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O IMPACTO DA SÍNDROME DE RETT NO AMBIENTE ESCOLAR: DESAFIOS E NECESSIDADES
THE IMPACT OF RETT SYNDROME IN THE SCHOOL ENVIRONMENT: CHALLENGES AND NEEDS
IMPACTO DEL SÍNDROME DE RETT EN EL ÁMBITO ESCOLAR: DESAFÍOS Y NECESIDADES

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RESUMO

Introdução: A síndrome de Rett é uma doença genética associada a perturbações neurológicas e do desenvolvimento, tipicamente causada por mutações no gene MECP2. Afecta principalmente o sexo feminino e pode levar a graves anomalias físicas e mentais.
Objetivo: Conhecer as necessidades educativas de uma aluna com Síndrome de Rett a frequentar uma escola regular, desde a infância e tendo em conta a perspetiva familiar.
Métodos: Foi realizado um estudo de caso intrínseco a uma menina de 9 anos a quem foi diagnosticada a síndrome de Rett aos 5 anos de idade e que se encontra na fase 3 (período pseudo-estacionário). Foram utilizados como instrumentos de recolha de dados os relatórios escolares e médicos da criança, observações sistemáticas e o testemunho da mãe.
Resultados: Os resultados mostram o impacto da doença do ponto de vista educativo, as necessidades educativas, assistenciais e sociais e as principais vantagens e dificuldades da educação inclusiva. Os resultados revelam preocupações sobre a falta de coordenação entre os sistemas de saúde e de educação, bem como a contradição na informação fornecida, levando à confusão sobre as melhores práticas para o desenvolvimento holístico das pessoas afectadas e as diferenças entre a intervenção precoce e os cuidados posteriores, que são escassos.
Conclusão: Estes resultados sublinham a importância de abordar a educação dos indivíduos com síndrome de Rett de uma forma holística, reconhecendo tanto as suas necessidades específicas como as oportunidades e dificuldades associadas à inclusão educativa. A atenção às necessidades deve ser interdisciplinar, contínua e a longo prazo.

Palavras-chave: síndrome de Rett; doença rara; estudo de caso intrínseco; educação inclusiva

ABSTRACT

Introduction: Rett syndrome is a genetic disease associated with neurological and developmental disorders typically caused by mutations in the MECP2 gene. It mainly affects females and can lead to severe physical and mental abnormalities.
Objective: To learn about the educational needs of a student with Rett Syndrome attending a mainstream school, from an early age and taking into account her family's perspective.
Methods: An intrinsic case study was conducted on a 9-year-old girl who was diagnosed with Rett syndrome at the age of 5 years and is in stage 3 (pseudo-stationary period). The child's school and medical reports, systematic observations, and the mother's testimony were used as data collection instruments.
Results: The results show the impact of the disease from an educational point of view, the educational, care, and social needs, and the main advantages and difficulties of inclusive education. The findings reveal concerns about the lack of coordination between the health and education systems, as well as the contradiction in the information provided, leading to confusion about the best practices for the holistic development of the affected persons and the differences between early intervention and later care which is scarce.
Conclusion: These findings underline the importance of addressing the education of individuals with Rett syndrome in a holistic manner, recognizing both their specific needs and the opportunities and difficulties associated with educational inclusion. Attention to needs must be interdisciplinary, continuous, and long-term.

Keywords: Rett syndrome; rare disease; intrinsic case study; inclusive education

RESUMEN

Introducción: El síndrome de Rett es una enfermedad genética que se relaciona con trastornos neurológicos y del desarrollo causados típicamente por mutaciones en el gen MECP2. Afecta principalmente al sexo femenino y puede conducir a graves anomalías físicas y mentales.
Objetivo: Conocer las necesidades educativas de una alumna con Síndrome de Rett escolarizada en un centro ordinario, desde una edad temprana y teniendo en cuenta la perspectiva de su familia.
Métodos: Se realizó un estudio de caso intrínseco, una niña de 9 años que fue diagnosticada con 5 años con síndrome de Rett y se encuentra en fase 3 (periodo pseudo-estacionario). Se utilizaron como instrumentos de recogida de datos los informes escolares y médicos de la niña, observaciones sistemáticas y el testimonio de la madre.
Resultados: Los resultados muestran la repercusión de la enfermedad desde el punto de vista educativo, las necesidades educativas, asistenciales y sociales y las principales ventajas y dificultades de la educación inclusiva. Los hallazgos revelan preocupaciones sobre la falta de coordinación entre los sistemas sanitario y educativo, así como la contradicción en la información proporcionada, generando confusión en cuanto a las mejores prácticas para el desarrollo integral de las personas afectadas y las diferencias entre la Atención Temprana y la atención posterior que es escasa.
Conclusión: Estos hallazgos subrayan la importancia de abordar de manera integral la educación de individuos con síndrome de Rett, reconociendo tanto sus necesidades específicas como las oportunidades y dificultades asociadas a la inclusión educativa. La atención a las necesidades debe ser interdisciplinar, continua y de larga duración.

Palabras Clave: síndrome de Rett; enfermedad rara; estudio de caso intrínseco; **e**ducación inclusiva

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INTRODUCTION

Rett syndrome is a severe, progressive neurological disorder with an X-linked inheritance pattern that affects multiple body systems and functions. This severe neurodevelopmental disorder is a so-called rare disease that causes developmental and nervous system problems. Furthermore, according to the Diagnostic and Statistical Manual of Mental Disorders (DSM-5) it states that in the regressive phase of Rett syndrome, dysfunction may be found in the social interaction of the child who meets the diagnostic criteria for autism spectrum disorders. It leads to severe disability, influencing almost all areas of life (Valer et al., 2021). It is mainly caused by loss-of-function mutations in the MECP2 gene (80% of cases of the classical form), which codes for the MeCP2 protein, involved in the development and differentiation of the central nervous system. Ninety-nine per cent of cases are de novo mutations (single cases in the family) (Candala et al., 2021; Jara-Ettinger et al., 2021).

Worldwide, the prevalence is 1/10000 and 1/15000 in girls and it is considered the second most common cause of severe intellectual disability in females after Down syndrome (Ellaway and Christodoulou, 2001; Fehr et al., 2011; Rose et al., 2013).

In recent genetic advances, cases have been discovered in the male sex when associated with mosaicism or gonosomal alterations (Klinefelter's syndrome type) (Candala et al., 2021).

1. THEORETICAL FRAMEWORK

1.1 Characterisation of Rett syndrome

In Spain, it affects an estimated 3000 people. The disease usually develops between 6 and 18 months (AESR, 2024). There is a regression of language and motor skills, as well as an evolutionary stagnation of skills in the long term, which has a great impact on their quality of life. Some people with the disease develop comorbidities such as respiratory irregularities, epilepsy or increased anxiety. Coexisting conditions may manifest themselves in different ways depending on the person, environment and time, or at different levels of severity throughout life (Townend et al., 2020).

The Spanish Rett Syndrome Association (2024) describes the four phases of the disease:

- Phase 1. Early-onset stagnation (6 to 18 months): previously normal or mildly disturbed developmental pattern. Frequent disturbance of behaviour and sleep pattern. Deceleration of head circumference growth. Duration of this phase: weeks to months.
- Phase 2. Rapid regression (1-4 years) loss of manual skills acquired language (although bisyllables may be maintained) and communicative interest. Once standing and walking have been achieved, an apraxic gait usually develops with a greater lateral component and asymmetrical support. Variable cognitive deficit. Manual stereotypies (hand washing, hand clapping or hand movements) are almost constant. Duration of this phase: weeks to months (up to one year)
- Phase 3. Pseudo-stationary period: progressive improvement in social contact and connection with the environment. Partial restitution of ambulation and use of hands. Most of the comorbidities appear. Duration of this phase: years-decades, may last into adulthood without further deterioration.
- Phase 4. Late motor impairment: not all people go through this period. Worsening of mobility with cessation of ambulation, severe disability with atrophy, increased spasticity, sometimes parkinsonian symptoms with on-off phenomena and behavioural changes with apathy and refusal to eat. There are four main diagnostic criteria: loss of manual skills, loss of communication skills, hand stereotypies and gait abnormalities. Developmental regression is the hallmark diagnostic feature of this condition; although recovery of function is common, it is usually partial. A large proportion of affected individuals meet the diagnostic criteria for autism spectrum disorder (ASD) (Banerjee et al., 2019).

The main features and comorbidities of this condition are visual impairment, auditory processing difficulties or hearing loss, gastrointestinal problems, sensory regulation difficulties (overstimulation or understimulation), fatigue and inattention, mood and anxiety, seizures, scoliosis and kyphosis, disturbed sleep patterns, respiratory problems (breath holding, hyperventilation and air swallowing) and movement disorders (Townend et al., 2020).

People with Rett syndrome may have comorbidities, including reduced clarity or sharpness of vision (reduced visual acuity), neurological problems in the cortex and brain activity (cortical visual impairment) or reduced voluntary control of purposeful eye movements (oculomotor apraxia). The main gastrointestinal problems are reflux, abdominal bloating, constipation or diarrhoea associated with abdominal pain. Other conditions include interrupted or poor-quality night sleep, seizures, side effects of medication, poor nutritional status and sensory regulation difficulties. They may have periods of insomnia with nocturnal laughing and screaming. In the daytime they may fall asleep and need breaks, decreasing their performance. Many people, especially in adulthood, experience problems of low mood and depression. Disturbed breathing, holding your breath and wringing your hands are signs of increased anxiety. It should also be noted that 60-80% of people with Rett syndrome suffer from seizures. About 80% of people with this genetic condition can be affected by a sideways curvature of the spine (scoliosis).

Finally, the main movement disorders are manual stereotypies, loss of fine and gross motor skills, dyspraxia/apraxia and impaired muscle tone.

The US Food and Drug Administration (FDA) approved Daybue (trofinetide) oral solution as the first treatment for Rett syndrome

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in both adults and children from the age of 2 years and up (FDA, 2023). FDA approval was based on a 12-week phase 3 study evaluating the efficacy and safety of trofinetide versus placebo in 187 women with Rett syndrome between the ages of 5 and 20. Treatment showed statistically significant improved symptoms: OBJ et al., 2023).

1.2 Inclusive education

In this study, we will focus on the consequences of this disease in the educational environment. Students with rare diseases are characterized by their diversity, presenting changing and varied needs that require continuous observation and attention. Attention to diversity is a need that covers all educational stages, guaranteeing the development of all students, favoring equity, and contributing to greater social cohesion.

The Organic Law for the Modification of the Organic Law on Education (LOMLOE), Organic Law 3/2020, of 29 December, specifies that pupils with specific educational support needs are those with special educational needs (disability or serious behavioral, communication or language disorders); maturational delay; language and communication development disorders; attention or learning disorders; severe lack of knowledge of the language of learning; socio-educational vulnerability; high intellectual abilities; late entry into the education system; personal conditions or school history.

People with Rett syndrome have a range of special educational needs depending on the following areas affected: cognitive, socio-affective, motor, communication and language, and curricular areas (Merchán, 2010; Townend et al., 2020):

- At the cognitive level, specific treatment is needed to develop the basic cognitive aspects such as multi-sensory methodology, use of stimulating and diversified teaching material, and encouraging generalization.
- In the socio-affective area, it is important to learn strategies for maintaining and building relationships with your peer group.
- Regarding the motor area, one way to improve motor skills and development is by working on spatial-temporal orientations.
- Communication and language. Girls with Rett syndrome have compromised communication, but eye gaze is their strong point, which is why the use of Alternative Communication systems is recommended, especially for those with eye control. They naturally use their eyes to communicate, looking at people and focusing on their faces rather than objects. Access to literacy is crucial as it is the most accurate means of communication. Literacy plays an important role in the lives of these people. Thanks to the use of technology, it is now known that girls with Rett syndrome are more intelligent than previously thought, that they have the ability to read and write, and that they can be taught in an adapted way and with the presumption of competence, that is, by providing opportunities. Individual attention is essential at the curriculum level.

The research gap in Rett syndrome persists due to its low prevalence and complexity. Researchers such as Banerjee et al. (2019), Fehr et al. (2011), Jara-Ettinger et al. (2021), Neul et al. (2023) and Rose et al. (2013) have highlighted the paucity of patient case literature, highlighting the negative impact of the investment of funds aimed at improving health and educational strategies. This case study seeks to address this lack of research on educational and health integration, providing valuable insights to guide future studies toward more effective and personalized educational interventions for people with Rett syndrome. Therefore, the general aim of this study was to understand the educational needs of a pupil with Rett syndrome attending a mainstream school, starting from infancy and considering the family perspective. The specific objectives were: (1) To learn about the experiences of a girl with Rett syndrome in a regular school; (2) To understand the repercussions of Rett syndrome in the school and family environment.

2. METHODS

A qualitative methodology was used, specifically the intrinsic case study (Stake, 1995; Yin, 2014), which allows us to analyze and understand a particular case that is of interest, whose purpose is not to understand the generic phenomenon or to build a theory. The medical and school reports of the girl with Rett syndrome were analyzed, with systematic observations and the mother's testimony. The following timeline (Figure 1) shows the evolution of the disease:

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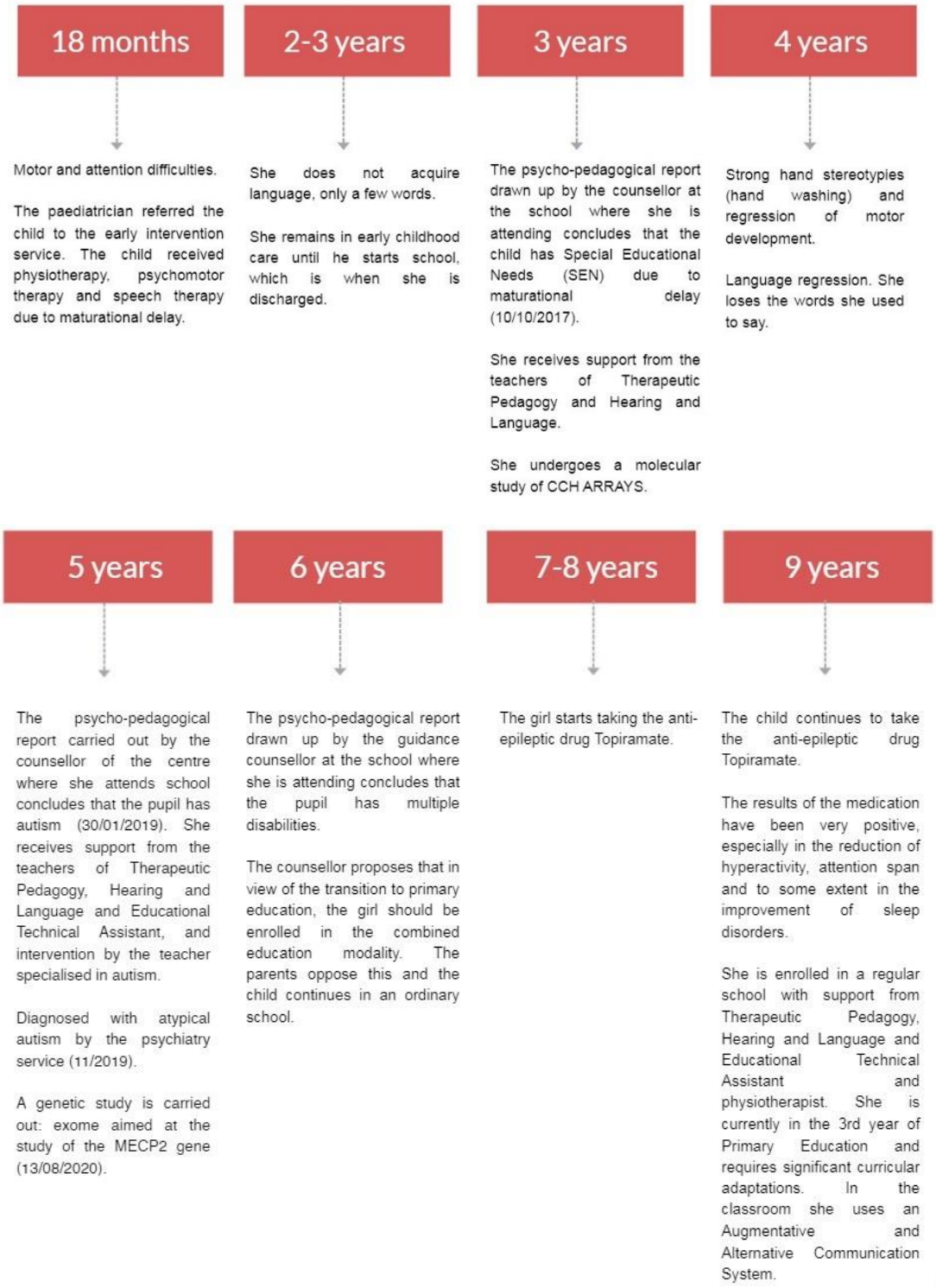
Figure 1 - Timeline of the evolution of Rett Syndrome in the interviewed child



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2.1 Participants and Context

In this study, the sample consisted of a girl with Rett syndrome (Participant 1) and her mother (Participant 2), who provided the data. The mode of sample selection was purposive in terms of our research interests and objectives.

- Participant 1. A 9-year-old girl who was diagnosed at the age of 5 and is in phase 3. She attends an ordinary school in a small rural town in the autonomous community of Castilla y León where she receives classroom support from a teacher specialised in Hearing and Language and Therapeutic Pedagogy. She also receives support from the physiotherapist. She has hearing problems, hypermetropia and has difficulties in using her hands. She uses a high-tech Augmentative and Alternative Communication System (AACS), a personalised communication board that grows with her. The board is controlled by an eye-reader which she uses in all contexts and is mainly modelled for her learning. This system allows her to communicate her needs and to relate to others. On the other hand, we work on reading and writing as one of the most relevant objectives, as it is the most complete system of communication, and we also prioritise basic mathematical skills. All the contents must be adapted to the motor and communication barriers of the pupil.
- Participant 2. Mother of the pupil who works professionally in education as a counsellor in a secondary school. She belongs to and participates in associations related to the world of functional diversity, several of which are specific to Rett syndrome. Since the birth of her daughter, she has always provided educational support focused on meeting her needs, but, above all, on improving her quality of life. Her involvement in her daughter's academic life is total, with a daily relationship, as she is responsible for setting up the child's communication system at school every morning and coordinating with all the teachers to develop teaching materials adapted to her communication system.

2.2 Data collection instruments

In the development of this study, a qualitative methodology was used following the guidelines established by COREQ (COnsolidated criteria for REporting Qualitative research) to ensure rigor in the presentation of the results (Sillero et al., 2023; Tong et al., 2007). This approach allowed for an in-depth exploration of the experiences and perspectives of the student with Rett syndrome, as well as her family, in the educational context. Semi-structured interviews were conducted with the mother, teachers, and other professionals involved, providing a comprehensive framework for analyzing educational needs from multiple perspectives. The interview script was organized around four key points: 1) Family experience in the educational environment, 2) Relationship with educational and health professionals, 3) Documentary analysis of health and school documentation, and 4) Observation of inclusion strategies in the educational and health center. The triangulation of data was achieved through the combination of school reports, medical reports, systematic observations and the testimonies of the report that were contrasted with family members who attended the child, contributing to the validity and reliability of the findings. This robust methodological approach allows for a holistic understanding of the educational implications of Rett syndrome and highlights the importance of addressing these needs in a comprehensive manner.

2.3 Data analysis

The data analysis phase was carried out using Braun & Clarke's (2006) procedure for thematic analysis. Initially, familiarisation with the data collected was undertaken through repeated review of interview transcripts, reports, and observations. This was followed by the generation of codes, identifying patterns and emerging themes in relation to the educational needs of the pupil with Rett syndrome. This coding process was conducted interactively and systematically, allowing for a thorough exploration of the experiences narrated by the mother, teachers, and other participants (Braun & Clarke, 2019). The codes were then grouped into broader thematic categories reflecting significant aspects of educational needs. The identification and definition of these themes were done through discussions and consensus among the researchers, ensuring validity and consistency in the interpretation of the data. Finally, a coherent narrative was developed that integrated the different four identified themes, providing an in-depth understanding of the educational and care dimensions related to Rett syndrome. This rigorous and reflective process of thematic analysis highlights the complexity of the student's experiences and needs, contributing to the validity and interpretative richness of the results.

2.4 Ethical aspects

The study was conducted according to the guidelines of the Declaration of Helsinki and was approved in advance by the relevant Ethics Committee (protocol code IR 29/2023).

3. RESULTS

The main results were related to the impact of the disease from the educational point of view, the attention to this special educational need and the main advantages and difficulties of inclusive education. In each of them, a small fragment appears with the mother's textual words in relation to this.

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3.1 Impact of the disease on education

The disease is very different from one girl to another, so it is not possible to generalise. Most girls, especially at the primary education stage, are enrolled in special education centres. In the case presented here, the girl is enrolled in a regular school; this means that she requires very individualised attention. Due to motor difficulties and hand dysfunctions, she needs the continuous help of a person and a communication system, as well as peers to teach and accompany her in the implementation of this system. This allows them to belong and participate in the life of the classroom and the school, encouraging socialisation.

"Undoubtedly, we have an unfinished business, I say this with total conviction, defending it with the scientific evidence in hand: in order to advance on the road to true inclusion, we must focus on the barriers and not on the disease and the disabilities it generates" (Mother's testimony). "The evolution during the second semester has been quite unstable. This is due to the child's health problems during these months, which have prevented her from focusing on her work at many times, with episodes of nervousness, hyperventilation and apnoea causing her discomfort. We continue to work with the same methodology as before, so that the learning process is well established" (Qualitative assessment of the second semester (academic year 2023-2024) in 3rd year of Primary Education by the teacher of Therapeutic Pedagogy (PT) and the tutor).

3.2 Needs of pupils with Rett syndrome

Attention to all the needs of pupils with Rett syndrome is insufficient, as resources are scarce, even though Royal Decree 193/2023, which regulates the basic conditions of accessibility and non-discrimination of people with disabilities for access to and use of goods and services available to the public, indicates that adjustments must be reasonable. However, economic reasons continue to prevail, and support is distributed according to administrative criteria and student ratios.

"I believe that there is still a long way to go to achieve true inclusive quality education because of lack of resources, because of the prevailing school training, and because of the lack of competence.

"As a mother, the phrase that represents me and in moments of weakness I almost recite as a mantra is: the limit is in the sky, we cannot put barriers to the sea" (Mother's testimony).

"As for the keyboard and the calculator, there are still difficulties in using it correctly" (Qualitative assessment of the second semester (academic year 2023-2024) in 3rd year of Primary Education by the teacher of Therapeutic Pedagogy (PT) and the tutor).

3.3 Charting a Path to Inclusion: Unlocking the Benefits of Inclusive Education for People with Rett syndrome

The first aspect is socialization, which is a benefit not only for the pupil but also for the rest of the classmates, as they internalize values, and it helps to make them aware that we all have different abilities and have our roles in society. The second aspect is stimulation at all levels, as with the challenges in communication and motor areas, it is very difficult to establish what abilities they have. It would be necessary to promote different areas of development through work in the ordinary classroom, adapting the contexts and methodologies to the pupil. To this end, the Universal Design for Learning (UDL) is the teaching methodology that will cater for diversity in the classroom, not only for students with Rett syndrome, but for any student. In this case, one of the greatest benefits is the development of communication in relation to their peers; they are a key element in the implementation of the SAAC in natural contexts based on belonging and participation.

"The quality of life of people, I think, is measured in the possible experiences they can participate in and there is no greater participation than being included in all levels of social participation, with the school being a crucial agent in all of this" (Mother's testimony).

"It is worth highlighting the use the child has made of the communicator this term. Both because of the teachers' greater knowledge of Grid and because of the child's communicative intention, we have been able to understand each other much better. This provokes a much wider conversational exchange" (Qualitative evaluation of the second semester (academic year 2023-2024) in 3rd year of Primary Education by the Therapeutic Pedagogy teacher (PT) and the tutor).

3.4 Educational challenges in the school inclusion of people with Rett syndrome

The first is the lack of resources, especially personal resources, but also material resources; for example, the communication system available to the pupil is provided by the social security system, and the family must move it from one place to another, given the importance of communication and the fact that it is a fundamental right. The pupil should have one in the school, but these systems are very expensive. On the other hand, the lack of training of professionals means that they continue to develop practices based on old paradigms that clash with the inclusion and empowerment vision that still prevails in society.

"From my experience at the family level, I would like to highlight the emotional exhaustion that the illness entails, and that is added to the fatigue of having to fight continuously for the rights of people with functional diversity recognized in the current law on education and in other regulations related to special education and functional diversity to be fulfilled" (Testimony of the mother).

"In terms of new learning, the contents taught in his classroom have been adapted so that he can understand them correctly. A lot of work has been done on multiplication, reading and writing, reading comprehension, types of animals, rivers, water, and content that has been fun and fun to work with. The biggest difficulty has been in division, as it is a rather abstract concept and has not been motivating for them either. In general, it has been a term with progress, but also with moments of stagnation" (Qualitative

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assessment of the second semester (academic year 2023-2024) in 3rd year of Primary Education by the teacher of Therapeutic Pedagogy (PT) and the tutor).

4. DISCUSSION

The main objective of understanding the educational needs of a pupil with Rett syndrome and her family was achieved. In the current context, as pointed out by Candala et al. (2021), the lack of a cure for Rett syndrome highlights the urgent need for a multidisciplinary therapeutic approach. This approach is supported by the results of our study, where relatives express concern about the lack of coordination between the health and educational levels. It is highlighted that the information provided is sometimes contradictory, leading to confusion as to the best practices to support the motor and psychological development of affected individuals. This finding underlines the importance of effective communication and close collaboration between health and education professionals to provide coherent guidance and comprehensive support to families affected by this rare disease.

Early developmental intervention, as indicated by Valer et al. (2021), is presented as an essential element to optimize capacities and extend life expectancy in those individuals affected by this neurological pathology. In this context, gaze-based communication technology emerges as a resource of considerable relevance, facilitating the literacy process and providing new avenues of expression for people with Rett syndrome. This aspect is significantly evidenced in the present case, as it is apparent that, initially, early care teams are more accessible and intervention is more effective during the early stages of development, but as complexity increases, greater helplessness and unfamiliarity is experienced, as one cannot rely exclusively on a single specialist.

As highlighted by Banerjee et al. (2019), the training of future health professionals and educators in the field of Rett syndrome is limited, as experts are only beginning to understand what the most appropriate treatments for the management of Rett syndrome might be. This gap affects children's progress, highlighting the importance of planning. In relation to these findings, our study also confirms that the role of educators and health professionals in implementing these goals is essential to encourage the active participation of girls with Rett syndrome in their daily lives. As pointed out by Rose et al. (2013) and explained in our study, the integration of gaze-based communication technologies in educational strategies emerges as a crucial resource to enhance the autonomy and improve the quality of life of these individuals.

Health advances through genetic testing for Rett syndrome favor early detection of the disease, with families experiencing less stress and emotional strain compared to those with later diagnosis. It should also be noted that diagnosis is more complex in rural than in urban areas (Fehr et al., 2011). This research clearly manifests this perspective by stating that the worst thing is the lack of knowledge and not knowing what is wrong with your daughter because you feel lost and without access to resources, recognizing that it is easier if you live in urban areas than in rural areas.

Another case study analyzed by Jara-Ettinger et al. (2021) also highlighted the difficulty of diagnosis and the need to facilitate interdisciplinary follow-up to reduce the impact of complications, highlighting in this study the difficulty of coordinated access to these professionals. It also highlights the improvements that exist between a 5-year-old and a 17-year-old.

In our study, Rose et al. (2013) and Jara-Ettinger et al. (2021) point out that the collaborative relationship between the family and the school is an extremely important factor in the comprehensive development of students with Rett syndrome. These two educational agents, acting in a complementary manner, play a crucial role in the physical, emotional, social, and intellectual progress of affected individuals, directly influencing their learning process.

As Neul et al. (2019) point out, specific difficulties associated with Rett syndrome, such as dyspraxia, apraxia, and sensory regulation, have a significant impact on communication and the acquisition of literacy skills. Dyspraxia and apraxia affect the execution of motor movements (including eye movements) and may influence speed and responsiveness, without necessarily indicating a lack of understanding of the task being performed.

Our study highlights the need to design specific educational activities related to sensory regulation. The main limitation of the study lies in the scarcity of analyzable cases, which prevents generalized extrapolation of the findings. However, it is stressed that the uniqueness of this specific case, linked to a rare disease of low prevalence such as Rett syndrome, confers substantial intrinsic value to the research.

In terms of future lines of research, it is proposed that a longitudinal follow-up be established to evaluate and review the efficacy of the treatments recently approved by the FDA in 2023. This evaluation should focus on the observation of possible improvements in symptoms such as vocalization, facial expression, and gaze, among others. In addition, there is a call to examine the impact of these health developments on education, with a particular focus on the need to review and adapt educational methods to school settings, thus establishing a more holistic approach to the care of people affected by Rett syndrome.

CONCLUSION

In this study, the educational needs of a pupil with Rett syndrome have been explored in depth, highlighting the complexities inherent in this neurological genetic disorder. The results reveal the complexity of the educational, care and social challenges associated with this rare disease, highlighting the importance of interdisciplinary and long-term care. The limited availability of research focusing on Rett syndrome underlines the neglect faced by people affected by rare diseases in education and care.

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SAACs are alternative and/or augmentative communication systems that enable the elimination of communication barriers, reducing social isolation, exclusion, and vulnerability, and enhancing the emergence of speech or improving existing capacity, providing the person with greater autonomy and having a positive impact on literacy. Learning this system is slow and requires the involvement and participation of the family, educational agents, and various specialist professionals. There is a great lack of knowledge of the SAACs by health and educational staff. The use of these devices varies depending on the Autonomous Community. In the case of this child, it was prescribed by the hospital itself, and the family did not have to bear the high cost. Initial and ongoing training in the use of SAACs is necessary, adapted to technological and methodological advances to ensure that people receive the best possible support in their communication.

Communication and language are fundamental aspects for all people. It is essential to ensure that people with disabilities have full access to effective forms of communication, a key right in their inclusion and participation in society, which is leading to situations of inequality as some people are unable to access these systems. Rett syndrome, as a rare disease, poses unique challenges in terms of research, awareness, and healthcare. The low visibility and resources allocated to rare diseases often mean a lack of support and resources. It is essential that the education community and health professionals work together to promote research and awareness of this disease to improve the quality of life of people affected by this disease and their families. Inclusive education focuses on ensuring that all students, regardless of their abilities, needs, or characteristics, have access to quality education, promoting comprehensive care that encompasses academic, social, and emotional aspects, which contributes to creating an equitable and diverse educational environment.

However, the findings also reveal concerns about the lack of coordination between the health and education systems, as well as contradictory information provided, leading to confusion about best practices for the holistic development of affected individuals. It is crucial to recognize that while early care teams can be accessible and effective during the early stages of development, the increasing complexity of needs requires a continuous and coordinated approach that cannot rely solely on a single specialist. In addition, it is essential to consider geographical disparities, as accessibility to resources and supports may be more limited in rural compared to urban areas. Ultimately, it highlights the need to design specific educational activities that address sensory regulation, recognizing its importance in the development and well-being of individuals with Rett syndrome. This study not only identifies current challenges but also points to key areas for future research and action to improve the lives of people affected by Rett syndrome and their families.

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AUTHORS' CONTRIBUTIONS

Conceptualisation, L.A.J., J.P.P. and L.A.M.; data processing, L.A.J., J.P.P. and L.A.M.; formal analysis, L.A.J. and L.A.M.; research, L.A.J., J.P.P. and L.A.M.; methodology, L.A.J., J.P.P. and L.A.M.; project management, L.A.J., J.P.P. and L.A.M.; resources, L.A.J., J.P.P. and L.A.M.; software, L.A.J., J.P.P. and L.A.M.; supervision, L.A.J., J.P.P. and L.A.M.; validation, L.A.J., J.P.P. and L.A.M.; visualisation, L.A.J., J.P.P. and L.A.M.; writing - preparation of the original draft, L.A.J., J.P.P. and L.A.M.; writing - revision and editing, L.A.J., J.P.P. and L.A.M.

CONFLICTS OF INTEREST

The authors declare that they have no conflicts of interest.

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