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ORIGINAL ARTICLE

Beyond the diagnosis of Autism Spectrum Disorder: comorbidities and utilization of health and educational services

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Abstract

Autism spectrum disorder (ASD) has a high prevalence, but there are gaps in the national literature on comorbidities and the use of health and school services. Objective: To describe comorbidities and the use of health and school services in children and adolescents with ASD treated at a basic health unit for pediatric teaching in Rio de Janeiro. Methods: Retrospective, descriptive, exploratory study, with data collected from medical records of children/adolescents treated at a basic health unit for medical teaching in Rio de Janeiro, a partner of the Unified Health System (SUS), from February 2023 to June 2024. Results: The sample consisted of 291 medical records of children and adolescents. The mean age was 82.5 ± 43.9 months; 72.2% (210/291) were male, 67.1% (204/291) were brown/black. They arrived at the outpatient clinic for diagnosis at 60.8 ± 35.2 months; 96.6% of the mothers had incomplete elementary school education. Comorbidities were present in 72.9% (212/291), 79.2% (168/291) had at least 2 comorbidities, with allergies being the most prevalent; mental disorders accounted for 16.4% (63/385). 72.5% had been hospitalized once, the most frequent cause was respiratory diseases. 13.9% (39/281) were not enrolled in school; 86% (141/164) did not have a mediator in the classroom, of which 54.6% (77/141) were unsuccessfully requested. 68.6% (190/277) were without treatment. Conclusion: We observed barriers to access to treatment and more inclusive education in the context of an overloaded SUS, and in which treatment improves the prognosis.

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INTRODUCTION

Autism Spectrum Disorder (ASD) is defined by persistent deficits in social communication and social interaction across multiple contexts, as well as restricted and repetitive patterns of behavior, interests, or activities. These deficits must be present early in development and cause clinically significant impairment in social functioning or other areas of an individual's life.[1]

The prevalence of ASD in Brazil remains unknown, pending the official results of the 2022 national census conducted by the Brazilian Institute of Geography and Statistics (IBGE). Meanwhile, the IBGE estimates that there are approximately two million individuals with autism in Brazil, corresponding to about 1% of the population.[2] According to Manner et al., in a multicenter study conducted for the Centers for Disease Control and Prevention (CDC), the prevalence is one in every 36 children aged 8 years.[3]

Girianelli et al. (2023), when investigating early diagnosis of autism and other Pervasive Developmental Disorders (PDD)—a classification used at the time of the study in the fourth edition of the Diagnostic and Statistical Manual of Mental Disorders (DSM-IV)—in Brazil, found a mean age of 5.5 years for the diagnosis of childhood autism and 6.2 years for atypical autism.[4] In international epidemiological studies, the age at diagnosis ranges between 3 and 5 years.[5] The diagnosis is primarily clinical, as there are no biological markers to support the diagnostic process. Additional barriers to early diagnosis include the heterogeneity of ASD presentations, professionals' level of knowledge, place of residence, family socioeconomic status, recognition of possible signs by families, and the use of diagnostic instruments not always validated in Portuguese.[6]

Comorbidities contribute to the heterogeneity of ASD clinical presentations. Their recognition is particularly important because they require a more detailed and attentive clinical assessment during consultations and may mask an ASD diagnosis, resulting in delayed diagnosis or even a change from a previous ASD diagnosis. Thus, this represents a bidirectional pathway: ASD may be masked by a comorbidity, and awareness that ASD is frequently associated with comorbidities may facilitate the diagnosis of ASD. As a consequence of limited clinical awareness, the most appropriate therapeutic plan may not be implemented, leading to a cascade of further harm to these families and their children.[7,8] Nevertheless, few studies have been conducted in middle- or low-income countries, such as Brazil.[9]

The presence of comorbidities also impacts healthcare services, not only in outpatient settings but also in hospital care. These services are frequently utilized by individuals with ASD, with high rates of hospitalization and outpatient visits described in the international literature; however, little is known about this profile in Brazil.[10] Knowledge regarding the reality of autism in Brazil remains limited, and existing publications are concentrated primarily in the states of Rio Grande do Sul and São Paulo.[11,12]

In light of the above, the objective of this study is to describe comorbidities, healthcare service utilization, and school attendance among children and adolescents with ASD treated at a teaching primary healthcare unit in pediatrics in the city of Rio de Janeiro.

METHODS

This was a retrospective, descriptive, exploratory study of diagnoses of ASD, autism, childhood autism, or Pervasive Developmental Disorders (PDD) in children and adolescents (0 to 18 completed years of age) treated at the Pediatric Outpatient Clinic of a primary healthcare teaching unit affiliated with Faculdade de Medicina Souza Marques, Itanhangá Campus, in the municipality of Rio de Janeiro (RJ), from February 1, 2023 (when the electronic medical record system was implemented) to June 30, 2024. Data were collected from electronic medical records using a search system with the following keywords: "autism," "childhood autism," "ASD," and "PDD," according to the corresponding codes of the International Classification of Diseases, 10th revision (ICD-10).

A search of physical medical records was also conducted for patients previously followed at this outpatient clinic who were seen during the study period. The physical record number is registered in the electronic medical record. All records of children and adolescents (0 to 18 completed years) with a diagnosis of ASD, PDD (included due to changes in diagnostic grouping and nomenclature in DSM-5 replacing DSM-IV), or autism, who were seen during the specified period, were included. Records that were incomplete or not properly filled out were excluded.

The Itanhangá Pediatric Outpatient Clinic is a primary care teaching unit that provides free care without territorial restrictions. It also includes a child neurologist, child psychiatrist, developmental pediatrician, among other specialties. Evaluation by these professionals occurs at a second stage, only after referral by pediatricians. Other services, such as specialty care (except pulmonology, cardiology, and allergy), rehabilitation, laboratory and imaging tests, procedures, and surgeries, are referred to the Unified Health System (SUS) network, as the clinic operates as a SUS-affiliated partner institution.

Although there is no defined catchment area, most patients reside in the Itanhangá neighborhood, located in the western zone of Rio de Janeiro. This region is characterized by marked socioeconomic contrasts, including gated communities and six surrounding communities (predominantly Rio das Pedras, Tijuquinha, Recanto da Barra da Tijuca, Morro do Banco, Vila da Paz, Sítio Pai João, Muzema, and Pedra do Itanhangá). According to the 2022 IBGE census, Muzema and Rio das Pedras are among the nine most populous communities in Rio de Janeiro.[13]

The diagnosis of ASD was established according to the specific guidelines of the Brazilian Society of Pediatrics (SBP), including an extensive medical history, observation of behavior and developmental milestones, administration of screening tools, and detailed physical examination with active investigation of signs and symptoms.[14] DSM-5 diagnostic criteria were also applied, as well as complementary testing when clinically indicated. It is important to emphasize that the diagnostic process is interdisciplinary and involves longitudinal follow-up with appropriate guidance and treatment. Accordingly, pediatricians, the child psychiatrist, child neurologist, and developmental pediatrician at our outpatient clinic participate in the diagnostic process, tailored to the individual child/adolescent and family context.

Some variables require clarification of their definitions. The variable "small for gestational age" (SGA) was defined according to World Health Organization criteria as newborns with birth weight below the 10th percentile for gestational age.[15] The growth curves used followed SBP recommendations.[16] Information regarding "Neonatal Asphyxia" was recorded when documented in the medical record. Additionally, an Apgar score at 5 minutes of less than seven was used as an indicator for investigation of birth conditions.[17] The variable "reason for hospitalization" was classified according to categories of the International Classification of Diseases and Related Health Problems, 10th revision (ICD-10).[18]

The variable "duration of follow-up in the outpatient clinic" was calculated by subtracting the date of the first visit from the date of the last visit. In addition to cases lacking this information, records in which the date of diagnosis coincided with the date of the first outpatient visit (patients diagnosed at their initial consultation) were excluded.

Regarding the variable "Comorbidities," as this was a descriptive study, all identified comorbidities were listed, with grouping limited to phacomatoses (Neurofibromatosis and Tuberous Sclerosis) and gastrointestinal disorders recorded in the charts. For the variable "Treatment," absence of treatment was defined as a minimum of six months without therapeutic intervention. Within this variable, the modality "intellectual rehabilitation" refers to the multidisciplinary therapy provided by the SUS for patients with ASD.

For the variable "enrollment in educational institution," available information in the medical records did not allow clear differentiation between daycare and school attendance for children aged 6 to 7 years. Therefore, all available information was grouped under "daycare/school" to ensure data reliability.

Categorical variables were presented as observed frequency/number of available records (n/N) and percentage. Quantitative variables were described using mean, standard deviation, median, and minimum and maximum values (in parentheses). Data analysis was performed using SPSS version 21. The study was submitted to and approved by the Research Ethics Committee under CAAE number 81133724.5.0000.5239.

RESULTS

The study population corresponded to 8% of the children and adolescents seen at the Itanhangá pediatric outpatient clinic during the study period. Of the 295 medical records of children and adolescents attended with a diagnosis of ASD, 4 records were excluded due to insufficient information. Thus, the final sample comprised 291 medical records of children and adolescents, of whom 79.7% (232/291) were children and 72.2% (210/291) were male (Table 1).

When race/color categories (mixed-race and Black) were grouped, they accounted for 67.1% (204/291) of the sample. In 94.1% (274/291) of cases, mothers had not completed elementary school, with 2.5% (7/291) reported as illiterate (Table 1). The mean age was 82.5 ± 43.9 months, and 50% of the sample were 75 months old (Table 2).

Regarding perinatal history, Table 1 shows that 15.2% (37/243) were born prematurely, of whom 97.3% (36/37) had a history of moderate to late prematurity. In 8.1% (20/248), newborns were small for gestational age (SGA). Neonatal asphyxia was identified in 2.4% (6/250). In 10.7% (29/270), mothers were smokers and/or reported alcohol and/or illicit drug use. Comorbidities were present in 72.9% (212/291), with one comorbidity per individual being the most prevalent (44.8%; 95/212). When considering at least two comorbidities, this prevalence increased to 79.2% (168/212).

This population has been followed in our outpatient clinic for 31 ± 35.3 months (median of 21 months), and in cases diagnosed at the first visit, patients presented to the clinic at a mean age of 60.8 ± 35.2 months (median of 53.5 months) (Table 2).

Figure 1 presents all comorbidities identified in the study. Allergic conditions (rhinitis, asthma, and atopic dermatitis) were the most prevalent, accounting for 30.4% (117/385), 16.6% (64/385), and 6.2% (24/385), respectively. Overweight and obesity combined accounted for 8.6% (33/385); epilepsy/seizures for 6% (23/385); intellectual disability for 4.7% (18/385); and constipation for 3.4% (13/385).

The mental disorders described in Figure 1 (Disruptive Disorder, Psychotic Disorder, Anxiety Disorder, Oppositional Defiant Disorder [ODD], Attention-Deficit/Hyperactivity Disorder [ADHD], social phobia, and intellectual disability), when grouped, accounted for 16.4% (63/385). When constipation was included among gastrointestinal symptoms, the prevalence increased to 4.2% (16/385).

Within the category of genetic syndromes, among the 16 affected individuals, 81.2% (13/385) were still under investigation.

In Table 3, it is observed that 61.5% (174/283) had never been hospitalized, and among those who had been hospitalized, 72.5% (79/109) had experienced one hospitalization. The most prevalent causes were diseases of the respiratory system and perinatal conditions, accounting for 27.7% (38/137) and 24.1% (33/137), respectively.

Table 1. Characterization of the study population – categorical variables.

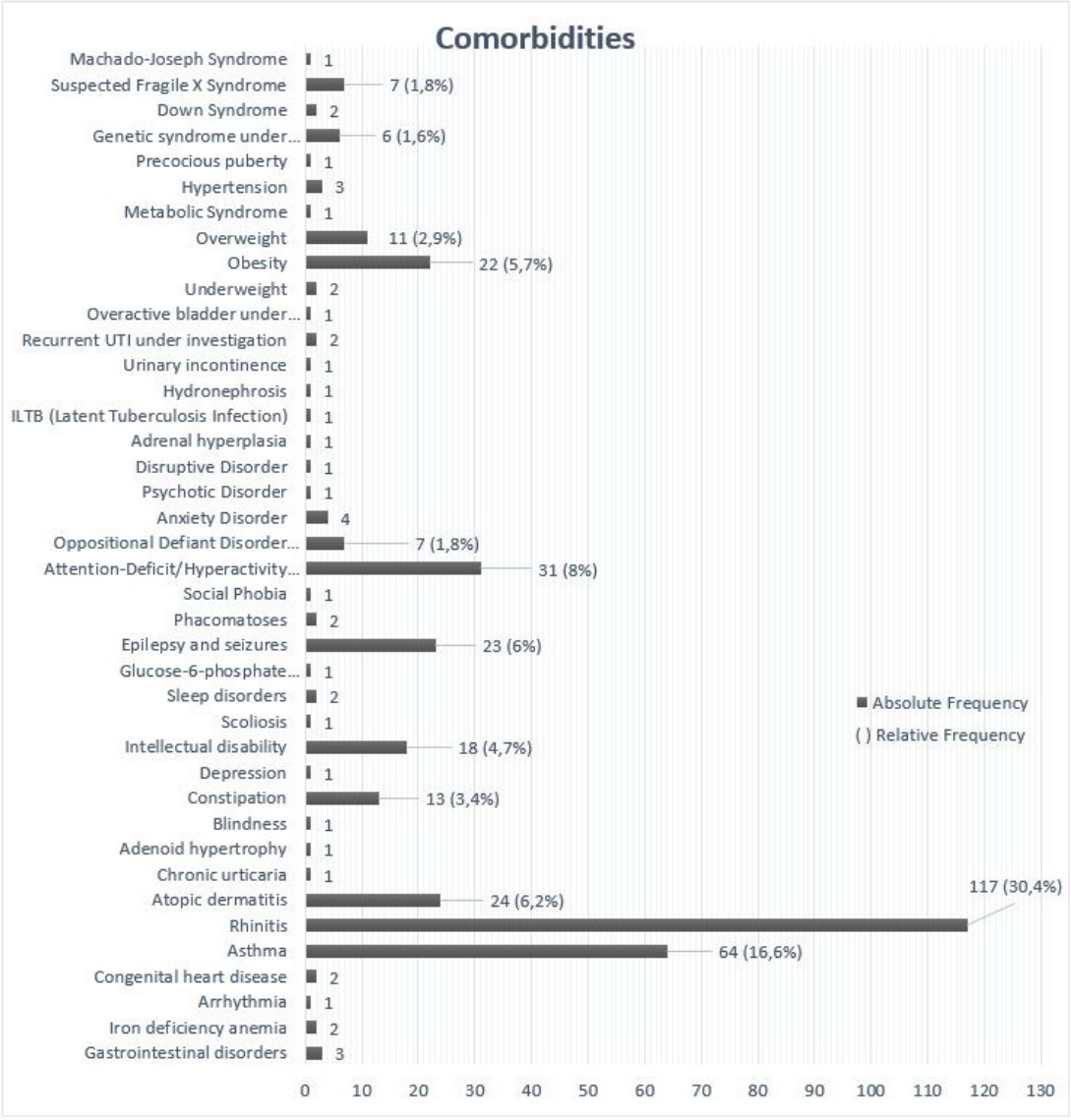
Study Categorical Variables	n/N*	Percentage (%)
Sex		
Female	81/291	27.8
Male	210/291	72.2
Age Group		
Children	232/291	79.7
Adolescents	59/291	20.3
Race/Color		
Mixed-race	195/291	67.1
Black	9/291	3.1
White	87/291	29.8
Maternal Education		
Illiterate	7/291	2.5
Incomplete elementary school	274/291	94.1
Complete elementary school	7/291	2.4
Incomplete high school	3/291	1.0
Perinatal History		
Small for gestational age	20/248	8.1
Prematurity	37/243	15.2
Moderate to Late	36/37	97.3
Severe	1/37	2.7
Maternal Hypertension	69/270	25.6
Maternal Diabetes	21/242	8.7
HIV	3/275	1.1
Neonatal Asphyxia	6/250	2.4
Smoking and/or alcohol and/or illicit drug use	29/270	10.7
Comorbidities		
Presence of comorbidities		
Yes	212/291	72.9
No	79/291	27.1
Number of Comorbidities per Person		
1	95/212	44.8
2	73/212	34.4
3	27/212	12.7
4–5	17/212	8.1

n – number corresponding to the specific variable response;
N – total number of medical records in which the variable was available, regardless of the response, excluding records with missing information;
HIV – Human Immunodeficiency Virus.

Table 2. Characterization of the Study Population – Continuous Variables

Study Continuous Variables	n/N	Mean ± Standard Deviation Median	Minimum – Maximum
Age (months)	291/291	82.5 ± 43.9 75	21 – 206
Duration of outpatient follow-up (months)	239/291	31 ± 35.3 21	1 – 199
Age at first outpatient visit and diagnosis (months)	50/291	60.8 ± 35.2 53.5	21 – 168

Figure 1. Prevalence of comorbidities in children/adolescents with ASD.



Legend: ILTB – Latent Tuberculosis Infection; ODD – Oppositional Defiant Disorder; ADHD – Attention-Deficit/Hyperactivity Disorder; Percentages of 0.3%, 0.5%, 0.8%, and 1% correspond to conditions with absolute frequencies of 1, 2, 3, and 4 cases, respectively.

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Table 3. Characterization of hospitalizations and access to health and educational services.

Study Variables	n/N	Percentage (%)
Hospitalizations		
Yes	109/283	38.5
No	174/283	61.5
Number of Hospitalizations		
1	79/109	72.5
2	18/109	16.5
3–9	12/109	11.0
Reasons for Hospitalizations (137 hospitalizations)		
Perinatal conditions	33/137	24.1
Respiratory system diseases	38/137	27.7
Infectious and parasitic diseases	27/137	19.7
External causes	4/137	2.9
Seizures	4/137	2.9
Surgical conditions	19/137	13.9
Not specified	12/137	8.8
Enrollment in Educational Institution		
Enrolled in school/daycare	242/281	86.1
Not enrolled	39/281	13.9
Unable to secure enrollment despite online registration	12/39	30.8
In daycare	8/12	66.7
In school	4/12	33.3
Classroom Support (Educational Mediator)		
With mediator	23/164	14.0
Without mediator	141/164	86.0
Without mediator despite request	77/141	54.6
Treatment		
Receiving treatment	87/277	31.4
Not receiving treatment	190/277	68.6
Treatment Modality		
Speech therapy and occupational therapy	32/87	36.8
Speech therapy only	13/87	14.9
Occupational therapy only	8/87	9.2
Intellectual rehabilitation	11/87	12.6
Motor physiotherapy	1/87	1.2
Psychology	22/87	25.3
Follow-up with specialist*	157/291	53.9

Legend: n – number corresponding to the specific variable response; N – total number of medical records in which the variable was available, regardless of the response, excluding records with missing information; *Specialist refers to psychiatry, neurology, or developmental pediatrics.

In 86.1% (242/281) of cases, this population was enrolled in an educational institution. Among those not enrolled, 66.7% (8/12) were unable to secure a daycare placement despite registration in the online system (Table 3). In 86% (141/164), classroom learning occurred without an educational mediator; however, among these, 54.6% (77/141) of caregivers had requested school mediation without success. In 68.6% (190/277), children/adolescents were not receiving treatment, and speech therapy combined with occupational therapy was the most prevalent modality, accounting for 36.8% (32/87). Follow-up with specialists is provided exclusively by professionals from our outpatient clinic.

DISCUSSION

Our study highlights a high burden of comorbidities, with at least 79.2% presenting two comorbidities, in addition to a high frequency of hospitalizations due to respiratory diseases and a substantial proportion of children and adolescents without treatment.

The high prevalence of comorbidities observed in this study was primarily driven by allergic conditions. International literature reports a higher prevalence of these diseases among children/adolescents with ASD compared to adults.[19,20] In another recent population-based study, the risk ratio for dermatological and respiratory diseases in children/adolescents with ASD compared to those without ASD was 1.4 and 1.5, respectively.[21]

Comorbidities are particularly relevant in patients with ASD, who may have difficulty verbally expressing symptoms and needs, even among those who are verbal. Altered sensory perception may further hinder symptom localization, complicating diagnosis and potentially leading to underreporting in medical records.[7,22] Therefore, clinicians must remain attentive and proactive in identifying signs and symptoms that may not be recognized by families. It is important to emphasize that healthy children/adolescents with ASD have greater opportunities to learn—at school, in therapy, and in daily interactions with family and peers.[19]

These children/adolescents have a well-established increased risk of obesity, and recent evidence also suggests an elevated risk of cardiometabolic disease.[23] Thus, vigilance regarding physical examination and counseling for healthy lifestyle habits is essential, even in patients who may be difficult to examine and exhibit selective eating behaviors.

Neurological and psychiatric disorders are highly prevalent, affecting 59.6% of children/adolescents with ASD,[8] with ADHD being the most frequent, affecting approximately 1 in 6 children/adolescents.[24] ADHD was also the most prevalent mental disorder in our study; however, mental disorders were not the most prevalent comorbidities overall, as discussed above. In addition to methodological differences, one hypothesis for this discrepancy relates to the greater availability of professionals managing allergic conditions in our outpatient clinic, including pediatricians, allergists, and pulmonologists. Conversely, we do not have a sufficient number of professionals for mental health diagnoses, as only slightly more than half of the children (53.9%) are followed by a child psychiatrist, child neurologist, or developmental pediatrician.

Furthermore, there are significant barriers to referral for child and adolescent neurological or psychiatric care within the municipal network through the National Regulation System (SISREG) or through direct access to Child and Adolescent Psychosocial Care Centers (CAPSi) for those we cannot accommodate. This bottleneck is well documented by the Rio de Janeiro Municipal Health Department, which reports an average waiting time of 354 days for neurology and 160 days for child and adolescent psychiatry.[25] Our data suggest that the issue extends beyond specialty availability to professional qualification, as children/adolescents are arriving for their first ASD diagnosis at approximately 5 years of age.

Perinatal conditions were the second leading cause of hospitalization and may be associated with maternal risk factors and prematurity described in the study. Prematurity is a known factor associated with ASD and may be exacerbated by hospitalization-related procedures.[26] Moderate and late prematurity carry risks of 1.75 and 1.42, respectively, compared to term births, as demonstrated in a recent meta-analysis.[27] Surgical conditions were the fourth most prevalent cause of hospitalization in this study. Supporting this finding, Engelhard (2020) observed a 1.5-fold increased risk of surgical hospitalization among children/adolescents with ASD compared to those with typical development and ADHD.[26]

The pattern of comorbidities reflects the hospitalization profile observed when compared to international literature, which reports a higher frequency of psychiatric hospitalizations in children and adolescents.[27,28] In contrast, we observed a higher prevalence of hospitalizations due to respiratory diseases, consistent with allergic conditions being the most prevalent comorbidities. In Brazil, psychiatric reform—characterized by a reduction in psychiatric hospital beds and expansion of community-based services such as CAPS and CAPSi—may contribute to the lower prevalence of psychiatric hospitalizations.[29] However, factors such as service overload (approximately one-third of CAPSi-RJ consultations involve individuals with ASD), limited hours of care for children/adolescents, and insufficient integration with other SUS services create additional barriers to access.[30]

Hospitalization occurred at least twice in 89% of our population, suggesting that hospitalization may represent an additional opportunity for ASD diagnosis beyond outpatient consultations. An inherent advantage is the extended observation period afforded by inpatient care, enabling comprehensive assessment by the healthcare team.

Beyond outpatient shortages of specialists within the SUS, our study also demonstrates difficulty accessing treatment services. This finding aligns with the reported 336-

day waiting list for pediatric intellectual rehabilitation within the established care pathway.[25] School mediation serves as an important facilitator of learning and social skills. Although we identified a substantial unmet demand in this area, we found no published data addressing this issue.

In conclusion, we observe an overburdened public health system with extensive waiting lists, creating barriers to treatment access and inclusive education, in a context where early and appropriate treatment improves prognosis. ASD emerges as a complex and heterogeneous condition extending beyond diagnosis, requiring attentive, proactive, and compassionate care from healthcare teams, particularly in light of the numerous barriers faced by families.

This study has limitations, including its retrospective design, incomplete information in medical records, and the fact that the population is served within a teaching unit, which may introduce bias. Nonetheless, this initial exploration of the population treated in our outpatient clinic highlights significant gaps not only in understanding ASD beyond diagnosis but also within the national literature, reinforcing the need for further Brazilian studies.

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