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Between Science and Community: development and validation of an educational booklet on early diagnosis of Autism Spectrum Disorder and molecular biomarkers

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Portuguese title: "Entre a Ciência e a Comunidade: construção e validação de cartilha educativa sobre diagnóstico precoce do Transtorno do Espectro Autista e biomarcadores moleculares"

ABSTRACT

Autism Spectrum Disorder (ASD) is a neurodevelopmental condition with an estimated prevalence of 1.1% of the population. In Mato Grosso do Sul, Brazil, more than 29,000 cases are estimated, although many remain underreported due to diagnostic barriers. This study describes the development and validation of an educational guide that integrates scientific communication on salivary biomarkers (microRNAs) with community engagement in translational research. Validation of the educational guide involved 15 health and education professionals and 8 members, formed by caregivers, family members, and adults with ASD. The Content Validity Index (CVI) was used for this purpose. All items achieved a CVI > 0.80. This guide constitutes an important instrument for scientific communication on ASD, democratization of knowledge, and community engagement in research.

Keywords: Autism Spectrum Disorder; microRNAs; Health Education.

RESUMEN

El Trastorno del Espectro Autista (TEA) es una condición del neurodesarrollo con una prevalencia estimada del 1,1% de la población. En Mato Grosso do Sul, Brasil, se estiman más de 29.000 casos, aunque muchos permanecen sin notificar debido a barreras diagnósticas. Este estudio describe el desarrollo y la validación de un folleto educativo que integra la comunicación científica sobre biomarcadores salivales (microARN) con la participación comunitaria en la investigación traslacional. La validación del folleto involucró a 15 profesionales de la salud y la educación, y a 8 miembros, conformados por cuidadores, familiares y adultos con TEA. Para ello se utilizó el Índice de Validez de Contenido (IVC). Todos los ítems alcanzaron un $IVC > 0,80$. El folleto constituye un instrumento importante para la comunicación científica sobre el TEA, la democratización del conocimiento y la participación comunitaria en la investigación.

Palabras clave: Trastorno del Espectro Autista; microARN; Educación en Salud.

RESUMO

O Transtorno do Espectro Autista (TEA) é uma condição neurodesenvolvimental com prevalência estimada em 1,1% da população. Em Mato Grosso do Sul, Brasil, estimam-se mais de 29 mil casos, embora muitos permaneçam subnotificados devido a barreiras diagnósticas. Este estudo descreve o desenvolvimento e a validação de uma cartilha educativa que integra a comunicação científica sobre biomarcadores salivares (microRNAs) com o engajamento comunitário na pesquisa translacional. A validação da cartilha envolveu 15 profissionais da saúde e da educação e 8 membros, formados por cuidadores, familiares e adultos com TEA. Para esse fim, utilizou-se o Índice de Validade de Conteúdo (IVC). Todos os itens alcançaram $IVC > 0,80$. A cartilha constitui um importante instrumento para a comunicação científica sobre o TEA, a democratização do conhecimento e o engajamento comunitário na pesquisa.

Palavras-chave: Transtorno do Espectro Autista; microRNAs; Educação em Saúde.

1. INTRODUCTION

Autism Spectrum Disorder (ASD) is a neurodevelopmental condition characterized by challenges in social communication and restrictive, repetitive behaviors, with a multifactorial etiology involving genetic and environmental components (AMERICAN PSYCHIATRIC ASSOCIATION, 2013). Recent epidemiological data indicate a global prevalence of approximately 1% of the population.

Diagnosis of ASD relies primarily on clinical and behavioral assessment, following the criteria of the Diagnostic and Statistical Manual of Mental Disorders (DSM-5) (LORD et al., 2018). This approach, although well established, has significant limitations, including inherent subjectivity, the need for specialized professionals, and restricted access to assessment services, particularly in smaller or underserved cities (RIBEIRO et al., 2017).

Early detection of ASD is crucial for the timely initiation of interventions, which are associated with better developmental outcomes (GIRIANELLI et al., 2023). According to IBGE, 2.4 million people in Brazil reported an ASD diagnosis, corresponding to 1.2% of the population. In the state of Mato Grosso do Sul, more than 29,000 people are estimated to be on the spectrum (IBGE, 2025). In this context, and in view of regional challenges, the mirTEA Project was conceived and is currently underway in Mato Grosso do Sul. We are a multidisciplinary research group from the Graduate Program in Biotechnology at Universidade Católica Dom Bosco (UCDB).

The mirTEA project seeks to identify salivary microRNAs as reliable biomarkers for early detection of ASD. MicroRNAs are small non-coding RNA molecules that regulate gene expression and have shown promise as biomarkers for several conditions, including ASD (KALEMAJ et al., 2022).

Studies indicate that microRNA profiles in biological fluids, such as saliva, differ between individuals with ASD and those with typical development (HICKS et al., 2019; SEHOVIC et al., 2020). Our mission, however, extends beyond research: we believe that science leads to understanding, understanding generates empathy, and empathy builds connections. By investigating the molecular signatures of ASD, we aim to promote deeper acceptance of neurological diversity. Development of a complementary, objective, and accessible diagnostic tool has the potential to directly impact the reality of families in Mato Grosso do Sul, shortening the long path to a definitive diagnosis.

Communication between the university and society is a persistent challenge, particularly when it involves cutting-edge biomedical research. In the field of ASD, this communication gap is especially pronounced: while families seek immediate answers, researchers investigate

molecular biomarkers whose results take years to yield. How can these different timeframes be reconciled? How can science be translated into hope without generating false expectations? Health education practices are therefore essential to equip families, educators, and professionals to identify early signs of ASD and understand scientific advances in the field (ANDRADE et al., 2025). Appropriate educational materials can strengthen guidance for caregivers in the process of early identification and intervention (COLUCI et al., 2015).

2. OBJECTIVE

This study aims to develop and validate an educational guide on ASD, focused on early diagnosis and on the research on molecular biomarkers conducted by the mirTEA Project in Mato Grosso do Sul.

3. METHODOLOGY

3.1 Study design, period, and location

This is a methodological study conducted between March and October 2025 in Campo Grande, Mato Grosso do Sul, Brazil. The development and validation process of the educational guide followed three stages based on Giordani (GIORDANI, 2020), described below.

3.2 First stage: Development of the guide

This stage consisted of developing the theoretical content and graphic design of the educational guide based on a systematic review of the scientific literature on ASD, microRNAs, and early diagnosis, complemented by the authors' clinical and research experience in the mirTEA project (Olsen, I., & Hicks, 2020 et al., 2020; LEVITSKIY et al., 2021; SEHOVIC et al., 2020). The material was developed considering the sociodemographic profile of the population of Mato Grosso do Sul.

3.3 Second stage: Validation of content, appearance, and language

For validation of the first version of the guide, the material was distributed by e-mail to evaluators, accompanied by a questionnaire developed on the Google Forms platform. Participation in the evaluation of the educational guide was voluntary, and data were treated with confidentiality. This study was approved by the Research Ethics Committee of Universidade Católica Dom Bosco (CAAE: 56241522.9.0000.516).

3.3.1 Selection and recruitment of evaluators

For validation of the guide, specific inclusion criteria were established for each group of evaluators. Professionals were required to hold a complete degree in health or education fields, with a minimum of two years of experience providing care or follow-up to people with ASD, and to work in Mato Grosso do Sul. Family members were required to be direct caregivers or relatives of people with a confirmed ASD diagnosis, aged 18 years or older.

Participants were recruited by convenience, using multiple dissemination strategies. Contact with family members and professionals occurred mainly through local associations and WhatsApp groups dedicated to the topic. Invitations to participate were sent by e-mail, containing a detailed presentation of the research, the educational guide in PDF format, and a link to the evaluation questionnaire developed on the Google Forms platform.

3.3.2 Development of the material evaluation questionnaire

The instrument combined closed-ended fields, intended for objective evaluation, with open-ended questions that allowed participants to make free suggestions, report difficulties encountered, and point out specific disagreements regarding the content, visual presentation, and language of the material. This mixed approach aimed to ensure a comprehensive evaluation, confirming that the guide was attractive and easy to understand for the target audience.

For objective measurement of responses to the closed-ended fields, the Likert scale was used. Developed by Rensis Likert in 1932, this instrument allows respondents to express their degree of agreement with a series of statements, ranging from one extreme to the other (for example, from "strongly disagree" to "strongly agree"). The central position of the scale was defined to reflect unambiguous neutrality, thus avoiding interpretive ambiguity (LIKERT, 1932).

3.3.3 Analysis of the applied questionnaire

Quantitative analysis of the Likert scale responses was performed by calculating the Content Validity Index (CVI). This index measures the proportion of agreement among judges regarding the adequacy of each evaluated item. The CVI was calculated by dividing the number of responses categorized as 1: "strongly disagree," 2: "partially disagree," 3: "neutral," 4: "partially agree," and 5: "strongly agree" by the total number of responses for each item. According to the literature in the field, a minimum CVI of 0.80 was established as the acceptance parameter, with values above 0.90 preferred.

3.3.4 Data processing

Data processing and development of the visualization tools were performed in the RStudio environment (version 2026.6.0.242), using the R language, structured in two sequential scripts. The first script, dedicated to static analysis, employed the `readr` and `readxl` packages for data import; `dplyr`, `tidyr`, and `stringr` for data manipulation and categorization; `psych`, `rstatix`, and `lrm` for statistical analyses; and `knitr` with `kableExtra` for tabulation of results.

The second script focused on building an interactive graphical interface via the Shiny ecosystem (`shinydashboard` and `shinythemes`), integrating `plotly`, `DT`, `forcats`, `scales`, `wordcloud`, and `RColorBrewer` for dynamic rendering and customization of charts and tables, while retaining the manipulation and statistical packages for real-time reactive processing and high-resolution export.

Development and debugging of both codes were optimized using the DeepSeek-V3 model, whose textual externalization of the reasoning process (chain of thought) enabled continuous auditing of programming logic, precise identification of prompt inconsistencies, and rapid correction of syntax errors, function chaining, and package dependencies, ensuring the robustness of the scripts and the stability of the reactive interface.

3.4 Third stage: Author revisions

Disagreements and suggestions identified in the previous stage were analyzed by the authors, and initial adjustments were made to the material. After these modifications, the educational guide was published in e-book format and printed.

4. RESULTS

The educational guide, entitled "Autism Spectrum Disorder in Mato Grosso do Sul: The Science of Early Diagnosis," was developed with accessible language and illustrations. Its content addressed innovative ASD research in the state, the methodology of research on salivary microRNAs, the importance of early diagnosis, an overview of the mirTEA project, and ways to participate in the research.



Figure 1. Cover image of the developed educational guide. This material is freely available in the repository of UCDB Press (Catholic University of Dom Bosco) at <https://site.ucdb.br/a-ucdb/editora/livros>.

The educational material was structured in 11 pages, A5 format, with 6 sections organized in accessible language and 8 colored illustrations. The content included: (1) Innovative ASD Research in Mato Grosso do Sul; (2) How the Research Is Conducted; (3) Why This Research Matters; (4) Meet the mirTEA Project; (5) How to Participate in the Research; (6) References. The material presented recent, previously unpublished IBGE data on the prevalence of ASD in the state of Mato Grosso do Sul. The language adopted avoided excessive technical terms and

prioritized visual metaphors for complex concepts, such as comparing microRNAs to "cellular messengers" and molecular diagnosis to "reading biological clues."

Twenty-three judges took part in the evaluation, divided into two groups: one composed of 15 health and education professionals who work in the care of people with ASD, and another with 8 members, formed by caregivers, family members, and adults with ASD who had the cognitive capacity to understand the material and answer the questionnaire.

The quantitative results, obtained through the Content Validity Index (CVI), reached the following values: 0.88 for the Content domain; 0.84 for the guide overall; 0.90 for Language; and 0.83 for the Visual aspect.

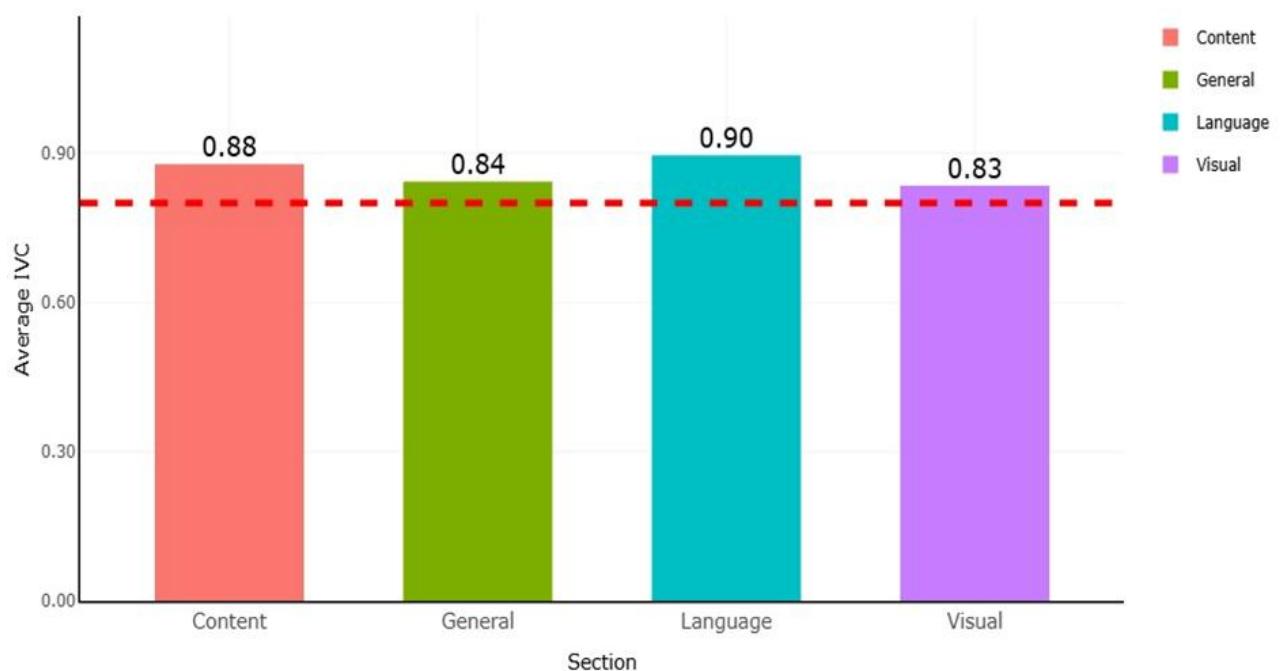


Figure 2. Content Validity Index (CVI) by evaluated item (content, overall, language, and visual).

In the comparison between groups, the mean scores given by professionals were: 4.56 (content), 4.68 (overall), 4.67 (language), and 4.48 (visual). Family members and people with ASD gave mean scores of: 4.20 (content), 3.89 (overall), 4.03 (language), and 4.00 (visual).

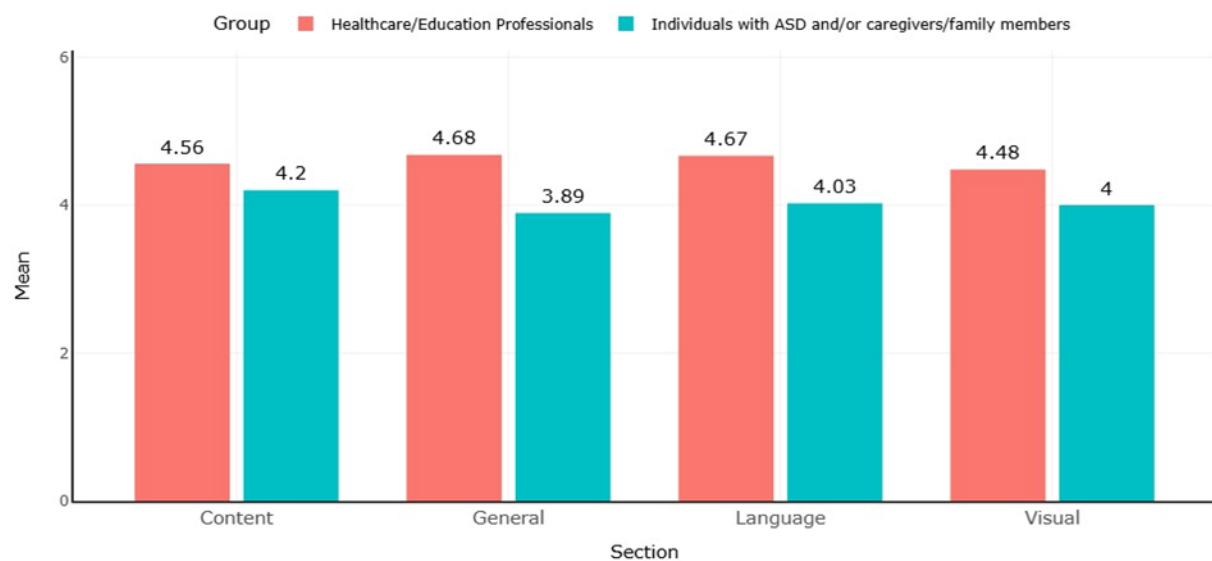


Figure 3. Comparison of mean agreement scores between groups. Red represents the group of health and education professionals who work in the care of people with Autism Spectrum Disorder (ASD); blue represents the group of caregivers, family members, and individuals with ASD.

Qualitative analysis of the open-ended responses revealed the following themes. As strengths, reviewers highlighted the scientific grounding of the content and clarity in addressing the importance of ASD research; accessible, direct, and well-structured language; and an attractive visual design, with appropriate illustrations, balanced colors, and easy-to-read typography.

As suggestions for improvement, evaluators indicated the need to make the language even more objective; to simplify or remove technical terms such as "microRNA"; to expand information on ASD-related care; to reduce the length of the text; and to include an introductory summary of the disorder. After team analysis, the judges' suggestions were not incorporated into the final version of the guide.

Following the validation of the educational guide content and appearance by the experts, the material was made publicly and freely available in the repository of UCDB Press (Catholic University of Dom Bosco), at <https://site.ucdb.br/a-ucdb/editora/livros>. Additionally, 100 printed copies were distributed at events related to ASD, as well as in specialized clinics and partner non-governmental organizations. This dissemination strategy aimed not only to broaden access to the generated knowledge but also to encourage the participation of both the general population and professionals in the field in the study, thereby reinforcing the applied nature of the research.

5. DISCUSSION

The educational guide developed represents an unprecedented educational instrument in the context of Mato Grosso do Sul, aligning scientific communication on ASD with community engagement in translational research. The educational approach to molecular biomarkers for ASD is particularly relevant given the limitations of diagnosis based exclusively on behavioral criteria.

The high Content Validity indices (all above 0.80) confirm the robustness of the material, indicating that it is scientifically grounded, clear, attractive, and appropriate to its informational purposes. The CVI of 0.90 in the Language domain suggests that this was the most appreciated aspect, in line with the literature on public communication of science, which identifies molecular biomarkers among the topics most difficult to translate for lay audiences (MASSARANI & MOREIRA, 2016).

Although professionals' evaluations were numerically higher than those of family members across all domains, both groups assigned mean scores above the cutoff point, attesting to the good acceptance of the material by both specialists and non-specialist audiences. More than a disagreement, this difference in perception reveals a necessary complementarity: while professionals tended to more strongly recognize technical quality and scientific rigor, family members and individuals with ASD (who assigned slightly lower scores) validated the practical applicability and informational relevance of the content, showing that the educational guide met real needs of this group. This participatory approach aligns with the principle "nothing about us without us," central to the autistic self-advocacy movement (NICOLAIDIS, 2012), and contrasts with materials developed exclusively by specialists, which are often criticized for being disconnected from everyday needs (EVERT et al., 2003).

Despite qualitative suggestions to simplify terms such as "microRNA" and shorten the text, the authors chose not to adopt them, based on three considerations: (1) the guide was designed with the specific purpose of informing about this particular research, without the intention of comprehensively addressing ASD, which justifies the absence of an introductory summary of the disorder; (2) the scientific terminology used is essential to ensure the accuracy and reliability of the information, and the high CVI for Language (0.90) itself attests that this choice did not compromise comprehension by the target audience; and (3) the compact, direct structure was intentionally maintained to facilitate retention of the core message, without compromising clarity, as demonstrated by the validity indices. This tension between oversimplifying (losing precision) and maintaining complexity (losing comprehensibility) is a

classic challenge in translating microRNA research for lay audiences, and the successful strategy adopted was the use of visual metaphors.

Finally, the contextualization in Mato Grosso do Sul was intentional: the educational guide includes local epidemiological data and contacts for regional associations. Although this regionalization limits direct generalization to other states, it responds to a common criticism of national educational materials, namely that they fail to address local specificities (SILVA & ALVES, 2018). The validation methodology, however, is replicable, allowing other research groups to adapt the guide to their own contexts while maintaining the validated structure and customizing regional content.

The free dissemination of the guide was a crucial strategy for translating scientific knowledge into a practical and accessible tool. This initiative reinforced the study's objective of not only validating an instrument but also effectively promoting health education about ASD, directly reaching the target population and strengthening partnerships with community institutions.

6. CONCLUSION

The guide showed adequate content validity, integrating health education, scientific communication, and community engagement. Participatory validation demonstrates its accessible language and practical utility, strengthening the link between university and society. Future studies will assess its impact on scientific literacy, while the methodology employed may inspire similar initiatives in other regions.

7. CONFLICT OF INTEREST

The authors declare no conflict of interest.

8. AUTHOR CONTRIBUTIONS STATEMENT (CRediT Taxonomy)

- Júlia Cunha Gonzales: Conceptualization, Methodology, Formal analysis, Data curation, Investigation, Writing – original draft, Writing – review & editing, Visualization.
- Willy Rilston Souza de Jesus: Data curation, Formal analysis, Investigation, Writing – original draft.
- Marcos Fernandes Morales: Data curation, Formal analysis, Software.

- Romulo Diniz de Arruda: Data curation, Investigation, Writing – original draft.
- Alinne Pereira de Castro: Conceptualization, Supervision, Methodology, Writing – review & editing, Funding acquisition.

All authors reviewed and approved the final version of the manuscript and agree with its submission.

9. DATA AVAILABILITY STATEMENT

All data generated or analyzed during this study are included in this manuscript and its supplementary figures. The raw data from the validation questionnaire are not publicly available to preserve participant anonymity, as approved by the Research Ethics Committee of Dom Bosco Catholic University (CAAE: 56241522.9.0000.516).

10. STATEMENT ON THE USE OF ARTIFICIAL INTELLIGENCE

During the preparation of this manuscript, the authors used the artificial intelligence tool DeepSeek-V3 to assist in the development and debugging of the R language scripts used for data analysis and visualization (as described in section 3.3.4). The model was employed for code optimization, identification of inconsistencies in programming logic, and correction of syntax errors, ensuring the robustness of the scripts and the stability of the reactive interface developed in the Shiny environment. All outputs generated by the tool were reviewed, validated, and adjusted by the authors, who assume full responsibility for the final content of the manuscript and the integrity of the data presented.

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