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**Review Article**

# Characterization of the Language, Speech, Voice, Swallowing, and Hearing Domains in Prader-Willi Syndrome: A Scoping Review

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**ABSTRACT**

Prader–Willi Syndrome (PWS) is classified as a rare genetic disorder with widespread developmental consequences, affecting multiple domains. This study aimed to characterize PWS in relation to swallowing, psychomotor development, speech, voice, language, and hearing in order to identify communication, feeding, and hearing needs. To this end, a scoping review was conducted across seven databases to identify articles published between 2000 and 2022. Thirty-two articles were selected from this search, from countries such as the United States, the United Kingdom, Spain, and Brazil. The reviewed studies report disorders in speech and swallowing associated with hypotonia, which results in difficulties in articulation and in the coordination of sucking, breathing, and swallowing. Additionally, the articles describe delays in language development, voice disturbances such as weak intensity and hypernasality, and generally normal hearing. It is concluded that, due to the range of disorders affecting psychomotor development, speech, swallowing, and language, people with PWS require early speech-language therapy intervention to support these areas.

**Keywords:**
Prader-Willi Syndrome; Rare Diseases; Speech-Language Therapy

## Caracterización de las áreas de lenguaje, habla, voz, deglución, y audición en el Síndrome de Prader Willi: Una revisión de alcance

**RESUMEN**

El Síndrome de Prader Willi (SPW) pertenece a las enfermedades huérfanas y se caracteriza por ser un trastorno genético que tiene consecuencias generalizadas en el desarrollo, por lo que afecta a diferentes áreas. El objetivo del presente trabajo es caracterizar el SPW en relación a la deglución, el desarrollo psicomotor, el habla, la voz, el lenguaje y la audición, mediante una revisión de alcance, con el fin de identificar las necesidades comunicativas, de alimentación y de audición. Se realizó una revisión de alcance, considerando 7 bases de datos para identificar artículos publicados entre los años 2000 y 2022. De ellos se seleccionaron 32 artículos procedentes de países como EEUU, Reino Unido, España y Brasil. Los artículos seleccionados indican que manifiestan alteraciones en las áreas de habla y deglución relacionadas con la hipotonía, la cual genera dificultades en la articulación y coordinación succión-respiración-deglución. Además, presentan retrasos del lenguaje, alteraciones en la voz como intensidad débil e hipernasalización y generalmente presentan audición normal. Se concluye que debido a los diferentes trastornos en el desarrollo psicomotor, el habla, la deglución y el lenguaje, los usuarios con SPW requieren de intervención temprana desde fonoaudiología para apoyar las áreas mencionadas.

**Palabras clave:**
Síndrome de Prader-Willi; Enfermedades Raras; Fonoaudiología

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## INTRODUCTION

Orphanet, the Portal for Rare Diseases and Orphan Drugs, defines a rare disease as a condition affecting 1 in 2,000 people (Orphanet, 2012). There is typically limited knowledge on the characteristics of these diseases, and they follow a severe, chronic course, often involving multiple motor, sensory, and cognitive deficits. These features make rare diseases highly complex from a clinical perspective. Moreover, they are challenging to recognize and diagnose (Posada et al., 2008), and there is generally a lack of specific treatments due to the limited understanding of their etiology. They are also referred to as "orphan diseases" because they are considered insufficiently "attractive" for clinical research (Cortés, 2015).

Prader-Willi Syndrome (PWS) can be found among the range of rare diseases. This condition affects around 15,000 to 25,000 children, occurring equally in both sexes and across all racial groups (International Prader Willi Syndrome Organization [IPWSO], 2022).

People with PWS present with a highly variable multisystem disorder. The phenotype is commonly associated with the following features: delayed motor development characterized by child hypotonia, hypogonadism, short stature, small hands and feet, and a distinctive facial appearance; mild to moderate cognitive delay, and language delays affecting phonological, lexical, syntactic, and morphological components. Additionally, children with PWS might exhibit behavioral difficulties, including obsessive-compulsive symptoms, verbal perseveration, hyperphagia, low activity levels, and behavioral outbursts (Atkin & Lorch, 2007).

Given the broad impact on multiple areas of development, people with PWS require ongoing support involving their environment and continuous interdisciplinary intervention (Barañón-Trujillo, 2015). Consequently, there is a need for research that allows speech-language therapy to clearly define its role in addressing this condition. This knowledge would enable speech-language therapists to implement evidence-based interventions that positively impact the quality of life of this population across all domains of functioning.

The Colombian Prader-Willi Syndrome Association (Asociación Colombiana Síndrome de Prader-Willi, 2022) asserts that formal empirical publications on this disease are limited, including assessments, diagnoses, and therapeutic follow-ups. This lack of evidence creates barriers to advocating for appropriate access to specialized services required by people with PWS. Moreover, it hinders the development of intervention guidelines in countries lacking national-level organizations.

The Colombian Federation of Rare Diseases (Federación Colombiana de Enfermedades Raras [FECOER] (2012) has noted the existing gaps in PWS research, highlighting the need to reduce them:

> In Colombia, there is little knowledge about Prader-Willi Syndrome. Consequently, some people remain undiagnosed. Early diagnosis undoubtedly improves prognosis, and the implementation of strategies aimed at appropriate symptom management, emotional support, communication, information provision, family care, and research is essential to achieving significant improvements in the quality of life for both the family unit and people with PWS in particular (p. 5).

Taken together, the combination of three characteristic variables—a high pathological complexity, low prevalence, and limited scientific production—frequently results in individuals with PWS not receiving timely diagnoses, which may lead to inadequate treatments and delay the appropriate multidisciplinary support (Martínez Franco et al., 2019).

Accordingly, this scoping review aims to characterize PWS in relation to swallowing, psychomotor development, speech, voice, language, and hearing, to identify communicative, feeding, and auditory needs.

## MATERIALS AND METHODS

A scoping review methodology was considered appropriate for literature mapping due to the complex and heterogeneous nature of the topic (Peters et al., 2015). Chambergo-Michilot et al. (2021) describe a scoping review as a rapid literature review that synthesizes scientific findings and identifies gaps in knowledge on a specific topic.

An exhaustive search was conducted in the following databases: Scopus, Web of Science, PubMed, ScienceDirect, SciELO, the Virtual Health Library (BVS), and Google Scholar. The review followed the framework by Arksey & O'Malley (2005) and the guidelines of the Joanna Briggs Institute (Peters et al., 2020). The PRISMA-ScR checklist (Tricco et al., 2016) and the PCC framework (Population, Concept, and Context) (Peters et al., 2020) were used to guide the process.

Table 1 presents the PCC strategy employed in this study.

**Table 1**. PCC Framework.

| PCC | Definition |
|---|---|
| Population | People diagnosed with PWS. |
| Concept | Swallowing, psychomotor development, speech, voice, language, and hearing characteristics. |
| Context | Research articles published between 2000 and 2022.<br>Articles retrieved from the following databases: Scopus, Web of Science, PubMed, ScienceDirect, SciELO, the Virtual Health Library (BVS), and Google Scholar.<br>Open Access articles.<br>Papers written in Spanish, English, or Portuguese.<br>Studies in the fields of health sciences and neurosciences.<br>Various study designs, such as experimental studies, case studies, and synthesis works, including systematic reviews. |

The inclusion criteria were: (a) articles published between 2000 and 2022, to cover a broad range of at least 20 years; (b) publications written in English, Spanish, or Portuguese; (c) open-access articles; and (d) studies belonging to the fields of health sciences and neuroscience. The exclusion criteria were: (a) articles without open access and (b) studies that did not address characteristics of PWS in areas such as language, motor development, speech, swallowing, or hearing.

Due to the limited information available on this topic, gray literature was also included. The search equation used the MeSH terms "Prader-Willi Syndrome" and "Speech Therapy," combined with the Boolean operator AND. This search strategy was replicated in Spanish and Portuguese.

Experimental studies, case studies, and synthesis works such as systematic reviews were selected and analyzed using a qualitative methodology. Additionally, titles and abstracts were screened using a hypothetical-deductive approach. Subsequently, the team read the full texts of the final selection. Finally, a data extraction grid was designed to systematize the information, which included the following elements: title, year, author, bibliographic reference, objective, sample, methods and instruments, statistical results, conclusion, and observations.

### Search Results

The initial search yielded 6,305 potential registers, which were reduced to 4,577 after removing duplicates. After applying filters and eligibility criteria, 116 articles were selected. Following a critical reading of these articles, including titles, abstracts, and full texts, 32 registers were ultimately included in the review. Table 2 provides a summary of the search strategy, and Figure 1 illustrates the information screening process.

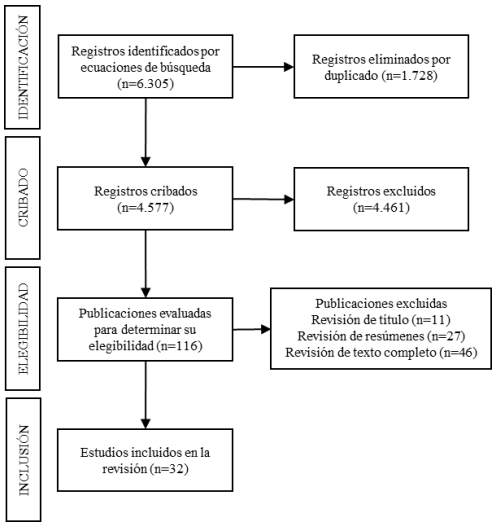


**Figure 1**. Flowchart of the selection process.

**Comentado [AB1]:** Traducción figura 1 (de izq a derecha y arriba-abajo)

**Identification** / Records identified from the search equation (n=6,305)
> Duplicate records removed (n=1,728)

**Screening** / Records screened
(n = 4,577)
> Records excluded (n=4,461)

**Eligibility /** Reports assessed for eligibility (n=116)
> Reports excluded
Title review (n=11)
Abstract review (n=27)
Full text review (n=46)

**Included /** Studies included in review (n=32)

## RESULTS

The results section is organized into two subsections. The first subsection presents the general characteristics of the studies included in this review. The second subsection provides a detailed qualitative description of the features of Prader-Willi Syndrome (PWS), considering the areas of swallowing, psychomotor development, speech, voice, language, and hearing, as identified in the selected articles.

**Table 2**. Search equations.

| Language | Search Equation |
|---|---|
| English | ("Prader-Willi Syndrome"[Mesh]) AND "Speech Therapy"[Mesh]) |
| Spanish | ("Síndrome de Prader-Willi" AND "Terapia del Lenguaje") |
| Portuguese | ("Síndrome de Prader-Willi" AND "Terapia da Fala e da Linguagem") |

### General Characteristics of the Studies Included in the Review

Table 3 presents the main characteristics of the studies included in this review.

### Characteristics of Prader-Willi Syndrome

#### *Motor Development*

Infants with PWS exhibit marked central hypotonia, with reduced muscle tone in the neck and extremities. They typically present with a small forehead, narrow face, almond-shaped eyes, a small mouth, and thin lips. These infants cry only sporadically and with low intensity; some may not cry or babble. Their movements are often repetitive, and early motor milestones are generally achieved with delays.

**Table 3.** Summary of Studies Included in the Review

| Author | Year | Design | Objective | Participants |
|---|---|---|---|---|
| Defloor et al. | 2000 | Experimental | Determine the prevalence and nature of disfluencies in individuals with PWS | 15 people with PWS, chronological age 9.9–20.0 years |
| Defloor et al. | 2001 | Experimental | Further delineate the prevalence and nature of voice problems in individuals with PWS through acoustic voice analysis | 22 people with PWS; children aged 6 years, 7 months to 11 years, 7 months; adolescents and adults aged 17 years, 1 month to 29 years, 5 months |
| Lewis et al. | 2002 | Experimental | Examine existing literature on speech and language skills in PWS | 55 people with PWS, aged 6 months to 42 years |
| Defloor et al. | 2002 | Experimental | Contribute to the documentation and delineation of the prevalence, nature, and severity of articulation difficulties in PWS | 13 people with PWS, chronological age 7–29 years |
| Bellon-Harn | 2005 | Case study | Evaluate a single case to highlight heterogeneity in PWS-MatUPD and describe clinical treatment and outcomes. | One 6-year-old girl with PWS and maternal uniparental disomy |
| Stein et al. | 2006 | Experimental | Examine the link between SSD phenotypes and microsatellite markers on chromosome 15q14-21 associated with autism, PWS, and dyslexia | 151 families with children aged 2–16 years diagnosed with SSD |
| Atkin & Lorch | 2007 | Case study | Analyze the lexical development of a child with PWS over 18 play sessions across 4 months. | One child with PWS, aged 3.7–3.11 years during the study |
| Van Borsel & Tetnowski | 2007 | Literature review | Describe different genetic syndromes associated with fluency disorders | Not Applicable |
| Pituch et al. | 2010 | Descriptive | Identify rehabilitation priorities of parents with children, including adult children, with PWS, and determine their relation to the adaptive behavior levels of the child | 50 mothers and 8 fathers of children with PWS, aged 21–61 years |
| Misquiatti et al. | 2011 | Case study (Longitudinal) | Describe the course and outcomes of speech therapy at a teaching clinic over 4 years, in a child with PWS. | One 8-year-old child with PWS |
| Saeves et al. | 2011 | Experimental | Evaluate orofacial dysfunction in individuals with PWS compared with a healthy reference group | 45 people with PWS (23 M, 22 F), mean age 19.8 ± 9.5 years; 40 healthy controls (18 M, 22 F), mean age 24.0 ± 16.3 years |
| Allen | 2011 | Descriptive | Explore the management of PWS within families | 80 participants: 34 parents, 20 youth with PWS, 26 siblings; participants with PWS aged 11–15 years at study onset |
| Zambotti & Souza | 2013 | Case study | Describe and analyze speech therapy effects in a cooking workshop for a patient with PWS. | One 5-year-old child with PWS |

| | | | | |
|---|---|---|---|---|
| Dimitropoulos et al. | 2013 | Descriptive | Further characterize expressive and receptive language skills in 35 participants with PWS and compare by genetic subtype using CELF-IV (Clinical Evaluation of Language Fundamentals-4) | 35 people with PWS, aged 7–44 years |
| Caixàs Pedragós et al. | 2014 | Literature review | Describe PWS and its main features | Not Applicable |
| Crockett et al. | 2014 | Retrospective | Review the occurrence of velopharyngeal dysfunction in PWS patients post-adenotonsillectomy for obstructive sleep apnea | 11 patients (5 M, 6 F), aged 2–9 years |
| Martín Santana | 2015 | Case study | Propose a swallowing therapy program for a PWS infant to transition from nasogastric tube to oral feeding. | One 2-month-old infant with PWS |
| Pérez Pérez | 2015 | Systematic Review | Critically analyze studies over the past 25 years on speech therapy interventions in PWS | Not Applicable |
| Barañón-Trujillo | 2015 | Literature review | Understand PWS, special educational needs, and behavior management for this population in the classroom. | Not Applicable |
| Rogers et al. | 2015 | Systematic Review | Highlight factors influencing early language development in intellectual disability and identify other relevant influences. | Not Applicable |
| Salles et al. | 2016 | Case study | Determine how individuals with PWS process multisensory information, including visual and auditory communication cues | 26 French-speaking PWS patients (10 M, 16 F), aged 27.6–33.5 years |
| Mourelo et al. | 2017 | Literature review | Establish evidence-based recommendations to unify care criteria for individuals with PWS. | Not Applicable |
| Salehi et al. | 2017 | Systematic Review | Characterize swallowing dysfunction and aspiration risk via videofluoroscopy in infants with PWS. | Not Applicable |
| Dianesi | 2018 | Experimental | Identify carriers of PWS in newborns with hypotonia using methylation analysis of 15q11-q13 | Two newborns under 1 month with neonatal hypotonia and clinical suspicion of PWS |
| Ramón Krauel et al. | 2019 | Literature review | Characterize PWS from multiple perspectives to support interdisciplinary care | Not Applicable |
| González Aboitiz | 2019 | Descriptive | Analyze language development in members of the Spanish PWS Association | 34 families of people with PWS (21 M, 13 F), aged 3–18 years |
| Pansy et al. | 2019 | Case study (Longitudinal / Prospective) | Describe early motor and prelinguistic verbal development in a PWS infant | One infant with PWS aged 2–27 weeks post-term |
| Lobo | 2020 | Case study | Carry out a case study involving speech therapy using play-based activities in a person with PWS | One male 20-year-old patient with PWS |
| McGregor) | 2020 | Bibliometric | Quantify research efforts directed at DLD compared to other neurodevelopmental disorders. | Not Applicable |
| García Álvarez | 2021 | Experimental | Design an intervention to achieve age-appropriate linguistic competence in students with PWS and spina bifida | One 11-year-old fifth-grader with PWS and one 10-year-old fifth-grader with lumbar myelomeningocele spina bifida |
| Llorente Rodríguez | 2021 | Experimental | Design an educational program to improve affective, mathematical, and linguistic development in children with PWS. | One 8-year-old primary school student with PWS |
| Miles et al. | 2022 | Bibliometric | Establish a standard protocol for quantitative VFSS analysis in children | Not Applicable |

### *Speech*

Most children with PWS exhibit delays in oral expression compared with the general population. Their first vocalizations typically occur between 6 and 7 months of age and are characterized by being monotonous and having a distinct harmonic structure. Articulatory difficulties include imprecision, reduced speech rate, and low volume (Atkin & Lorch, 2007; Pansy et al., 2019). These difficulties are often associated with the anatomical characteristics of oral structures involved in speech production, such as a narrow oral cavity, high-arched palate, and hypotonic tongue and lips (Caixàs Pedragós et al., 2014; Stein et al., 2006). Approximately 91% of children have some degree of motor deficit, ranging from mild to severe, which affects tongue mobility and coordination of the articulatory organs. Additionally, they experience difficulties with lip protrusion and retraction, alternate tongue movements, and overall articulatory coordination (Bellon-Harn, 2005; Misquiatti et al., 2011; Ramón Krauel et al., 2019). These impairments negatively impact speech intelligibility, and children with PWS often rely on gestures to communicate.

Regarding speech fluency, interjections are the most common type, accounting for 52.58% of disfluencies. Other frequent disfluencies include revisions (11.10%), partial word repetitions (10.84%), and whole word repetitions (9.85%). Less common disfluencies (≤5%) include phrase repetitions, prolongations, incomplete phrases, blocks, and broken words (Defloor et al., 2000). Despite the high frequency of disfluencies, children with PWS are generally not considered to have stuttering. Evidence indicates that although many individuals with PWS display disfluency, they do not typically present a clinical stuttering profile (Defloor et al., 2000; Van Borsel & Tetnowski, 2007).

### *Swallowing*

Feeding issues in newborns with PWS are a major clinical concern, and videofluoroscopic swallowing studies (VFSS) are frequently employed to assess swallowing safety. These studies have revealed a relationship between feeding dysfunction and hypotonia, which affects oral musculature, impairs coordination of the suck-swallow-breathe sequence, and reduces airway protective mechanisms (Miles et al., 2022; Salehi et al., 2017).

Salehi et al. (2017) described the swallowing physiology of 10 infants with PWS, reporting that 87% of VFSS trials showed aspiration with at least one consistency. Aspiration was more likely with thin liquids (84%) than with thickened liquids (60%) or purees (14%). Pharyngeal residue was observed in 71% of cases. Notably, all aspiration events in the infants were silent, which is particularly concerning, as clinical signs such as gagging or coughing may be absent. Silent aspiration can result in long-term respiratory complications, including lung damage, respiratory failure, infection, and asphyxia. Respiratory conditions are common in children with PWS, with respiratory illnesses accounting for 61% of deaths in this population (Salehi et al., 2017).

In speech-language therapy assessments, it is common to observe an absence of oral reflexes, including rooting, sucking, swallowing, extrusion, gagging, coughing, and vomiting. In some cases, the suction process may be weak or absent. Additionally, the tongue is often anteriorized, the palate is high-arched, and hypotonia affects the entire oral musculature. Functional impairments include slow orofacial movements and poor coordination of the suck-swallow-breathe sequence (Martín Santana, 2015). Chewing is typically unilateral, corresponding to the less hypotonic side, and is accompanied by compensatory perioral movements and increased chin tension (Lobo, 2020). Velopharyngeal dysfunction (VPD) is also frequently observed, with signs such as nasal regurgitation, which can compromise feeding safety (Crockett et al., 2014). In some cases, infants show limited interest in feeding (Pansy et al., 2019). Therefore, the management of feeding in children with PWS aims to ensure adequate nutrition, often requiring specialized feeding techniques, such as manual manipulation of the jaw and oral structures, as well as the use of specialized nipples and feeding tubes (Llorente Rodríguez, 2021).

Children with PWS also exhibit a high prevalence of obstructive sleep apnea. Adenotonsillectomy is commonly performed as a surgical treatment; however, VPD is a known postoperative complication. Consequently, additional speech-language evaluations and interventions are required prior to surgery. Speech-language therapists should assess orofacial function post-surgery, documenting structural characteristics and changes in oral motor function (Saeves et al., 2011).

Alongside feeding safety issues, hyperphagia typically emerges between the first and second year of life and is considered a hallmark feature of PWS. Children develop compulsive behaviors related to food, exhibiting excessive concern with eating, continuously seeking food, consuming unusual substances, and throwing temper tantrums when access to food is restricted (Pituch et al., 2010). The underlying cause of hyperphagia is not fully understood but is thought to involve hypothalamic dysfunction, particularly in the arcuate nucleus, which regulates hunger and satiety. This dysfunction impairs autonomous regulation of feeding behavior (Caixàs Pedragós et al., 2014).

Feeding patterns in children with PWS often include open-mouth, immature chewing and swallowing, anterior tongue positioning, and tongue-thrusting over the teeth during swallowing (Atkin & Lorch, 2007; Zambotti & Souza, 2013). Additionally, many children chew and suck on objects, and drooling is common (Saeves et al., 2011). These children also experience difficulty vomiting, sometimes requiring pharmacological intervention. When medications are ineffective, doses may be increased, potentially leading to toxicity without eliciting vomiting. Therefore, environmental control is the primary measure to prevent choking, aspiration, or foreign-body-related infections (Barañón-Trujillo, 2015).

***Language***

Most people with PWS exhibit impairments in both receptive and expressive language, with expressive language being more severely affected—a pattern commonly observed in genetic disorders (McGregor, 2020; Rogers et al., 2015). Expressive deficits include delays in phonological, syntactic, grammatical, and pragmatic skills (Dimitropoulos et al., 2013; Pérez Pérez, 2015; Ramón Krauel et al., 2019). Since comprehension tends to be relatively preserved, early stimulation should target this domain. Regarding expression, it is recommended that children learn to use gestures as a means of communication during language acquisition (Caixàs Pedragós et al., 2014; González Aboitiz, 2019). In some cases, language difficulties are so severe that children do not develop spoken language and rely exclusively on gestures. Despite the lack of speech, their comprehension is relatively strong, allowing them to organize stories sequentially, recognize body schema, primary colors, basic shapes, and concepts of quantity and size, as well as demonstrate appropriate temporal and spatial orientation (Lobo, 2020; Mourelo et al., 2017).

Language difficulties are evident from early childhood, with delays appearing across multiple language components. First words typically emerge between the ages of 4 and 5. Initial constructions are generally two-word phrases accompanied by gestures and pointing, often replacing complete sentence constructions, which may not appear until as late as 20 years of age (Dimitropoulos et al., 2013; Lobo, 2020; Stein et al., 2006).

The phonetic-phonological component is frequently affected in children with PWS, with approximately 85% showing phonological impairments ranging from mild to severe; younger children tend to exhibit more severe deficits (Lewis et al., 2002). These impairments manifest in several ways, such as delays in phoneme acquisition, with initial sounds typically emerging around the age of 3 years (Lewis et al., 2002). Another characteristic is the occurrence of common errors, such as omissions, substitutions, and distortions, in initial and/or final phoneme positions (Arconada, 2012; González Muñoz, 1999, cited in González Aboitiz, 2019).

Additional phonological difficulties include rhotacization and the loss of occlusive or fricative characteristics in voiceless stops and fricatives, with the production of /r/ being particularly challenging. Consonants produced in prepalatal and alveolar positions, as well as fricative and plosive sounds, are among the most frequently affected. Despite these difficulties, most children with PWS retain a phonological inventory that includes vowels, diphthongs, and most consonants (Defloor et al., 2002). Evidence indicates that phonological difficulties tend to improve with age, whereas articulatory errors, including sound distortions, persist in 18–29% of cases. In other words, as children age, phonological deficits gradually resolve, but phonetic difficulties remain (Saeves et al., 2011).

Lexical performance is variable; some children display a performance within the expected range for their age (Dimitropoulos et al., 2013). In a study involving parents of children with PWS, 11.8% reported that their child had an abundant vocabulary, 44.1% reported age-appropriate vocabulary, and 44.1% reported limited vocabulary (González Aboitiz, 2019).

Morphosyntactic difficulties include gender and number agreement errors and challenges in constructing simple sentences (subject + verb + complement). Most children (47.1%) begin producing two- to three-word combinations between 1 and 3 years of age (Mourelo et al., 2017).

In the pragmatic domain, children with PWS often exhibit reduced auditory and visual attention spans, difficulties in interaction, apathy, social withdrawal, and inconsolable crying. They communicate intentions primarily through facial expressions, gestures, and limited vocalizations (Misquiatti et al., 2011). Speech may sometimes include repetitive features, reflecting perseveration. These characteristics suggest difficulties with speech output processing and pragmatic language use (Atkin & Lorch, 2007). In some cases, echolalia and unintelligible responses are observed, accompanied by limited and primitive play (Zambotti & Souza, 2013).

In addition, people with PWS often exhibit difficulties in reading comprehension, partly due to mechanical reading with incorrect intonation and cadence. They fail to pause appropriately at punctuation marks, resulting in monotonous and difficult-to-

understand reading. Moreover, making inferences from written text is challenging. Writing is also affected by reduced syllabic and phonemic awareness, leading to incorrect spelling and segmentation of words into syllables. Common errors include improper word spacing and mistakes in natural, rule-based, and arbitrary orthography (García Álvarez, 2021).

***Hearing***

People with PWS generally have hearing thresholds within normal limits (above 20 dB), which remain stable over time, as indicated by audiometric testing (Misquiatti et al., 2011). However, compared with typically developing individuals, they show a specific deficit in discriminating human voices from environmental sounds. In PWS, multisensory information does not converge or interact before initiating behavioral responses, suggesting that social behavioral deficits may partly stem from difficulty interpreting social cues conveyed through voice, facial expressions, or a combination of both. Neuroimaging studies indicate disturbances in the "social brain network" in PWS, including the superior temporal sulcus (STS), which is involved in processing human voices (Salles et al., 2016).

Interestingly, a neuroimaging study of a subject with PWS revealed a restricted set of hypoperfused cortical areas at rest. This included the anterior cingulate and cingulate cortex—regions associated with theory of mind and empathy—as well as the auditory area of the superior temporal gyrus, specialized for processing vocal and speech information. Furthermore, analysis of the relationship between emotional and behavioral disorders indicated that deficits in processing social information, such as vocal stimuli, are related to the severity of clinical and social impairments in PWS (Misquiatti et al., 2011).

***Voice***

The studied population often exhibits voice difficulties characterized by weak vocal intensity, high pitch, intermittent pitch changes, and a voice quality that may be choppy, rough, and/or hoarse (Defloor et al., 2001; Lewis et al., 2002; Pérez Pérez, 2015). Issues like constant or intermittent nasal emission on pressured sounds and a general quality of hypernasality are particularly notable. Articulatory assessments have found nasal airflow associated with the production of fricatives, affricates, and occasionally plosives, contributing to the nasal distortion of these sounds (Branson, 1981 as cited by Defloor et al., 2001). These issues are directly related to hypotonia, which results in insufficient velopharyngeal closure, and orofacial anomalies may further exacerbate resonance difficulties (Munson, 1998, cited by Defloor et al., 2001; Bellon-Harn, 2005).

Voice quality in PWS is comparable to that observed in patients with flaccid dysarthria, where hypernasal resonance is a characteristic feature. Anomalies in pitch and resonance are likely associated with muscular hypotonia and endocrine dysfunction, resulting in oral motor dysfunction (particularly inadequate velopharyngeal closure and movement) and deficient laryngeal growth, respectively (Kleppe et al, 1990, cited in Defloor et al., 2001; Ramón Krauel et al., 2019).

Overall, individuals with PWS tend to perform poorly on aerodynamic measures. Children, adolescents, and adults have been found to score more than two standard deviations below the mean for MPT (Munson, 1988, as cited in Defloor et al., 2001). Although they are capable of generating sufficient oral airflow to sound horns or whistles, their MPT has been observed at only five seconds, which is far below the expected adult value (Bellon-Harn, 2005).

Finally, snoring in people with PWS is often abnormally loud, which may be associated with severe respiratory symptoms (Saeves et al., 2011).

## DISCUSSION

The findings of this study highlight the need for a comprehensive approach to support people with Prader-Willi Syndrome (PWS). This syndrome not only affects overall motor development but also profoundly impacts oral functions related to speech, swallowing, voice, and language. The analysis indicates that many of the observed difficulties in PWS originate from hypotonia, which manifests in the first months of life and persists throughout development.

Central hypotonia not only explains delayed motor milestones but also affects the efficiency of sucking, swallowing, and the development of complex motor patterns necessary for oral speech production. These results are consistent with Salehi et al. (2017), who directly link muscle weakness with swallowing dysfunction. Moreover, there is a high incidence of silent aspiration and poor coordination of the suck-swallow-breathe triad, emphasizing the importance of early speech-language therapy intervention.

The above aligns with the management guidelines for patients with PWS, a consensus statement by experienced clinicians in the United Kingdom and Ireland, which notes that this population exhibits delays in oromotor and swallowing skills. Accordingly, speech-language therapists play a critical role in their development, beginning in early childhood and continuing

throughout life as part of a multidisciplinary team (Shaikh et al., 2024).

Regarding speech, the data reveal an impaired phonetic profile, characterized by frequent articulation errors (omissions, distortions, substitutions) and reduced intelligibility, which often requires compensatory gestural communication. This, coupled with uncoordinated articulatory movements, confirms the significant functional impact of hypotonia on speech and underscores the need for therapeutic interventions aimed at improving oral motor control.

Although disfluencies are present in PWS, they do not constitute stuttering. This distinction is important because it identifies disfluency as a structural feature of the PWS speech profile rather than a disorder with psychomotor or emotional origins, as seen in other clinical populations.

Language comprehension is generally more preserved than expression, with expressive language being severely affected (Dimitropoulos et al., 2013; Rogers et al., 2015). Phonological, morphosyntactic, and pragmatic difficulties reflect a delay in language acquisition that, in many cases, may remain at pre-verbal levels. The presence of echolalia, perseveration, and compensatory gestures supports the hypothesis that PWS constitutes a neurological and motor-based language disorder in which speech processing and control systems are compromised. These findings are consistent with Westby (2014), who reports that linguistic skills in PWS, both comprehension and expression, are significantly impaired, while verbal intelligence remains relatively preserved.

Additionally, although auditory thresholds in children with PWS are within normal limits, there is a notable deficit in social auditory cue discrimination, particularly with human voices. This finding is relevant because it links auditory processing deficits with the pragmatic and social difficulties observed in this syndrome.

The voice domain exhibits a pattern of hypernasality, abnormal pitch, reduced vocal intensity, and maximum phonation times (MPT) well below the average, comparable to a flaccid dysarthria profile. These vocal impairments directly impact speech intelligibility and communicative effectiveness. Thus, they should be considered a secondary clinical manifestation arising from generalized hypotonia and orofacial anatomical anomalies in PWS.

Finally, the relationship between early swallowing difficulties and the subsequent emergence of hyperphagia constitutes a clinical paradox in PWS. While infants initially show low interest in feeding, in later stages this interest becomes obsessive, potentially placing the patient at risk of choking, aspiration, or infection. These observations are consistent with Bravo et al. (2021), who outline a nutritional trajectory in individuals with PWS. They first describe feeding difficulties in newborns due to a weak sucking pattern, leading to secondary malnutrition and apathy toward feeding. This is followed by a phase of normalized appetite, and ultimately, gradual weight gain and development of hyperphagia, resulting in obesity.

The findings of this study emphasize the need for comprehensive and periodic speech-language therapy assessments in patients with PWS, ideally starting from birth. Early and continuous intervention can mitigate long-term functional consequences, enhance quality of life, and support the development of communication. Therefore, it is essential to incorporate these clinical characteristics into the diagnostic and follow-up profile of this syndrome, promoting individualized interventions that address motor, sensory, cognitive, and emotional components of communicative development.

Regarding the nature of studies on PWS, it is noteworthy that most research in this area has provided clinical descriptions of the participants, reflecting a medical/rehabilitative model of disability in which disability is located within the biological body. Consequently, the strategies proposed are focused on rehabilitating structural and functional deficiencies that "disable" people in order to normalize their functioning. The theoretical framework of research must incorporate a systemic model, which would allow for a comprehensive analysis of the subjects. This approach would not only address people with PWS from their symptomatology (Allen, 2011) but also encourage reflection on the types of support that can be provided from familial, social, and political contexts.

In line with this, Currie et al. (2024) highlight the pathologization and medicalization experienced by patients with such conditions, as well as the isolation, lack of medical support, and pressure to “be normal” that mothers of children with PWS often encounter. Therefore, it is pertinent to conduct research that moves beyond the medical characterization of PWS and explores the broader lived experiences of this population and their families.

The primary limitation of this study is the restricted amount of available information, despite considering multiple sources over a 22-year period. This scarcity may be due to the low prevalence of PWS, which can restrict research interest. Consequently, it is imperative to conduct further studies to generate accurate,

evidence-based information that supports optimal speech-language intervention from a comprehensive and critical perspective.

## CONCLUSION

This research shows that Prader-Willi Syndrome (PWS) is a complex genetic condition that affects multiple areas of communicative development, with hypotonia being the central pathophysiological factor underlying many of the observed difficulties in speech, swallowing, and voice. It is also evident that oral motor impairments not only affect feeding but significantly compromise speech production and communicative effectiveness, highlighting the need for early, continuous, and specialized speech-language therapy assessment and intervention.

In conclusion, people with PWS require a transdisciplinary approach, where speech-language therapy takes a fundamental role within the care team, from the earliest stages of life and throughout development. Furthermore, it is recommended to adopt a critical and inclusive perspective that goes beyond the traditional medical/rehabilitative model. This perspective should consider the social, familial, and emotional dimensions of this population, and promote contextual support that enhances participation and quality of life.

Ultimately, further research is necessary to establish a strong theoretical and clinical framework for addressing PWS. Given its low prevalence, academic and scientific interest in this area has been limited. Robust research will enable the design of evidence-based interventions that are sensitive to the complexity of this condition and oriented toward the real well-being of people with PWS and their environment.

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