







# Physiotherapeutic intervention for a child with Koolen-de Vries Syndrome

*Intervenção fisioterapêutica para uma criança com Síndrome Koolen-de Vries*

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## Abstract

**Introduction:** Koolen-de Vries Syndrome (KdVS) is a rare syndrome characterized by motor development delay, hypotonia, intellectual disability, language delay, and distinctive facial features. **Objective:** To describe a case report of the clinical and functional profile of a child with KdVS and to present the implementation of a physiotherapy intervention for this child. **Methods:** This is a retrospective case report of a male child, aged 3 years and 9 months, diagnosed with KdVS. Sociodemographic characterization was performed using the Poverty Income Ratio and the Brazil Economic Classification Criterion. Denver Developmental Screening Test II was used to screen for developmental delays. The physiotherapy program followed current recommendations focusing on task-specific training. Environmental factors were assessed by Affordances in the Home Environment for Motor Development. Additional assessments of age-expected motor skills and International Classification of Functioning, Disability and Health qualifiers were used. **Results:** Treatment goals were partially achieved. The child showed improvement in activity levels and maintenance of home environment affordances. **Conclusion:** This case report showed the benefits of physiotherapy intervention for a child with KdVS.

**Keywords:** Koolen-de Vries Syndrome. Case report. Developmental delay. International Classification of Functioning, Disability and Health. Physiotherapy.

## Resumo

**Introdução:** A Síndrome de Koolen-de Vries (KdVS) é uma síndrome rara caracterizada por atraso no desenvolvimento motor, hipotonia, deficiência intelectual, atraso na linguagem e características faciais distintivas. **Objetivo:** Descrever um relato de caso do perfil clínico e funcional de uma criança com KdVS e apresentar a implementação de uma intervenção fisioterapêutica para essa criança. **Métodos:** Trata-se de um relato de caso retrospectivo de uma criança do sexo masculino, com 3 anos e 9 meses, diagnosticada com KdVS. A caracterização sociodemográfica foi realizada utilizando a Razão de Rendimento para a Pobreza e o Critério de Classificação Econômica Brasil. O Teste de Triagem de Desenvolvimento de Denver II foi utilizado para identificar atrasos no desenvolvimento. O programa fisioterapêutico seguiu as recomendações atuais, focando no treinamento específico de tarefas. Fatores ambientais foram avaliados através do instrumento Affordances in the Home Environment for Motor Development. Avaliações adicionais das habilidades motoras esperadas para a idade e qualificadores da Classificação Internacional de Funcionalidade, Incapacidade e Saúde foram utilizados. **Resultados:** Os objetivos do tratamento foram parcialmente alcançados. A criança apresentou melhora nos níveis de atividade e manutenção das oportunidades no ambiente domiciliar. **Conclusão:** Este relato de caso demonstrou os benefícios da intervenção fisioterapêutica para uma criança com KdVS.

**Palavras-chave:** Síndrome Koolen-de Vries. Estudo de caso. Atraso do desenvolvimento. Classificação Internacional de Funcionalidade, Incapacidade e Saúde. Fisioterapia.

## Introduction

In 2006, a microdeletion on chromosome 17q21.31 was identified in three individuals with common features named Koolen-de Vries Syndrome (KdVS).<sup>1</sup> This is a rare syndrome characterized by motor development delay, hypotonia, intellectual disability, language delay, and distinctive facial features such as ptosis and blepharophimosis.<sup>2</sup>

The prevalence of KdVS is still unknown, however, it is estimated that it occurs in one in every 55,000 individuals, with no difference in prevalence between the sexes.<sup>3,4</sup> Early diagnosis are based on molecular genetic tests, with different method possibilities, such as

Chromosomal Microarray Analysis (CMA) and sequence analysis.<sup>2,3</sup> The life expectancy of individuals with KdVS is still unknown.<sup>3,5</sup>

Due to the characteristic developmental delay of the syndrome, infants and children diagnosed with KdVS should be included in early intervention multidisciplinary programs to prevent further activity limitations and participation restrictions.<sup>2</sup> Early physical therapy intervention is defined by Tudella and Formiga.<sup>6</sup> as a secondary prevention treatment approach aimed at newborns and infants at risk for neurosensorimotor development alterations and should be initiated by the 4th month of chronological age. This may not always be achieved for infants with KdVS, as the diagnosis of rare diseases is often delayed. These challenges are enhanced in low- and middle-income countries such as Brazil, where early referral to specialized health services without a medical diagnosis is not straightforward. Nevertheless, children with KdVS must be guaranteed access to health and education services as facilitators for their development, optimizing their participation in the home, school, and community. To the best of our knowledge, this is the first article addressing physical therapy intervention in a child with KdVS. The closest related study we found examined the profiles of patients undergoing hippotherapy in combination with speech therapy; among the 16 patients included, one was diagnosed with KdVS.<sup>7</sup> However, this study focuses on speech therapy rather than physical therapy, and while it reports that 87.5% of patients benefitted from hippotherapy, it does not specify which patients had positive outcomes, making it impossible to determine if the KdVS patient was among them.<sup>7</sup> Considering the role of physical therapy in rare syndromes, Campos et al.<sup>8</sup> examined the influence of a therapeutic program on the functional performance of a 6-year-old child diagnosed with Charcot-Marie-Tooth type 2. After 15 sessions, the authors observed significant improvement in self-care and mobility. Similarly, Kovala et al.<sup>9</sup> studied the effects of a physical therapy intervention on a 17-month-old infant with Rubinstein-Taybi syndrome. Following 12 weeks of intervention, the infant showed marked improvement, with over a 40% increase in gross motor function.<sup>9</sup> These findings emphasize the importance of physical therapy in rare syndromes.

Even though specific guidelines for early intervention in children with KdVS are currently not available, current evidence recommends that interventions follow a

family-centered model, which aims to meet the needs of the family members, in addition to those of the patient.<sup>10</sup> Motor learning techniques, including task-oriented training – which focuses on practice and is directed toward goals to improve performance in functional tasks<sup>11</sup> – are also currently supported by evidence<sup>12</sup> and should be included in early intervention programs. The present report aims to describe the clinical and functional profile of a child with KdVS and to present the objectives and effects of a PT early intervention for this child.

## Case report

This case report was conducted in accordance with the CAse REports Guidelines (CARE)<sup>13</sup> and approved by the Universidade Federal de São Carlos (UFSCar) Ethics Committee (CAAE: 67518423.5.0000.5504). The child's guardians consented to the publication of the case description.

### History

Male child, 3 years and 9 months old, diagnosed with KdVS. Born at 36 weeks and 6 days of gestational age (late preterm), cesarean delivery without complications, birth weight of 1,950g, 40 cm high, and Apgar scores of 7 and 9 in the first and fifth minutes, respectively. The mother, aged 33 years, and the father, aged 36 years, had no family history of health conditions and did not have other children. Four days after birth, the child was admitted to the Intensive Care Unit for 18 days due to thrombocytopenia and jaundice.

Since his birth, both parents and the child's physicians noticed delayed motor development and muscle hypotonia. As a result, the diagnostic investigation was initiated, and at 3 months of age, the child was referred to an early intervention physical therapy program which started in another specialized service, which was not ours. According to the mother's report, at 2 years and 1 month old, the child acquired independent walking, however, after a seizure at 2 years and 2 months, he regressed this skill. During this period, he continued to be followed up by that one intervention physical therapy program, being discharged at 2 years and 6 months old.

At 2 years and 9 months of age, after another seizure, the child was referred to specific genetic testing through Array Comparative Genomic Hybridization (Array-CGH),

being diagnosed with KdVS. As a result of the diagnosis and the motor development delay still observed by the mother, at 2 years and 10 months old, the child was admitted to our program "Early intervention and follow-up program for babies at risk in the first three years of life" from the Núcleo de Estudos em Neuropediatria e Motricidade, which takes place on the premises of the School Health Unit of the UFSCar, in Brazil.

### Assessment

The physical therapy assessments and intervention in this case report were based on the biopsychosocial model using the International Classification of Functioning, Disability and Health (ICF),<sup>14,15</sup> and are related to the physical therapy intervention carried out in the "Early Intervention and Follow-up Program for Babies at Risk in the First Three Years of Life". It does not relate to any previous interventions the child received.

The evaluation form of our program was used to assess body structures and functions, activities, participation, and contextual and environmental factors. The form contains the following information: anamnesis, medical history, pregnancy and childbirth data, socioeconomic issues, physical examination (tonus and muscle strength, range of motion, and tactile sensibility), assessment of capacities and activity limitation, other therapies undertaken, access to resources and technologies (such as orthoses and medicines), the child's home routine (feeding, bathing routine, sleep), school attendance, among other variables related to contextual factors. The socioeconomic status (SES) was classified by the Poverty Income Ratio (PIR), which is the ratio between family income, the specific poverty level of the residence area (in Brazil, the value is R\$145.00, according to IBGE data from 2019), and the number of residents in the household. Thus, SES can be categorized as follows: low SES: maternal education below high school +  $PIR < 2$ ; high SES: maternal education is college degree or higher +  $PIR \geq 2$ ; medium SES: all other combinations.<sup>16</sup> In addition, the family's purchasing power was based on the Brazil Economic Classification Criterion (ABEP).<sup>17</sup>

The ICF qualifiers were used according to the most relevant components of body function and structure, activity and participation. The information for determining the ICF qualifiers was obtained from the assessment form of our program, based on the perceptions of the therapists and caregivers. Negative scale qualifiers were

used for the body function component to indicate the extent or magnitude of the impairment, while extent, nature, and location qualifiers were used for body structure. The capacity qualifier was used for the activity and participation component to describe the individual's inherent or intrinsic ability to execute a task or an action.<sup>14,15</sup> In the environmental factors' component (e.g. family support, access to health care services), when a resource had a positive effect, it was considered a facilitator, and when the effect was negative, it was considered a barrier.<sup>14,15</sup> A positive effect included any resource that provided better opportunities for the child's functional outcomes, such as access to healthcare services. In contrast, a negative effect included any resource that limited these opportunities, such as restrictions on participation.

In addition to the assessment form, the Brazilian version of the Denver Developmental Screening Test II was used to characterize the child and for an initial developmental screening.<sup>18</sup> The Affordances in the Home Environment for Motor Development (AHEMD) was used to identify the environmental factors.<sup>19</sup>

The Denver II is a normative developmental screening test that aims to identify delays in neuropsychomotor development in children between 15 days and 72 months of age.<sup>20,21</sup> The test consists of 125 items distributed across four domains: personal-social, fine-motor adaptive, gross motor, and language.<sup>20</sup> In this case report, the translated and cross-culturally adapted version for the Brazilian population was used, which demonstrated a reliability of 96.91%.<sup>18</sup> Each item is classified

into advanced, normal, caution, and delay.<sup>20-22</sup> Since this instrument is recognized as an essential tool for monitoring child development,<sup>23</sup> our goal in using it was to assess the child's motor development status and help set appropriate goals based on their age and developmental stage.

The AHEMD aims to verify the affordances offered in the home environment for the motor development of children between 18 and 42 months of age.<sup>19</sup> It consists of 67 items, with one section on child and family characteristics and another assessing affordances in space, stimuli, and motor toys.<sup>19,24</sup> The scoring goes from 0 to 4 points, where 0 represents a very low score and 4 represents a high score.<sup>24</sup> The Portuguese version of the AHEMD was used, which presented a high reliability and confidence index.<sup>19</sup>

### Physical therapy intervention description

The collected data and described intervention encompass the period from August 2021 to January 2022. The structure of this physical therapy program consisted of sessions that took place twice a week, with an average duration of 45 minutes, for 6 months. The total dose of the intervention was approximately 30.75 hours, considering the child's attendance at the sessions. The intervention goals were defined according to the family's expectations and clinical reasoning, focusing on age-expected activities through task-specific training (Table 1). For goal setting, three periods were considered: short (2 months), medium (4 months) and long (6 months) term.

**Table 1** - Physical therapy treatment goals

Period	Goals set
Short-term (2 months)	Being able to maintain single-leg support, without hand support, for 2 seconds to kick a ball.
Medium-term (4 months)	Being able to maintain single-leg support, without hand support, for 10 seconds, while his mother puts on his shoes.
Long-term (6 months)	Being able to walk and play on uneven terrain (e.g. grass, sand). Being able to independently go down a slide.

Intervention strategies were structured based on the ICF, and adapted from Tudella et al.,<sup>25</sup> considering the expected activities for the child's age (Figure 1). Each therapeutic session was divided into four phases, incorporating daily routine activities according to pre-established goals and using a playful approach: (1) The session

started with activities focused on body function and structure, usually incorporating balance and proprioception activities, (2) followed by activities that provided strengthening exercises used to enhance body function and capacities. (3) Then, functional exercises were stimulated to transfer the motor learning to daily routine

activities (e.g. circuits); (4) The last session phase was the parental training, where guidance was provided to the caregivers on activities to be performed at home and in the community, in addition to the therapy to enhance the child's participation. The exercises included daily routine activities, manual abilities, mobility, and balance. In this way, the activities were structured in a circuit format with psychomotor games, and gross and fine motor skills practice.

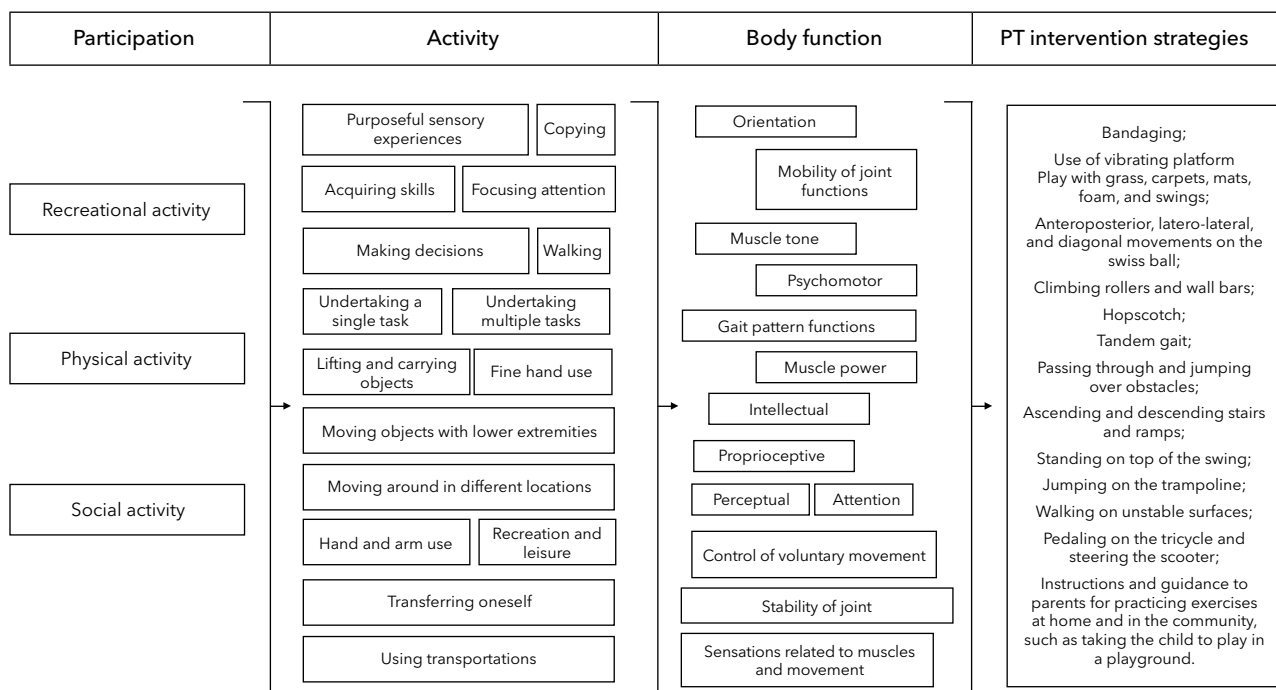
For the goal of remaining in single-leg support for kicking, we use practice and activity focused on the functional task of kicking a ball, to strengthen learning,<sup>26</sup> in addition to offering balls of different sizes and weights for kicking. For the goal of remaining in single-leg support for a longer period, aiming to put on shoes and be able to walk and play on uneven terrain, circuit activities were implemented. These circuits were designed based on the concept that movement emerges from the dynamic interaction among the individual, the task, and the environment where the task will be performed.<sup>27</sup> It included activities that encouraged him

to perform hopscotch, tandem walking, crossing and jumping over obstacles, going up and down steps and ramps, climbing backrests and rollers, standing on a swing, jumping on a trampoline, walking on unstable surfaces, pedaling a tricycle and driving a scooter.<sup>28-30</sup>

The circuits were carried out in clinical environments and in external environments, aiming to represent activities that the child experiences in daily life.<sup>26</sup> To increase the perception and difficulty of the activity, the circuits were performed with 0.5-kilogram shin weights.

To achieve the goal of going down a slide independently, it was used the principle of practice and repetition, decreasing the support as the child felt safe and increasing the motivation to perform the task.

The main supplies used in the activities were balance board, mat, ramps, wedge, roller, ladder, narrow surface, cushion, slide, swing, tricycle, scooter, ball, mats, foam, shin pads, trampoline, and various toys with the objective of improving motivation during activities. The use of toys was directed to a specific activity, also using them as therapeutic tools.



**Figure 1** - Interaction between the International Classification of Functioning, Disability and Health components and the intervention strategies used.

Note: Adapted from Tudella et al.<sup>25</sup> PT = physical therapy.

### Pre- and post-intervention outcomes

Following the initial assessments, a physical therapy diagnosis was established. It was observed that the child exhibited hypotonia associated with impaired gravitational proprioception and muscle weakness, which limits their community gait abilities.

According to the PIR, the socioeconomic status was considered high (mother's level of education = higher education completed + PIR  $\geq 2$ ) and, in the ABEP, the classification was B2 (class 3 in a system of classes from 1 to 6, in which 1 is the highest, and 6 the lowest).<sup>17</sup>

In the activity-level screening, according to Denver II (Table 2), 12 delays and five cautions were observed regarding developmental delays, which the majority were related to the language domain. Regarding gross motor skills, the child showed delays in activities such as jumping and balancing each foot for one second, which were incorporated into the physical therapy program.

Table 3 describes the functioning profile through the qualitative assessment of age-expected motor skills before and after the intervention, as well as the impairments in the body structures and functions related to each activity limitation, through observation of functional capacities.

The use of the Denver II for sample characterization supported a qualitative assessment of functional abilities, as it helped identify age-appropriate activities for the child as well as activities that should have been previously acquired.

**Table 2** - Characterization of motor development according to the Denver Developmental Screening Test II

Domain	Pre-intervention assessment	
	Delay	Caution
Personal-social	Put on clothing (F) Name friend (F)	Put on a t-shirt (F)
Fine motor-adaptive	Tower of 4 cubes (F) Tower of 6 cubes (F)	Tower of 8 cubes (F)
Language	3 words (F) 6 words (F) Combine words (F) Name 1 picture (F) Point 4 pictures (F) Speech half understandable (F) Name 4 pictures (F)	Know 2 actions (F) Know 4 actions (F)
Gross motor	Kickball forward (R) Broad jump (F)	Balance each foot 1 second (F)
Denver II results	12 delays	5 cautions

Note: F = failed; R = refused.

**Table 3** - Functional capacities and limited activities before and after intervention

Activity	Pre-intervention			Post-intervention	
	Functional capacities	Limited activities	Related body structure and function impairment	Functional capacities	Limited activities
Maintain single-leg support (short- and medium-term goals)	He is able to <b>maintain single-leg support with support from both hands</b> on a bench at hip height.	He is not able to maintain single-leg support without hand support.	Global hypotonia; Weakness of the quadriceps, hamstrings, and gluteus maximus, foot inverters (anterior tibialis), foot everters (long and short peroneus), and foot intrinsic muscles; Weakness and decreased synergy of trunk muscles; Gravitational insecurity.	He is able to <b>remain in a single-leg stance without support</b> for up to 2 seconds on each limb support.	He is not able to remain in a single-leg stance without support for more than 2 seconds on each limb support.
Play in a squatting position	He is able to play in a squatting position indefinitely.	N/A	N/A	He is able to play in a squatting position indefinitely.	N/A

Note: Note: N/A = not applicable. Bold text indicates functional capacities that improved post-intervention.

**Table 3** - Functional capacities and limited activities before and after intervention (continued)

Activity	Pre-intervention			Post-intervention	
	Functional capacities	Limited activities	Related body structure and function impairment	Functional capacities	Limited activities
Climb and descend stairs alternating lower limbs	He is able to go up and down stairs alternating lower limbs <b>with support of both hands</b> on a handrail.	He is not able to go up and down stairs alternating lower limbs without the support of both hands on a handrail.	Global hypotonia; Weakness of the quadriceps, foot inverters (anterior tibialis), foot eventers (long and short peroneus), and foot intrinsic muscles; Gravitational insecurity.	He is able to go up and down stairs alternating lower limbs <b>with the support of a single hand</b> on a handrail.	He is not able to go up and down stairs alternating lower limbs without hand support on a handrail.
Play on uneven terrains (long-term goal)	He is able to play on uneven ground for a <b>short period</b> .	He is not able to play on uneven ground indefinitely.	Sensorial impairment.	He is able to play on uneven ground <b>indefinitely</b> .	N/A
Slide down the slide (long-term goal)	He is able to go down the slide <b>with therapist support on his trunk</b> .	He is not able to go down the slide without support on his trunk.	Gravitational insecurity.	He is able to go down the slide <b>with one-hand support</b> .	He is not able to go down the slide without the support of one hand.
Perform tandem gait	He is able to perform tandem gait <b>with one-hand support</b> .	He is not able to perform tandem gait <b>without support</b> .	Global hypotonia; Weakness of the foot inverters (anterior tibialis), foot eventers (long and short peroneus), and foot intrinsic muscles; Decreased synergy between trunk muscles; Gravitational insecurity.	He is able to perform tandem gait <b>with one-hand support</b> .	He is not able to perform tandem gait <b>without support</b> .
Run long distances	He is able to run long distances.	N/A	N/A	He is able to run long distances.	N/A
Jump with bimanual support	He is able to jump <b>with the support of both hands</b> on a bench at hip height.	He is not able to jump <b>without hand support</b> .	Global hypotonia; Weakness of the quadriceps, hamstrings, gluteus maximus, and triceps surae; Decreased synergy and weakness of trunk muscles; Gravitational insecurity.	He is able to jump <b>without support</b> .	N/A

Note: N/A = not applicable. Bold text indicates functional capacities that improved post-intervention.

The ICF qualifiers, used for participant characterization purposes, were assigned considering both the therapists' and parents' perceptions. These assignments were based on the information collected through the assessment form, the standardized instruments, and the qualitative assessment of the ICF qualifiers according to the child's age (Table 4), pre- and post-intervention.

The AHEND score from the assessment at 36 months (pre-intervention) remained the same at 42 months of age (post-intervention) and was classified as average. In the post-intervention assessment, regarding the treatment goals established, the child was able to maintain one leg support, without hand support, for 2 seconds to kick a ball, fulfilling the short-term goal and partially the

medium-term goal; and he was able to walk and play on uneven terrain, fulfilling the long-term goal. Regarding the second long-term goal, the child was not able to go down the slide independently, mainly due to sensory insecurity issues and not specifically due to motor issues.

Based on the ICF qualifiers, muscle strength, the acquisition of basic skills, kicking, and jumping showed improved scores post-intervention. Similarly, the guidance provided to the family, such as referring the child to other health professionals (occupational and speech therapist) to optimize the development with a holistic

focus on the child's participation, also demonstrated improved scores post-intervention. In both evaluations of opportunities in the environment with AHMED, the scores obtained in all domains maintained the same. According to the AHMED, the affordances presented in the house offer sufficient opportunities for the child's motor development. During the intervention period, therapists actively encouraged the family to provide affordances for the child's development at home, recognizing it as the primary environment where the child spends the most time.

**Table 4** - Main pre- and post-intervention assessments using the International Classification of Functioning, Disability and Health qualifiers

Component/Category	Pre-intervention assessment	Post-intervention assessment
<b>Body structure and functions</b>		
b7300 - Power of isolated muscles and muscle groups (quadriceps and pronators)*	.2 (moderate problem)	.1 (mild problem)
b7305 - Power of muscles of the trunk	.1 (mild problem)	.1 (mild problem)
<b>Activity and participation</b>		
d1550 - Acquiring basic skills*	.2 (moderate problem)	.1 (mild problem)
d4351 - Kicking*	.2 (moderate problem)	.1 (mild problem)
d4502 - Walking on different surfaces	.1 (mild problem)	.1 (mild problem)
d4553 - Jumping*	.2 (moderate problem)	.1 (mild problem)
<b>Environmental factors</b>		
e1100 - Food	+8 (facilitator, not specified)	+8 (facilitator, not specified)
e1101 - Drugs	+8 (facilitator, not specified)	+8 (facilitator, not specified)
e1400 - General products and technology for culture, recreation and sport	+8 (facilitator, not specified)	+8 (facilitator, not specified)
e310 - Immediate family	+8 (facilitator, not specified)	+8 (facilitator, not specified)
e355 - Health professionals	+8 (facilitator, not specified)	+8 (facilitator, not specified)
e360 - Other professionals*	.8 (barrier, not specified)	+8 (facilitator, not specified)
e410 - Individual attitudes of immediate family members	+8 (facilitator, not specified)	+8 (facilitator, not specified)
e5800 - Health services	+8 (facilitator, not specified)	+8 (facilitator, not specified)
e585 - Education and training services, systems, and policies*	.8 (barrier, not specified)	+8 (facilitator, not specified)

Note: \*Categories that had post-intervention change.

## Discussion

This case report aimed to describe the clinical and functional profile of a child with KdVS, and to present the implementation of a physical therapy intervention for this child. As far as we know, this is the first case to functionally characterize and report a physical therapy intervention for a child with KdVS.

The ICF allows standardizing and describing the performance of skills performed in daily life according to the interference of components of body structures and functions, activity, and participation, as well as environmental factors and personal factors (motivation), according to the individual's needs.<sup>31</sup>

Improvements were observed in the child's activity level, such as going up and down stairs alternating lower limbs with the one hand support on a handrail, remaining in one leg support without hand support for up to 2 seconds on each foot, and jumping without hand support. These results allow us to corroborate with Koolen et al.,<sup>2</sup> when affirming the importance of physical therapy intervention to maximize skills acquisition and reduce the risk of late-onset orthopedic complications. To achieve the proposed goals and in coherence with the functional capabilities and limited activities presented by the child, the physical therapy strategies were performed in accordance with the most current evidence for interventions in children with disabilities, such as task-specific training, environmental enrichment, and strength training.<sup>32</sup> It should be noted that Novak et al.<sup>32</sup> highlighted this evidence for children with cerebral palsy. However, it is possible to extrapolate these intervention strategies to other health conditions when specific evidence is not available.<sup>33</sup> The results of this case report are important as they enrich the literature regarding physical therapy intervention strategies in motor outcomes for a child with KdVS.

The DENVER II was used for the child's activity-level characterization and for an initial screening of the development. The gross motor and fine motor-adaptative delays and cautions were identified and addressed in the physical therapy intervention. The cautions and delays in the "personal-social" and "language" domains, were not addressed in physical therapy, however, the family was oriented by the therapists to look for a multidisciplinary service to address these delays. Future studies could add another gross and fine motor assessment instruments to the assessment of children with KdVS.

We highlight the importance of family-centered practice, as the achieved goals were set in family-professional collaboration, and the intervention strategies were planned to achieve specific activities according to expectations, preferences, and family needs. In this case report, the age and cognitive ability of the child did not allow for his direct collaboration in the goal-setting approach, which would be more motivating and more likely to be achieved.

Initially, two environmental factors were identified as barriers: other professionals, and education and training services. In the pre-intervention assessment, the child was not followed up by other rehabilitation professionals besides physical therapy, such as speech

therapy. The child also did not attend school, which according to Koolen et al.<sup>2</sup> could help with other factors, such as intellectual disability associated with the syndrome diagnosis. Therefore, the family was instructed to seek these resources, and, in the post-intervention assessment, these barriers became facilitators.

The child presented as environmental facilitators: food, medication, general products and technologies for cultural, recreational, and sports activities, immediate family, individual attitudes of immediate family members, and health services. Among these, we highlight the immediate family and their individual attitudes, which corroborate with Anjos et al.,<sup>34</sup> who highlighted the support of the immediate family as the main facilitator for infants with Down syndrome. In addition, factors such as high socioeconomic status and high parental education were facilitators, as they brought benefits to the child's development.<sup>34</sup>

The AHMED was essential to analyze the opportunities offered in the home environment and toys, in order to guide the family in the activities to be done at home and in the community. Overall, no changes were observed in the scores before and after the intervention, which was positive, as the classification for the majority of the categories were either "high" or "good". Due to household limitations, improvements could not be made in the "outside space" category, so the family was instructed to look for playgrounds. The family was also instructed to pay attention to the "fine motor toys" category, which provided very few opportunities for the child's development. However, due to particularities concerning cognitive development and the child's personal preferences, this category did not show improvement. The other affordances were already sufficient for the child prior to the intervention.

It is recommended that future studies use validated instruments to assess gross motor function and child participation in different environments, such as home, school, and community, to generate results comparable with typical peers.

## Conclusion

The clinical and functional profile of the patient with KdVS was detailed in this case study. In addition to describing the child's clinical history, the results of standardized assessments were presented regarding the child's

developmental stage at the pre-intervention stage, as well as the functional abilities and activity limitations observed qualitatively, described in both the pre- and post-intervention stages. We believe that this functional description of a child with KdVS is novel information, with the potential to contribute to clinicians and researchers in the field. Through this case report, we highlight the benefits of physical therapy intervention focusing on task-specific training and on a family-centered approach for a child with KdVS. Physical therapy intervention may minimize delays in the motor development of a child with KdVS, improving functionality and minimizing possible impairments in body structures and functions, preventing limitations in activities and restrictions on participation.

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## Authors' contributions

APZS contributed with conceptualization, data curation, methodology, and writing the original draft. IPS contributed with conceptualization, methodology, and writing the original draft. MDW contributed with data curation, methodology, reviewing and editing the draft. SRSF contributed with methodology, and writing the original draft. ACC contributed with supervision, reviewing and editing the draft. ET contributed with supervision, project administration, reviewing and editing the draft. All authors actively participated in the discussion, review, and approval of the final version of the manuscript.

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