



## RESEARCH ARTICLE

# Autistic Disorder (DSM-IV) by Gender/Sex and Brazilian Public Health System: A 10-Year Retrospective Study

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**ABSTRACT**

**Background:** Diagnostic stability in Autistic Disorder (AD) remains a relevant and underexplored topic, particularly in public health systems of middle-income countries.

**Objective:** To investigate whether diagnostic transience of AD, according to DSM-IV criteria, is associated with origin of care within the public, Brazilian Public Health System versus private service (out of pocket, Non-SUS), and gender/sex.

**Methods:** Retrospective analysis of medical records from children evaluated between 2009 and 2020 at a tertiary public pediatric hospital and a private psychiatric office in Minas Gerais, Brazil. Inclusion required a DSM-IV diagnosis of AD for at least 36 months. Statistical analyses included Chi-square, Fisher's Exact, Mann-Whitney, and Kaplan-Meier survival estimates.

**Results:** Among 906 records analyzed, 186 were excluded, 68 had a transience diagnosis (average 9%): 1F:14M in the BPHS group versus 6F:47M in the non-BPHS group. No statistically significant association was found between diagnostic outcome and BPHS/non-BPHS origin or gender/sex ( $p > 0.05$ ). Diagnostic transience was associated with a greater number of consultations (older age).

**Discussion:** Some results were consistent with the literature, such as the higher prevalence of male gender/sex (82.34%), the high frequency of environmental disorders and the median transience percentage (9%) by the 5-year-old median age. The new one is the lack of outcome differences between sex/gender and healthcare systems: BPHS, private, and international literature. The absence of difference in these outcomes is one of the most striking findings, as they differ substantially in access pathways, frequency and intensity of non-medical interventions, and professional specialization in ASD.

**Conclusion:** Diagnostic transience of AD occurred at similar rates regardless of healthcare origin or gender/sex. These findings raise questions about the importance of all biological markers that can complement clinical assessment and identify the most effective therapeutic methods for each individual affected by ASD. Further longitudinal and biologically integrated studies are necessary to clarify predictors of diagnostic stability and long-term outcomes

**Keywords:** Autism, Autism Spectrum Disorder, sex/gender, outcome, diagnostic

## Introduction

Autism Spectrum Disorder (ASD) is a neurodevelopmental disorder that affects social interaction, communication, and behavior. Its symptoms begin in early childhood and more frequently affect boys, with an average ratio of 4:1, following a chronic course. It is known that other medical conditions with developmental delays clinically mimic ASD, making accurate diagnosis difficult. Recent prevalence estimates indicate 1 in every 54 children aged 8 years or younger in the United States<sup>1</sup>.

In everyday life, most professionals are still guided by concepts that are not scientifically grounded, such as the impossibility of diagnostic transience, that autism in women is less frequent and presents more severe clinical conditions, and that better outcomes depend on early diagnosis and treatment.

The transience of diagnosis still appears to be a fact of little relevance, as evidenced by the limited dissemination and exploration of the topic itself, especially in countries such as Brazil, where there are no publications on the subject, nor on the relationship with gender/sex, nor related to the Brazilian Public Health System (BPHS), where the majority of the population is treated.

Proportionally to the thousands of publications on various topics, there are still very few on this subject in international journals, considering that the first was almost 60 years ago, when Rutter et al.<sup>2</sup> reported that 9 out of 63 patients (14%) “could no longer be classified under the term autistic at follow-up.”

The topic is not directly addressed in terms of its importance, a fact reflected in its absence in diagnostic manuals—DSM’s and ICD’s.

The importance of widely disseminating the possibility of diagnostic transience goes beyond the mere transmission of information, reaching at least three dimensions: those of healthcare professionals, the family, and science. In the first group, it challenges the still deeply rooted notion that autism has an

irreversibly negative outcome, thus changing this paradigm. For families, when it occurs, it resolves doubts about the initial autism diagnosis, both for families and for the physicians themselves—a situation that still persists today. For science, it fosters a sense of progress in knowledge and motivates further investigation.

Studies reveal the transience diagnosis presence in themes such as diagnostic mobility, which is multidirectional: 1) migration from AD to PDD-NOS, from PDD-NOS to AD, or from any diagnosis to no diagnosis<sup>3-7</sup>; 2) transience<sup>8</sup>; 3) changes to non-ASD diagnoses, such as Other Developmental Disorders and Reading Disorders<sup>3</sup>; and 6) possible outcome trajectories under DSM-IV<sup>9,3,10-16</sup>. Most studies do not specify the difference between clinical outcomes of Autistic Disorder and Pervasive Developmental Disorder – Not Otherwise Specified (PDD-NOS), the latter being more likely to represent a transient diagnosis<sup>13,14</sup>. Earlier studies suggest that girls present with more severe clinical profiles and poorer outcomes<sup>7,17,18</sup>, suggesting that such severity is secondary to Intellectual Disability (ID). There are also reports of a higher prevalence of epilepsy, which is positively associated with ID<sup>19,20</sup>, regardless of an ASD diagnosis<sup>19</sup>, and that they are more likely to present with de novo mutations<sup>21</sup>, a known cause of ID.

Regarding sex/gender and outcomes, there is very little literature<sup>13,14</sup>. In 2023, Harstad et al.<sup>22</sup> reported that female sex/gender is predictive of a transient diagnosis, based on the Diagnostic and Statistical Manual of Mental Disorders – 5th Edition (DSM-5), without specifying severity levels. Another recent study<sup>23</sup> which specifically investigated the transience of an Autistic Disorder diagnosis clinically equivalent to Level 3 in DSM-5, did not find statistically significant differences between sexes/genders.

The clinical criteria for the diagnosis of Autistic Disorder (AD) – 299.00 – as defined in the Diagnostic and Statistical Manual of Mental Disorders – 4th Edition (DSM-IV)<sup>9</sup> are considered technically reliable and stable over time<sup>10,24</sup> although not for children few

36 months of age<sup>3,8</sup> reported cases of children who were diagnosed only at 36 months, with no identification in earlier assessments. The "gold standard" for diagnostic quality is the clinical judgment of experienced professionals<sup>25</sup> and clinical diagnosis after 36 months of age shows the highest diagnostic stability for AD<sup>12,18,10,26,27</sup>.

The Brazilian Public Health System (BPHS) is a nationally integrated public health system that provides free services universally to anyone within Brazilian territory, regardless of nationality, socioeconomic status, or other factors. Outside the SPSB, there are private health insurance plans and services paid directly out-of-pocket. There is a significant quantitative and qualitative gap in the care provided across these three service types.

Management of the BPHS is municipalized, and at the outpatient level, access to any specialty depends on authorization from a public clinical service located in the patient's residential region, designated as Primary Level. The Secondary Level includes all General Hospitals, while Specialized Hospitals constitute the Tertiary Level. In most Brazilian private health plans, clients can seek a specialist directly, without any authorization.

The primary objective of this study is to identify whether there is a correlation between transient clinical diagnosis of AD according to DSM-IV criteria and: a) sex/gender; b) origin from BPHS and non-BPHS. The secondary objective is to determine whether there is a difference in diagnostic outcome for Autistic Disorder based on age and/or number of visits between patients originating from BPHS and non-BPHS.

The significance of this publication lies in the fact that there are no prior studies examining the relationship between diagnostic transience in autism, sex/gender, and the SPSB, indicating that this topic is underexplored and underfunded.

It is important to clarify that in Brazil, there are diagnostic tests that are not authorized for clinical use

by the Federal Council of Psychology, such as the Autism Diagnostic Observation Schedule (ADOS).

The term sex/gender is used here to reflect the understanding that the effects of biological "sex" and socially constructed "gender" cannot be easily separated, and that the identity of most individuals is shaped by both sex and gender.

## Methods

The study is retrospective, where the medical records of children treated at the Infant Development Ambulatory / Autism Spectrum Disorders of a tertiary public pediatric (Hospital Infantil João Paulo II, Fundação Hospitalar de Minas Gerais- FHEMIG), which participates in the Unified Health Brazilian System (BPHS), and at the author's office, only by the private system (an unknown number of those treated by the private system were receiving non-medical treatments through health insurance), from November 2009 to June 2020 were analyzed.

In both locations, only psychiatric medical attendance was provided, and the other proposed treatments were left to the families' discretion; therefore, there was no control over them. Most patients originated from the capital of the state of Minas Gerais and surrounding cities, a smaller portion from cities throughout the state, and a very small portion from other states. Families/guardians scheduled appointments directly at the hospital, regardless of prior consultation, medical request, or medication use. There were also referrals from the hospital's neurologists and pediatricians. At the time, scheduling was conducted directly at the hospital, meaning it was not yet under municipal service. Scheduling was only available up to 48 months of age. The patients at the private practice also basically had the same residential background.

The diagnosis of AD was clinical, utilizing the DSM-IV criteria both as an "entry" criterion (from 36 months of age) and as a transitioning criterion for the diagnosis. The M-Chat<sup>28</sup> was used from 24 months of age as a triage for the diagnosis while waiting for

the definitive diagnosis. When the diagnosis of AD was positive/BPHS suspicious, families were encouraged to return for clinical follow-up; when not, they were referred to another medical service. There was no active search for patients who failed to return for scheduled appointments, indicating no control over absenteeism.

The data were grouped according to the number of consultations, from the first to the maximum reached by each patient, up to 12 years. The exclusions, "entries", and the transitioned cases of AD clinical diagnosis and the abandonment of the Ambulatory occurred at any of those periods.

Patients who presented with any other primary condition that, by itself, caused delays in child development of any kind were excluded, including Active Early-onset Epilepsies, Prematurity, Cerebral Palsy, Perinatal Injury, Genetic Diseases, Deafness, Bipolar Disorder, Down Syndrome, Blindness, Psychotic Disorders, and Profound Intellectual Deficiency. Patients with BPHS suspected non-AD clinical conditions who were referred to other specialists and did not return were also excluded from the "No diagnosis" classification.

The data were analyzed separately according to BPHS/non-BPHS origin and gender/sex.

#### INCLUSION CRITERIA:

- Children with suspicions of an AD diagnosis in the first consultation up to 48 months of age;
- Children who were diagnosed with AD by DSM-IV criteria at 36 months of age by a senior child psychiatrist, qualified for the task;

#### EXCLUSION CRITERIA:

- Children with a history, or who received a base/primary diagnosis during the ongoing monitoring, with any disorders or syndromes that cause delays in development: children with a history of birth at less than 32 weeks of gestation, weighting less than 1.5 kg, Apgar below 4 at the 5th or 10th minute, morbid events that occurred up to the complete end of the

seventh day of life, chromosomal abnormalities in general, genetic diseases, epilepsy difficult to control by the 2nd year of life, Cerebral Palsy, deafness, blindness, Profound Intellectual Disability, so on;

- Children who presented clinical conditions that did not have a precise diagnosis of another medical disorder that could cause delays in development / differential diagnosis of AD;
- Children who, during follow-up, had psychiatric diagnoses of Bipolar Disorder (BD) and Psychotic Disorder (PD).

#### ETHICAL ASPECTS:

The project was approved under CAAE register No. 49999521.1.0000.5119 (Certificate of Presentation for Ethical Consideration - CPEC), Review No.: 4,861,990 - FHEMIG / Brazil, and no Consent Form was required as it is a retrospective study.

#### STATISTICAL ANALYSIS<sup>32</sup>

The Chi-Square and Fisher's Exact tests were used. The Fisher's Exact test provides an alternative approach to the Chi-Square test when the conditions for its applicability are not met, such as in small samples, or when some cells in the contingency table have very low frequencies, to compare the occurrence of other delays in child development, not AD, between sexes.

To compare the exit of the diagnosis within the variables of interest, two approaches were used: the first consisted of using Chi-Square and Fisher's Exact tests for categorical variables. For numerical variables, the Mann-Whitney test was used to compare the distribution of values between groups. This non-parametric test is suitable for small samples or when the data distribution doesn't follow a normal distribution. For the study of the time until the occurrence of transient diagnosis, the Kaplan-Meier estimator was used. The analyses were performed in R, version 4.3.1. The level of statistical significance considered was  $p < 0.05$ .

## Results

Between November 1999 and June 2020, 906 medical records from the Development Outpatient Clinic were analyzed (Table 1): a) 82.33% were of the male gender/sex, b) 73.65% of the excluded patients were also of the male gender/sex, c) 29.47% were from the BPHS: 38.70% were excluded from the study and 39.24% had some medical condition unrelated to AD with the potential to cause developmental delay, d) eighteen children up to 12

months age at the first consultation, the youngest was 2 months and the oldest was at the 117 months at 20<sup>st</sup> consultation, e) the highest number of consultations was 21 at 109 months of age, f) 9,4% developed Transient Autism until the 21st appointment, g) 86,7% underwent diagnostic transition around the age of 5, h) statistical analyses included Chi-square, Fisher's Exact, Mann–Whitney, and Kaplan–Meier survival estimates.

**Table 1 – Description of categorical variables.**

	BPHS		Não BPHS		Total
	Female	Male	Female	Male	
Total	69	198	91	548	906
Excluded	29	41	20	96	186
Autistic Disorder*	40	157	71	452	720
Permanent Autism	38	144	64	404	652
Transient Autism	2	13	6	47	68
<b>Excluded</b>					
No diagnostic confirmed	6	3	7	17	33
No ASD**	3	2	6	15	26
ASD no AD	1	2	2	21	26
Active Early onset epilepsies	4	8	1	7	20
Prematurity***	-	5	1	13	19
Cerebral Palsy	4	9	-	5	18
Perinatal Injury	3	4	1	9	17
Genic Disease	6	5	-	1	12
Deafness, Bipolar Disorder	2	2	-	3	7
Down S, Blindness, Psychotic Disorder, Profound Intellectual Deficiency	1	3	-	4	8

\*AD: Autistic Disorder (DSM-IV, 299.00); \*\*ASD: Autism Spectrum Disorder;

\*\*\*Prematurity: gestational age below 32 weeks

As shown in Table 2, there was no statistically significant difference ( $p$ -value  $> 0.050$ ) in the percentage of patients with Permanent or Transient autism between gender/sex and BPHS/non-BPHS origin.

Table 2 – Comparison of categorical variables for patients with Permanent or Transient autism between gender/sex and BPHS/non-BPHS origin.

Variable	Stability		Transient		p-value <sup>1</sup>	
	N	%	N	%		
Gender/Sex	Female	107	93.9	7	6.1	0.254
	Male	545	89.9	61	10.1	
BPHS	No	469	89.8	53	10.2	0.361
	Yes	183	92.4	15	7.6	

<sup>1</sup> Chi-square test.

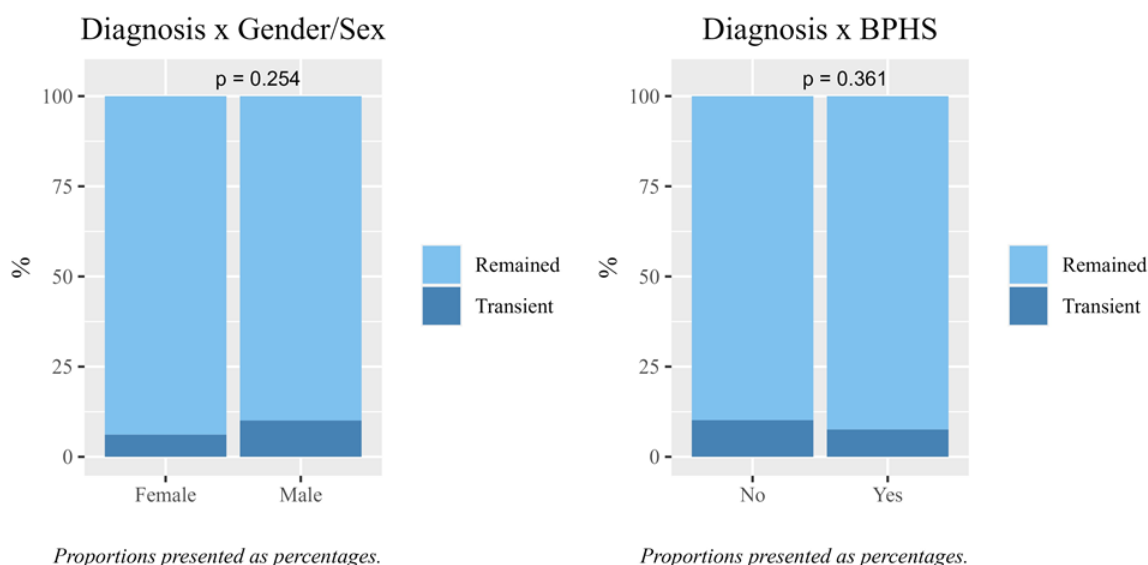


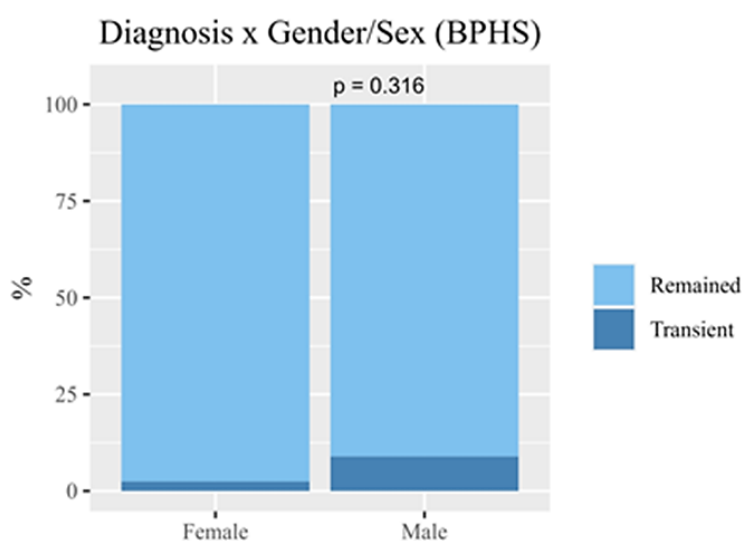
Figure 1 - Comparison of categorical variables for patients with Permanent or Transient autism between gender/sex and BPHS/non-BPHS origin.

There was no statistically significant difference ( $p > 0.050$ ) in the percentage of patients who experienced transient diagnosis in the BPHS group (Table 3).

**Table 3 – Comparison of categorical variables with Permanent or Transient AD patients by gender/sex – only BPHS.**

	Stability Variable	Stability		Transient		p- value <sup>1</sup>
		N	%	N	%	
Gender/Sex	Female	40	97.6	1	2.4	0.316
	Male	143	91.1	14	8.9	

<sup>1</sup> Fisher test.



**Figure 2 - Comparison of categorical variables with Permanent or Transient AD patients by gender/sex – only BPHS.**

Table 4 shows that there were significant differences in the number of consultations (older age) and the transience of the clinical diagnosis between patients of BHPS/non-BHPS origin.

**Table 4 – Comparison of numerical variables between Permanent or Transient autism and number of appointments.**

	Variables	N	Média	D.P.	1°Q	2°Q	3°Q	Valor-p <sup>1</sup>
Inicial age	Stability	652	2,78	0,74	2,23	2,82	3,37	0,143
	Transient	68	2,93	0,62	2,57	2,97	3,40	
Appointments	Stability	652	2,63	2,51	1,00	1,00	3,00	0,000
Appointments	Transience	68	5,52	3,02	3,00	5,00	7,00	

<sup>1</sup> Mann-Whitney Test.

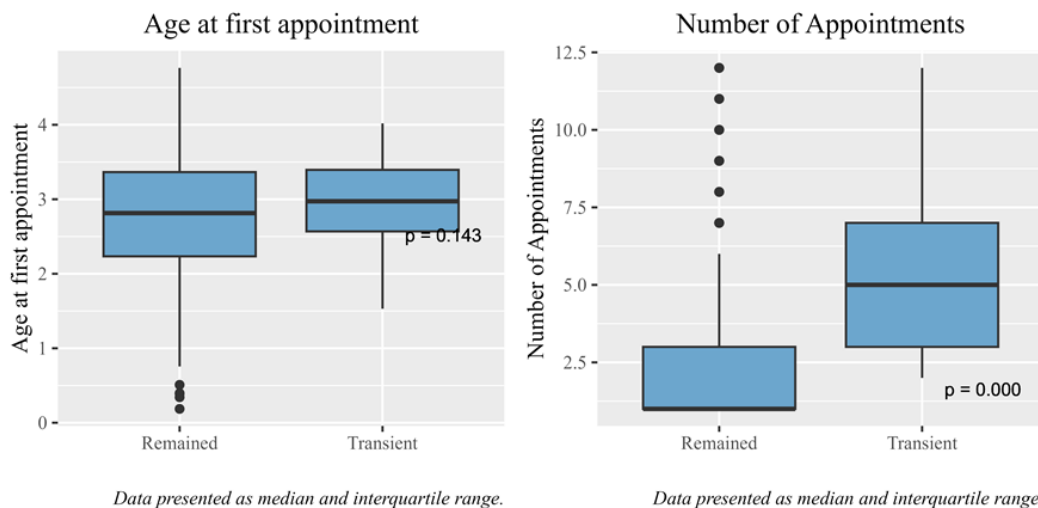


Figure 3 - Comparison of numerical variables between Permanent or Transient autism and number of appointments.

## Discussion

The possibility of clinical transience diagnosis may still be new information for many professionals, both medical and non-medical, as well as for families. This knowledge reduces the belief in the “extreme malignancy” of Autistic Disorder (AD), promotes greater optimism in the therapeutic approach for this population and their families, and strengthens engagement in physician–patient/family relationships, resulting in both technical and humanistic improvement.

Some results were consistent with the literature, such as the higher prevalence of the male gender/sex (82.34%) in the sample, the high frequency of environmental disorders<sup>33</sup>, including prematurity (10.22%), perinatal injury (9.14%), and cerebral palsy (9.68%) and the higher prevalence of AD, age three at the first consultation, Transient diagnosis was associated with the number of appointments, as shown in Table 4, related to older age, 59 out of 68 experienced transient diagnosis up to the 6th appointment, around 55 months of age, which aligns with Lord et al.<sup>10</sup>: “the classifications changed substantially more often from ages 2 to 5 years than from ages 5 to 9 years.”

This study shows the absence of statistically significant difference in AD Transience or Permanence diagnosis by BPHS/non-BPHS origin or by gender/sex (Tables

2 and 3), using DSM-IV as the diagnostic criterion. In the BPHS group, the gender ratio was 1 female to 14 males, versus 6 females to 47 males in the non-BPHS group. The median age at the first appointment was slightly higher in the Transient group (3.35 years) than in the Permanent group (3.06 years), although the difference was not statistically significant (Table 4). This may reflect a delay in seeking or accessing specialized services relative to the literature.

Although no statistically significant differences were found between BPHS/non-BPHS origin or gender/sex, the total frequency of diagnostic transience results is similar across Brazilian health systems, public health insurance, private medicine (median 8%), and international data (median 8.5%)<sup>2,3,5,13,15,34-37</sup>. These contexts have significantly different resources, which is intriguing: BPHS/Non-BPHS patients were mostly from the Metropolitan Region of Belo Horizonte, Minas Gerais, Brazil, and from the international literature. At the time, BPHS provided services were highly deficient, with limited and low ASD expertise, short-term non-medical treatment durations (sometimes 20-minute sessions), low frequency (sometimes monthly), and inconsistent follow-up. In contrast, non-BPHS patients were mostly treated by private professionals selected by families based on known technical competence, or through private health insurance, where provider choice was limited, and the technical quality was only slightly better

than that of BPHS. Meanwhile, international studies typically involved specialized clinics offering the therapies recommended in the scientific literature, with a high standard of care. Given these undeniable differences in treatment quality and quantity, why were the outcomes so similar? This question opens a necessary space for discussion regarding genetics, neurophysiology, psychopathology, current nosology, and treatment approaches related to ASD.

There is also a need for new studies that further explore this subject and develop methodologies for long-term follow-up of transient diagnosis, similar to what was proposed by Margaret H. Sibley et al.<sup>38</sup> for ADHD, a method not applied in the present study.

The strengths of this study include the long follow-up period, significant sample size, diagnosis performed by a senior child psychiatrist using a technically validated tool, as DSM-IV, and the fact that this is the first Brazilian study on the topic conducted within the public health system. The strengths of this study include the long follow-up period, significant sample size, diagnosis performed by a senior child psychiatrist using a technically validated tool, as DSM-IV, and the fact that this is the first Brazilian study on the topic conducted within the public health system, where there is no literature on the topic of this article.

The limitations are: (1) diagnosis performed by only one professional, even though senior in the field; (2) lack of tools such as the ADOS, which the Brazilian Federal Council of Psychology authorizes only for research use; (3) lack of data on functional levels, such as expressive/comprehension language or intelligence; (4) absence of diagnosis in 18% of the excluded cases (33/186), classified as "no confirmed diagnosis," which may have affected the accuracy of excluded case characterization; and (5) lack of active follow-up for all individuals in the database, limiting understanding of part of this population's reality.

This is the first research on the Transience BPHS and Permanence of the Autistic Disorder diagnosis in patients from BPHS.

A novel finding of this study is the statistical similarity in the Transience BPHS Permanence Autism Disorder diagnostic comparing BPHS and non-BPHS patients and across gender/sex. This opens a new window into a scientifically unexplored territory.

## Conclusion

It is particularly noteworthy that similar results have emerged in patients who received treatments that differed both in quality and quantity, whether they were from the Brazilian Public Health System, the private sector, health insurance plans, or those described in international studies.

The results contradict some assumptions: a) women are not affected more severely; b) early diagnosis does not have a decisive impact; c) current treatments may not be as effective as advertised; d) genetic anomalies may have a decisive influence; and e) we need to learn to combine all this information to offer patients truly personalized treatment.

From an outcome perspective, current knowledge in psychiatry, psychopathology, and nosology is insufficient, requiring better differentiation between genetic, immunological, and gestational/postpartum etiologies, and something similar applies to treatments in general. Further studies are necessary to clarify the above-mentioned issues.

### Conflicts of Interest:

None

### IA Use:

For translation

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### Abbreviations:

Autism Spectrum Disorder: ASD

Autistic Disorder: AD

Diagnostic and Statistical Manual of Mental Disorders:  
DSM

Pervasive Developmental Disorder Not Otherwise  
Specified: PDD-NOS

Intellectual Disability: ID

Bipolar Disorder: BD

Psychotic Disorder: PD

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