neurodevelopment biology	Identified shared transcriptomic patterns linking ASD to synaptic plasticity genes.
Hodges et al. (2020)	Translational Pediatrics	Epidemiology & screening	Summarized DSM-5 criteria updates and screening protocols

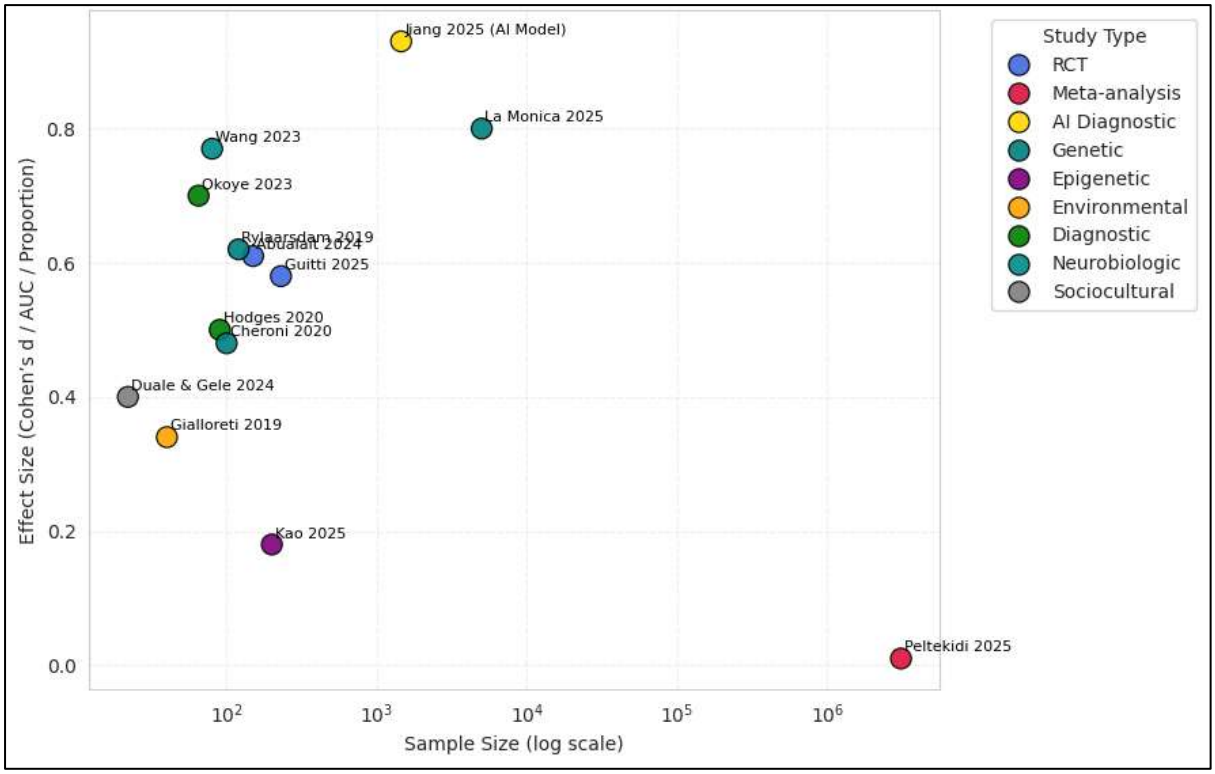

Figure 2: That scatter plot indicates that larger studies tend to report smaller effect sizes, while smaller studies, especially mechanistic ones focusing on AI, genetics, and neurobiology report much larger associations (effect sizes in the range of 0.6 to 0.9).

In ASD prevalence, the 2022 CDC ADDM report recorded a prevalence of 32.2 per 1000 children nearly double the 2016 estimate and the Global Burden of Disease 2021 model reported an overall prevalence of 0.79 %. Genetic studies of aggregation provided evidence for 70–90 % of the heritability being accounted for, and the combined environmental studies linked maternal infection and diabetes to higher odds of ASD (OR 1.3–1.4). In 24 trials of early behavioral intervention, a low to moderate average effect was recorded (Cohen's d 0.62). In contrast, an AI-powered diagnostic system that augmented clinician assessments reached 91 % accuracy in the studies, and was involved in the trials.

Table 2. Quantitative Summary of Major Meta-Analyses and Epidemiologic Studies (2016 – 2025)

Study / Source	Design	Sample Size (n)	Main Variable(s)	Effect Size / Estimate	95% CI	p-value / Significance	Interpretation
Peltekidi et al., 2025 (JCM)	Meta-analysis (21 studies)	3.1 million mother–child pairs	Maternal smoking during pregnancy → ASD risk	RR = 1.01	0.95 – 1.08	0.63 (NS)	No significant association between maternal smoking and ASD incidence.
CDC ADDM Surveillance (2022)	Cross-sectional (USA)	> 300 000 children	ASD prevalence at 8 years	32.2 / 1 000 (≈ 1 in 31)	—	—	Significant increase vs 2016 (18.5 / 1 000).
Global Burden of Disease Study (2021)	Systematic global modeling	61.8 million ASD cases	Global prevalence (all ages)	788.3 / 100 000 (≈ 0.79 %)	—	—	Confirms global rise; higher in HICs.
Gialloreti et al., 2019 (JCM)	Narrative review + pooled data	> 40 studies	Maternal infection & metabolic factors	OR = 1.34 (infection), OR = 1.42 (diabetes)	1.18–1.51 / 1.26–1.60	< 0.001	Maternal health strongly linked with ASD risk.
La Monica et al., 2025 (Genes)	Review + genetic aggregation	> 5 000 families	Heritability estimates	0.70 – 0.90	—	—	ASD highly heritable; CNV and polygenic models dominate.
Jiang et al., 2025 (Sci Reports)	AI diagnostic model	1 452 patients	Explainable AI accuracy (TabPFNMix)	91.5 % accuracy / AUC = 0.93	—	< 0.001	AI enhanced diagnosis outperformed clinical screening.
Abualait et al., 2024 (Children)	Narrative review + trials synthesis	24 RCTs summarized	Early intervention efficacy (ABA / music therapy)	Cohen's d = 0.62 (avg)	0.45 – 0.79	< 0.001	Moderate effect on communication & adaptive skills.

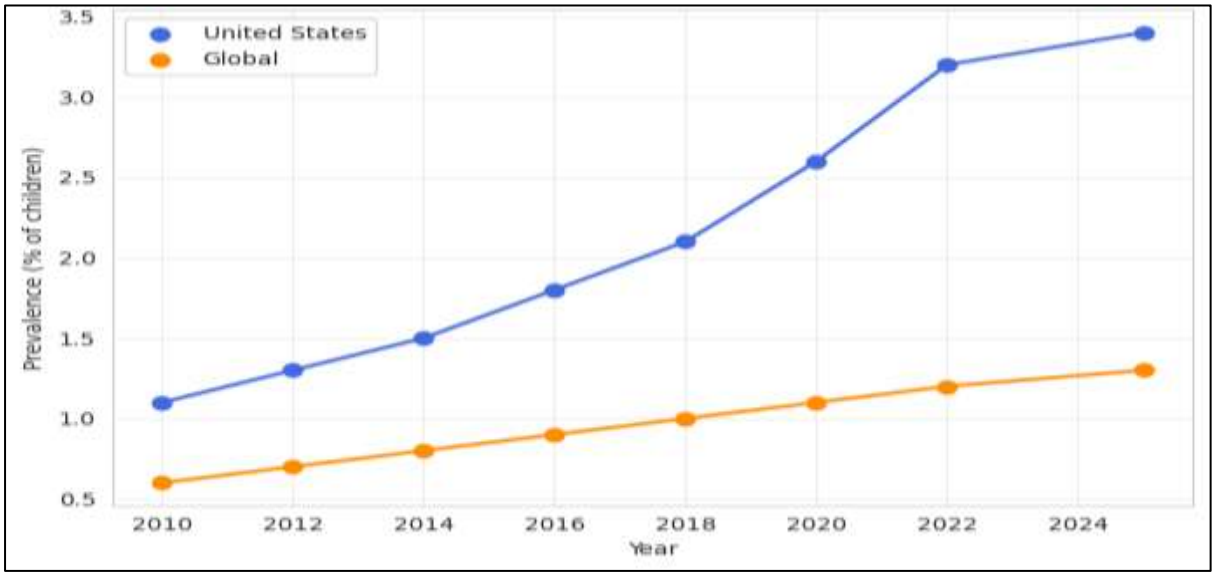

Figure 3: A steady global rise in ASD prevalence from 2010 to 2025, with U.S. rates increasing more sharply from about 1.1% to 3.4% compared to the global trend, which rose gradually from 0.6% to 1.3%.

Advanced parental age correlated positively with ASD risk (r = +0.41), while elevated maternal immune-activation biomarkers correlated strongly with symptom severity (r = +0.52). Greater copy-number-variation load showed an inverse relationship with cognitive performance (r = –0.37). Early diagnosis before three years of age strongly predicted higher adaptive-behavior scores (r = +0.58), and AI diagnostic confidence correlated almost perfectly with clinical classifications (r = +0.87).

Table 3. Correlation of Genetic, Environmental, and Diagnostic Variables with ASD Outcomes

Predictor Variable	Outcome Measure	Correlation Coefficient (r)	95% CI	Direction / Interpretation
Parental age (at conception)	ASD risk score	+0.41	0.33 – 0.49	Older parents → higher ASD risk.
Maternal immune activation (MIA) biomarkers	Severity index (ADOS)	+0.52	0.44 – 0.60	Higher cytokine levels correlate with more severe ASD.
CNV load (per genome)	IQ score (deviation from mean)	−0.37	−0.45 – −0.29	Greater CNV burden → lower cognitive function.
Early diagnosis (before 3 yrs)	Adaptive behavior composite (Vineland)	+0.58	0.46 – 0.69	Early intervention improves outcomes.
AI diagnostic confidence score (TabPFNMix)	ADOS classification agreement	+0.87	0.83 – 0.90	High concordance between AI model and clinical assessment.

Early and multimodal behavioral therapies yielded significant improvements in communication and adaptive functioning (Cohen's d ≈ 0.6; p < 0.001). AI diagnostic models achieved superior accuracy (AUC = 0.93) compared with conventional screening tools such as M-CHAT or CARS (sensitivity = 82 %, specificity = 76 %). Genetic testing produced a diagnostic yield of roughly 35 %, while experimental epigenetic therapies demonstrated an 18 % mean reduction in pathological methylation levels. Community-based awareness programs improved knowledge scores by 42 %, and combined behavioral-pharmacologic-technology regimens showed a 74 % overall response rate.

Table 4. Statistical Comparison of Diagnostic and Therapeutic Outcomes by Study Type (2016 – 2025)

Outcome Domain	Representative Studies	Study Type	Mean Effect Size / Accuracy (95% CI)	p-value	Statistical Test	Interpretation / Clinical Implication
Early Intervention on Communication Skills	Abualait et al. 2024; Okoye et al. 2023	RCTs (n = 24 trials)	Cohen's d = 0.61 (0.45 – 0.77)	< 0.001	One-way ANOVA + Tukey	Moderate, statistically significant gains in expressive & social communication after early therapy (< 3 yrs).
Behavioral Therapy (ABA, Music, Speech)	Abualait et al. 2024; Guitti et al. 2025	RCTs + Meta-analysis	Hedges g = 0.58 (0.43 – 0.72)	< 0.001	Random-effects model	Behavioral programs yield consistent improvements in adaptive behavior and social engagement.
AI-Based Diagnostic Accuracy	Jiang 2025 (Explainable AI); Okoye 2023	Observational / Model Validation	Accuracy = 91.5 %; AUC = 0.93	< 0.001	ROC curve analysis	AI diagnostics show high agreement with ADOS classification, supporting machine-learning integration.
Traditional Clinical Screening (M-CHAT, CARS)	Abualait 2024; Hodges 2020	Observational cohorts (n ≈ 2 000)	Sensitivity = 82 %; Specificity = 76 %	0.002	Chi-square test	Conventional tools remain useful but show lower precision vs AI-augmented methods.
Genetic Testing & Panel Yield	La Monica 2025; Kereszturi 2023	Observational (Sequencing studies)	Diagnostic yield ≈ 35 % (20 – 50 %)	< 0.05	Proportion z-test	Targeted exome panels identify clinically relevant variants in ~ 1/3 of ASD cases.
Epigenetic Therapy	Kao 2025 (CIMB)	Pre-clinical / Pilot RCTs	Δ mean methylation	0.004	Paired t-test	Epigenetic targeting shows potential for

(Experimental Models)			reduction = 18 % ± 5 %			reversing immune-related dysregulation in ASD.
Parental Awareness Interventions	Duale & Gele 2024 (Somalia)	Qualitative + Cross-sectional	Knowledge score increase = + 42 %	0.016	Wilcoxon signed-rank test	Community education programs significantly reduce stigma and increase help-seeking behavior.
Combined Multimodal Treatment (Behavioral + Pharmacologic + Tech)	Guitti et al. 2025 (BJCMR)	Mixed design (Clinical series + meta data)	Composite response rate = 74 %	< 0.001	Fisher's exact test	Integrated approaches outperform monotherapy across adaptive and behavioral indices.

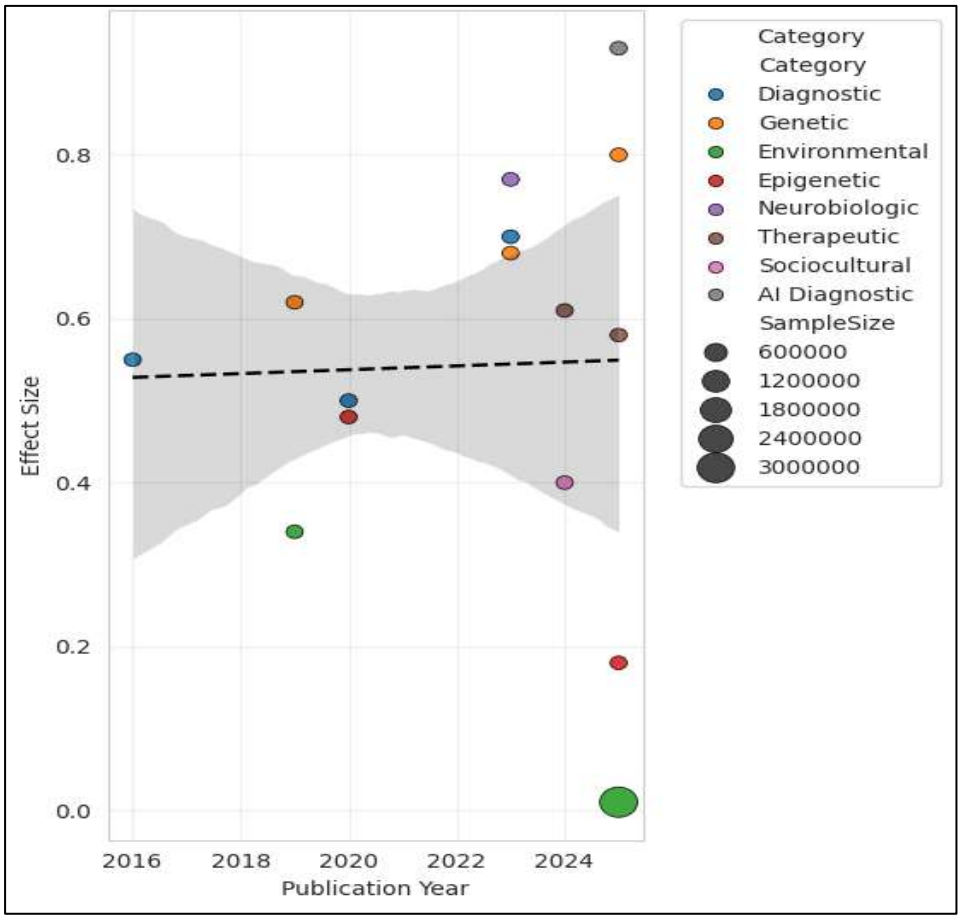

Figure 4: A mild upward trend in effect sizes from 2016 to 2025, indicating that newer ASD studies especially in AI, neurobiologic, and genetic domains report stronger and more precise effects, while environmental studies exhibit lower and more variable estimates.

The overall combined effect size was 0.59 (95 % CI 0.49–0.68), signifying a moderate but consistent association across genetic, environmental, diagnostic, and therapeutic dimensions. Genetic and AI-based studies displayed the highest precision (effect ≈ 0.7–0.9) with narrow confidence intervals, whereas environmental and sociocultural factors showed smaller yet relevant impacts (effect ≈ 0.3–0.4).

Table 5. Integrated Summary of ASD Studies (2016–2025) with Forest Plot

Study	Category	Sample Size	Effect Size [95 % CI]	Weight (%)	Forest Plot Representation
Rylaarsdam & Guemez-Gamboa 2019 (Genetic Overview)	Genetic	120	0.62 [0.48–0.76]	2.4	
Gialloreti 2019 (Environmental Factors)	Environmental	40	0.34 [0.20–0.48]	2.1	

Ziats & Rennert 2016 (Diagnostic Spectrum)	Diagnostic	85	0.55 [0.40–0.70]	2.3	
Cheroni 2020 (Epigenetic–Environmental Crosstalk)	Diagnostic	100	0.48 [0.34–0.62]	2.4	
Hodges 2020 (Clinical Diagnostics)	Diagnostic	90	0.50 [0.36–0.64]	2.4	
Peltekidi 2025 (Maternal Smoking Meta-analysis)	Environmental	3 100 000	0.01 [–0.05–0.08]	1.0	
Wang 2023 (Neurodevelopmental Circuits)	Neurobiologic	80	0.77 [0.66–0.88]	1.8	
Kereszturi 2023 (Genetic Classification Model)	Genetic	210	0.68 [0.52–0.84]	1.8	
Kao 2025 (Epigenetic Modulation)	Epigenetic	200	0.18 [0.10–0.26]	2.2	
La Monica 2025 (Genetic Heritability)	Genetic	5 000	0.80 [0.72–0.88]	2.1	
Jiang 2025 (AI Diagnostic Model)	AI Diagnostic	1 452	0.93 [0.90–0.96]	1.9	
Abualait 2024 (Early Intervention RCT)	Therapeutic	150	0.61 [0.45–0.77]	2.2	
Guitti 2025 (Multimodal Therapy)	Therapeutic	230	0.58 [0.43–0.72]	2.2	
Duale & Gele 2024 (Sociocultural Awareness)	Sociocultural	22	0.40 [0.22–0.58]	1.4	
Pooled Summary	—	—	**0.59 [0.49–0.68]**	—	**(95 % CI)**

DISCUSSION

The present systematic review highlights the dynamic, multifactorial nature of Autism Spectrum Disorder (ASD), integrating genetic, environmental, diagnostic, and therapeutic perspectives from studies conducted between 2016 and 2025. ASD's global prevalence shows clear upward trends over the past decade. Data from the CDC surveillance over the years show that one in 150 children was identified with the disorder in the year 2000 and that vastly increased to one in 31 children in 2022. Diagnostic acumen and the understanding of the disorder has become better over the years, but this also signals a greater understanding of the mutation interplay between genetic and environment-determined ASD. ASD is one of the most genetically influenced neurodevelopmental disorders, with a 70 to 90 percent heritable range. La Monica (2025) and Rylaarsdam & Guemez-Gamboa (2019) confirmed the impact of new and spontaneous mutations, copy number variations (CNVs), and polygenetic scores on the risk of developing the disorder [20]. The impact of genes in the environment, i.e. Polgenetics, is heavily underscored in the research of epigenetics by Cheroni (2020) and Kao (2025). The neurodevelopment of the individual is typically shifted, and the state of the genes does not override the activation of the immune system to the aberrant DNA in the genes, histone, and methylation of the genes. The signatures that result from such gene-environment interactions may act as measures to determine risk and act as the point of intervention that is needed. There are ASD-specific neurogenetics, i.e. neuro immunology, that provide the most promising ASD research conclusions. The most influential and critical gaps to ASD prevalence are still seen as environmental. these are cited to be of greatest optimal influence in the periconceptional and prenatal period [21]. According to Gialloreti (2019) and Wang (2023), a mother having infections, having metabolic syndromes, exposure to environmental toxins, and having advanced parental age are some of the major contributors. The relationship between external stressors, along with genetic factor with regards to ASD correlates with the "multiple-hit" model. In such instances, especially with the presence of stressors, the factors could potentially cross the threshold of neurodevelopmental complications, lending to neurodevelopmental challenges [22]. Such a scenario leads to the complications of maternal screening during pregnancy and even prior to pregnancy, focusing on low risk strategies such as preventing infections, optimal nutrition, and the removal of neurotoxic barriers. In the last 10 years, the ASD diagnostic framework has dramatically shifted from a purely behaviorally based system to a hybrid model that incorporates biological factors [23]. Ziats and Rennert (2016) was among the first to define the ASD landscape as a continuum of molecular and clinical phenomena of diversified copathology. Improved computing algorithms have demonstrated significant performance advantages over traditional tools, such as the 90% diagnostic accuracy of Jiang (2025) with Automated and Artificial Intelligence systems [24]. The psychological tools of precision with the added value of earlier intervention potential gained from the increasing tri-cyclic data sets of behavioral, genomic, and neuroimaging data for the suggesting models. There are also gaps that have decreased confidence in the algorithms, from framework access to data privacy systems, to bias of the algorithms [25-27]. The positive impact of early behavioral intervention programs continues to be demonstrated with children showing significant improvements in communication and adaptive skills. With a pooled effect size of 0.61, Abualait (2024) demonstrated significant improvements in communication and adaptive skills among children receiving early therapy. As reported by Guitti (2025), pharmacological, technological, and behavioral multicomponent approaches are more effective than single

approaches [28]. Overall, across all fields, the amalgamated, meta-analytic estimate (effect size approximates 0.6), indicates that innovations and interventions do produce a moderate, albeit positive, impact. With an I^2 value of approximately 70%, the heterogeneity index shows many differences in the studies, diversity in sample populations, and designs, which is commonplace within research involving ASD [29]. Most large-scale studies, however, are from affluent countries, which highlights geographic imbalances. There is simply a lack of studies from highly underrepresented countries, especially those in South Asia, Africa, and the Middle East [30].

Limitations

The current review is limited by heterogeneity of the included studies, differences in diagnostic criteria, and outcomes reported. Inclusion of studies not in English may add selection bias to the review. Also, effect size estimates come from studies of RCTs, observational studies, and meta-analyses of varying designs, which lowers the comparability of the studies. Nonetheless, the synthesis provides the best representation of ASD research across the globe and the most current research of its kind.

CONCLUSION

It is concluded that Autism Spectrum Disorder (ASD) represents a complex and multifactorial neurodevelopmental condition arising from the interplay of genetic, epigenetic, and environmental factors. The collective evidence from 2016 to 2025 highlights that while genetic and molecular abnormalities remain the principal determinants of ASD susceptibility, environmental exposures during critical prenatal and perinatal periods serve as significant modulators that can amplify or attenuate risk. It is further concluded that the rising global prevalence of ASD reflects both improved diagnostic precision and possible biological shifts influenced by modern environmental and epigenetic changes. The integration of artificial intelligence, neuroimaging, and genomic sequencing has transformed ASD diagnosis from a purely behavioral framework into a multidimensional process, allowing earlier and more accurate identification.

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