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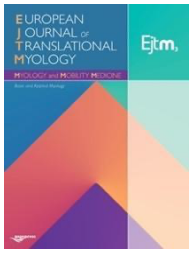

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Postural-motor development, spinal range of movement and caregiver burden in Prader-Willi syndrome-associated scoliosis: an observational study

Maria Chiara Maccarone,[1] Mariarosa Avenia,[2] Stefano Masiero[1,2]
[1]Rehabilitation Unit, Department of Neuroscience, University of Padua, Padua, Italy; [2]Physical medicine and rehabilitation School, Department of Neuroscience, University of Padua, Padua, Italy.

Correspondence: Maria Chiara Maccarone, Physical Medicine and Rehabilitation School, Department of Neuroscience, University of Padua, 35128 Padua, Italy.
E-mail: mariachiara.maccarone@phd.unipd.it
ORCID ID: 0000-0003-2793-1334

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Maria Chiara Maccarone
E-mail: mariachiara.maccarone@phd.unipd.it
ORCID ID: 0000-0003-2793-1334

Mariarosa Avenia
E-mail: mariarosa.avenia@gmail.com
ORCID ID: 0009-0008-2443-4221

Stefano Masiero
E-mail: stef.masiero@unipd.it
ORCID iD: 0000-0002-0361-4898

Abstract

Prader-Willi syndrome (PWS) is a rare genetic disorder characterized by hypothalamic dysfunction, hypotonia, cognitive deficits, and hyperphagia, primarily resulting from genetic abnormalities on chromosome 15. Among its varied manifestations, musculoskeletal issues, notably scoliosis, pose important challenges in management. This study aims to investigate differences in postural-motor development and spinal range of movement between preadolescents and adolescents with PWS, with and without scoliosis, while also exploring the potential impact of scoliosis on caregiving burden, an aspect yet to be thoroughly explored in existing literature. This observational study evaluated 13 individuals diagnosed with PWS, including 5 with scoliosis (PWS-Sc) and 7 without (PWS-NSc). Inclusion criteria comprised ages 8 to 18 years, confirmed PWS diagnosis through genetic testing, and scoliosis diagnosis. Anamnestic data, physical examinations, and surface measurements were collected, along with parental burden assessments using the Zarit Burden Interview (ZBI). Both groups displayed delays in achieving postural-motor milestones, with the PWS-Sc group exhibiting a more

pronounced delay, although statistical significance was not achieved. The main curve magnitude in the PWS-Sc group averaged 31.5° Cobb, with 60% of cases presenting an S-shaped curve. Surface measurements of physiological curves did not differ significantly between groups, but the scoliosis-affected group exhibited lower lumbar extension values ($p=0.04$). The overall ZBI revealed higher scores in the PWS-Sc group, although statistical significance was not reached. However, significant differences were observed in single questions score evaluating aspects such as social life and caregiver uncertainty ($p=0.04$ and $p=0.03$, respectively). Despite the small sample size, delays in achieving postural-motor milestones, particularly in individuals with scoliosis, were observed. The differences recorded in lumbo-pelvic movement suggest that tailored interventions may be beneficial. The heightened caregiving burden in the scoliosis group underscores the need for targeted support. Early intervention and ongoing monitoring should be important for accurate diagnosis and appropriate care, potentially with psychological support for caregivers.

Introduction

Prader-Willi syndrome (PWS) is a rare genetic disorder characterized by hypothalamic and pituitary dysfunction, hypotonia, cognitive deficits, and hyperphagia.[1,2] Imprinting-related mechanisms play a crucial role in its transmission. Genes on chromosome 15's long arm are physiologically imprinted with the maternal copy silenced and only the paternal copy expressed. PWS results if the paternal copy is missing, defective, or incorrectly silenced.[3] Paternal deletion of the 15q11-q13 region is the most common genetic error (75%), followed by maternal uniparental disomy (24%) and imprinting defects (1%).[4]

Hypothalamic dysfunction in PWS leads to pituitary hormone insufficiency, primarily growth hormone (GH), followed by luteinizing hormone (LH), follicle-stimulating hormone (FSH), and thyroid-stimulating hormone (TSH). Short stature is prevalent due to GH and gonadotropin deficiency.[1] Additionally, PWS may present central adrenal insufficiency, central hypothyroidism, reduced glucose tolerance, and diabetes mellitus, especially in adults with significant obesity.[1,5]

Common musculoskeletal findings in PWS include a high prevalence (40-80%) of scoliosis, along with hyperkyphosis, cifoscoliosis, flat or pronated feet, varus or valgus knees, ligament laxity, hip dysplasia, coxa valga, valgus knee, limb length discrepancy, and marked external

rotation of the lower limbs, contributing to a distinctive wide-based and externally rotated posture. Gait patterns often present as swaying or ataxic.[6]

The prevalence of scoliosis in individuals with Prader-Willi syndrome (PWS) ranges from 15% to 86%. Studies categorizing scoliosis based on onset age reveal 23% prevalence in infants (0-2 years), 29% in young children (3-9 years), and 80% in adolescents (10-17 years).[7,8] This distribution indicates an early peak likely linked to muscle hypotonia and a later peak during adolescence.[9] Females exhibit a higher incidence than males, with the risk of curve progression similar for both sexes.[7,9] Some studies suggest a slightly increased risk in individuals with maternal uniparental disomy of chromosome 15, but findings vary.[10] Most PWS-related scoliosis involves lumbar or thoracolumbar curves, and in particular the "C-shaped curve" characterized by a single curvature in the spine.[8]

Risk factors for curve progression in PWS differ from those identified for idiopathic scoliosis. These factors include prolonged hypotonia of the paravertebral muscles, which continues for an extended period after birth, failing to provide adequate support to the spine when the child starts assuming an upright position and walking. Additionally, obesity could contribute to increased mechanical load on bones and joints, potentially leading to postoperative complications if spinal surgery becomes necessary.[11-13]

Scoliosis represents a challenging management issue for individuals with PWS, not only due to the intrinsic characteristics of the condition but also owing to difficulties in adhering to prescribed treatments, intensified by behavioral disturbances in this population.[14]

This study aims to identify anamnestic and clinical differences in postural-motor development, as well as spinal range of movement, between a population of preadolescents and adolescents with PWS presenting scoliosis and a population of PWS individuals without scoliosis. Additionally, it explores whether scoliosis onset in individuals with PWS leads to increased caregiving burden, an emerging area of interest in current literature.

Materials and Methods

Study design

This study employs an observational design.

Population

The study cohort comprised 13 individuals diagnosed with PWS, including 5 subjects with scoliosis (PWS-Sc) and 7 subjects without a scoliosis diagnosis (PWS-NSc). Participants diagnosed with PWS were recruited from the Pediatric Endocrinology Division Outpatient

Clinic at the Department of Women's and Children's Health - University of Padua, Padua, Italy, spanning from September 2020 to June 2021.

The inclusion criteria comprised patients of both sexes within the age range of 8 to 18 years, with a confirmed diagnosis of PWS through genetic testing. For the scoliosis group, inclusion required a diagnosis of scoliosis based on the Scoliosis Research Society (SRS) criteria, confirming a Cobb angle of at least 10° in an anteroposterior standing spine X-ray. Conversely, exclusion criteria comprised the absence of informed consent from parents or legal guardians and the presence of vertebral malformations.

Initially, the study procedures and objectives were elucidated to the patients and their parents. Following the acquisition of their consent, along with parental consent, a thorough collection of medical history and a physical examination were conducted. Subsequently, caregivers underwent a telephone interview utilizing a questionnaire to assess the caregiving burden.

The study was carried out in compliance with the principles outlined in the Helsinki Declaration.

Demographical and postural-motor development data collection

The collection of anamnestic data was performed at the Rehabilitation Unit, Department of Neuroscience, University of Padua, Padua, Italy and involved a thorough investigation covering various aspects such as gender identification, date of birth, and evidence of reduced fetal growth, distinguishing between intrauterine growth restriction (IUGR) or small size for gestational age (SGA). Additionally, the data collection involved details on the type of delivery (natural or cesarean) and the presence of hypotonia at birth and in the initial months of life, with specific inquiries about difficulties in crying and sucking during the newborn stage. Further specifics included the age in months at achieving major psychomotor milestones. Details about scoliosis were also gathered, encompassing any prior diagnosis, age of onset, and X-ray history.

Physical examination and spinal range of movement assessment

The clinical assessment was performed using Inclimed® to measure static and dynamic surface parameters of spinal curves.[15] For static measurements, Inclimed® was placed at specific landmarks (T1, T12, and S2) on the patient's back while standing relaxed. T1 was positioned below C7 in the interspinous space, T12 at the maximum thoracolumbar inclination point, and S2 at the level of the line connecting the upper posterior iliac spines. Angular measurements at

T1 and T12 provided the thoracic kyphosis angle, while those at T12 and S2 yielded the lumbar lordosis angle.[16]

The study also involved measuring lumbo-pelvic flexion-extension and lateral inclination range of movement (ROM). Participants were asked to perform maximum trunk flexion, extension and lateral inclination using the double Inclimed®, a pair of inclinometers fixed in a support and placed at T12 and S2 landmarks. The acquired angles were then utilized to calculate net lumbar flexion (lumbosacral flexion ROM minus sacral flexion ROM) and net lumbar extension (lumbosacral extension ROM minus sacral extension ROM; Figure 1).[16]

Caregiver burden

Parental burden was assessed using the Zarit Burden Interview (ZBI), a self-administered questionnaire comprising 22 questions scored on a Likert scale. The questions explore various aspects, including physical and psychological well-being, financial resources, social life impact, relationships, and the caregiver's connection with the individual with the disease. Scores range from 0 (indicating no burden) to 88 (signifying very high burden).[17] In a previous study the ZBI has been demonstrated to be a good predictor of the impact of PWS on many aspects of the caregiver quality of life and thus could be considered an efficient instrument to capture the global impact of PWS on the caregiver.[18] The telephonic interview, lasting around 15 minutes, ensured anonymity in data collection.

Statistical analysis

A dedicated database was created for each study participant, including anamnestic data, objective examination results, radiographic data, and parents' questionnaire responses. Anthropometric values were statistically analyzed for differences between quantitative variables in the PWS-Sc and PWS-NSc groups using the Student's t-test for symmetrically distributed values and the Wilcoxon test for asymmetrically distributed values.

For the ZBI questionnaire responses, being non-parametric variables, the Mann-Whitney test was employed.

Statistical significance was considered when $p < 0.05$. Statistical analyses and graph construction were conducted using Microsoft Excel and Jasp 0.16.04, an open-source program for classical and Bayesian statistical analysis provided by the University of Amsterdam.

Results

Demographic characteristics and postural-motor development data

The PWS population consisted of 10 females (76.90%) and 3 males (23.10%), with an average age of 11.61 ± 2.63 years (ranging from 8 to 15 years). In the PWS-Sc group, there were 3 males and 2 females, while in the PWS-NSc group, there were no males and 8 females. The average age for PWS-Sc was 13.5 years (ranging from 10 to 15 years), and for PW-Nsc, it was 10.5 years, with no statistically significant difference ($p = 0.06$). Regarding BMI, there were no statistically significant differences between the two groups ($p = 0.32$), with an overall BMI of 21.9 (±2.72). The PWS-Sc group had a BMI of 21.3 (range: 17.5-25.4), while the PWS-NSc group had a BMI of 22.4 (range: 19.19-26.38).

During pregnancy, 53.84% of the participants (7 out of 13) manifested diminished intrauterine growth, with 71.14% receiving an IUGR diagnosis, 14.4% falling into the SGA category, and 14.4% unable to specify the type of growth delay. Specifically, the average birth weight in the PWS-Sc group was 1.93, while in the PWS-NSc group, it was 2.25, with no statistically significant differences ($p= 0.74$).

Among the participants, 7.69% experienced natural delivery (only one subject in the PWS-NSc group), while 92.31% were delivered via cesarean section. All subjects displayed hypotonia at birth, coupled with difficulty crying and sucking (100% prevalence).

In terms of achieving postural-motor milestones, the entire cohort displayed a deviation from the typical developmental timeline observed in unaffected children. Notably, the group with scoliosis showed a more pronounced average delay in reaching these milestones, although statistical significance was not achieved in the two-tailed T-test, except for crawling (see Table 1).

Individuals with both PWS and scoliosis exhibit an average main curve magnitude of 31.5° ± 18.3° Cobb. Among the 5 subjects in the PWS-Sc group, 3 developed scoliosis during adolescence (between 12-14 years), while 2 developed it in infancy (at 5 and 6 years old). Sixty percent of cases (3 out of 5 subjects) manifest an S-shaped curve, while 40% (2 subjects) exhibit a C-shaped curve. In terms of the main curve location, 40% of cases (2 subjects) were at the thoracolumbar level, with 1 case presenting a right convex thoracolumbar curve and another showing a left convex thoracolumbar curve. The remaining 60% (3 subjects) had the main curve at the lumbar level with left convexity. Therapeutically, 40% of subjects (2 patients) engaged in therapeutic exercises, 60% (3 patients) used a brace, and 20% (1 patient) underwent successful corrective scoliosis surgery.

Physical examination and spinal range of movement assessment

Surface measurements of physiological curves did not exhibit significant differences between the two groups. Specifically, the mean value of thoracic kyphosis observed in the population was 37.84° ± 12.89, with a thoracic hypokyphosis of 40° in the PWS-Sc group and of 36.5° in the PWS-NSc group. The measured lumbar lordosis was 39.84° ± 8.92, with an average value of 40.8° in the PWS-Sc group and of 39.5° in the PWS-NSc group.

In the dynamic measurements of lumbar excursion in both flexion and lateral bending, no differences were found in both groups. However, a statistically significant difference in spine extension was observed, with the scoliosis-affected group exhibiting lower values (Table 2).

Caregiver burden

Out of 13 parents of subjects in the study group, 12 agreed to participate in the telephone interview. Eleven mothers, reporting to be the primary caregivers in terms of time spent caring for the family member, and one father, who mentioned that the main caregiver was his wife, responded to the questionnaire.

On average, considering the entire study population, the mean ZBI score was 41.81 points, equivalent to a caregiving burden ranging from moderate to severe. The PWS-NSc group scored a total of 36.33 (ranging from 21 to 45), indicating a caregiving burden from mild to moderate. In contrast, the PWS-Sc group scored 48.4 (ranging from 30 to 59), approaching statistical significance without, however, reaching it (p= 0.07).

Considering the questions one by one, the PWS-Sc group showed higher scores approaching statistical significance for the negative impact of caregiving on the caregiver's health (p=0.07) and the feeling of embarrassment due to the family member's behaviors (p=0.06). In both groups, we recorded a burden related to concerns about the family member's future and the need for additional financial resources for caregiving. Statistically significant variations were observed in aspects such as social life (Do you feel that your relative currently affects your relationship with other family members or friends in a negative way? p=0.04), and in questions like "Do you wish you could just leave the care of your relative to someone else?" (p=0.01) or "Do you feel uncertain about what to do about your relative?" (p=0.03).

Discussion

Prader-Willi Syndrome (PWS) is a genetic disorder characterized by various physical, cognitive, and behavioral manifestations, and understanding its multifaceted impact is essential for addressing the challenges faced by individuals with PWS and their caregivers. Scoliosis is

one of the major concerns for PWS patients and therefore there is a need for regular systematic monitoring of spinal deformity from pediatric age.[19]

The findings of our study confirm a delay in achieving postural-motor developmental milestones among individuals with PWS, consistent with existing literature.[20-22]

Monotonous early motor and verbal behaviors in infants with PWS have been reported.[22] Monitoring milestones achievements and verbal behaviors in infants with PWS may reveal specific patterns and precursors of later development.[22] In our study the PW-Sc subgroup exhibited a more pronounced delay, suggesting potential factors such as increased axial hypotonia or impaired axial coordination and proprioception.[14] Moreover, surface measurements of dynamic spine ROM revealed a notable reduction in lumbar extension in the scoliosis group. This reduction may be attributed to a deficit in core muscle strength during trunk extension.[23] These alterations are documented in the literature to persist into adulthood. Adults with PWS exhibit increased thoracic kyphosis but reduced lumbar lordosis, along with diminished movements in lumbar flexion, lumbar extension, lumbar lateral flexion, and hip flexion and extension compared to those with normal weight.[24] Therefore, early recognition of these limitations can enable interventions with appropriate exercise programs to enhance trunk mobility. In addition, exercises for postural control in the sagittal plane should also be recommended. Encouraging activities that enhance core muscle control, such as swimming, dancing, or gymnastics, may also be beneficial.[9]

The limitations in mobility can also translate into a higher psychological burden on the caregivers of these patients. Caregivers of individuals with PWS can face a substantial burden, emphasizing the need for targeted support strategies.

All participants in our study scored above 20 points on the ZBI, with a mean ZBI score of 41.81 points. According to developers, a score of 20 is the threshold for requiring support. Notably, in our study caregivers of PWS scoliosis subjects scored higher averages across several domains, highlighting areas of increased concern, particularly in the personal and relational spheres. The concomitant differences in gender distribution between the two groups do not allow definitive conclusions on whether scoliosis determined the increased caregiving burden, contributed to it, or was incidental. However, this study identified key areas requiring monitoring and intervention, emphasizing the need for targeted support for caregivers of individuals with PWS, particularly those developing scoliosis. Kayadjanian *et al.* reported that caregivers of individuals with PWS experienced a significant burden, with an average ZBI score of 44.4 ± 15.4. Higher scores were noted for caregivers of teenagers and young adults with PWS, while lower scores were observed for older adults and the youngest age group.

Caregivers reported negative impacts on their romantic relationships, work, sleep, and mood due to caring for someone with PWS. Interestingly, caregiver burden in PWS appeared to be independent of income level and was weakly correlated with the amount of assistance received. Notably, caregiver burden in PWS exceeded that observed in caregivers for conditions such as dementia and traumatic brain injury.[18] Our findings reconfirm the need for interventions to alleviate caregiver burden, which could serve as a secondary endpoint in treating PWS. It should be advisable in clinical practice to screen caregiver burden, identifying cases of greater need and guiding caregivers to competent professionals addressing critical areas.[25] This screening should be particularly needed when proposing interventions that may impact family dynamics, such as prescribing a brace for conservative scoliosis treatment.[26]

However, some limitations should be acknowledged. In our population, patients with obesity were poorly represented. Increasing the sample size and monitoring over time could help evaluate the influence of obesity on the clinical history of scoliosis and musculoskeletal alterations in general. In addition, the sample size was relatively small in our study, limiting the generalizability of the findings. To confirm obtained data, future research should focus on expanding the sample size, adopting a longitudinal approach, and collaborating with pediatric endocrinology and relevant associations to further investigate the impact of musculoskeletal alterations on caregiving burdens. Continued interdisciplinary collaboration should be prioritized for the global management of individuals with PWS, guiding intervention strategies and support programs tailored for these subjects.

Conclusions

Our study highlighted the potential interplay between PWS, scoliosis, and the associated caregiving burden. Our findings, despite the small sample size, indicate a significant delay in achieving postural-motor milestones in individuals with PWS, particularly in those with scoliosis. Lumbo-pelvic movement analysis revealed differences in extension, suggesting potential implications for core muscle training programs in scoliosis-affected individuals. Finally, the caregiving burden was higher in the scoliosis group, emphasizing the need for targeted support programs. Therefore, the study underscored the importance of considering early intervention and long-term monitoring in this patient population to make a correct diagnosis and offer appropriate care, enhancing the well-being of individuals with PWS. Alleviating the caregiving burden on families may require psychological support for caregivers. Therefore, continued collaboration among diverse medical specialties and

professional figures should be necessary for a comprehensive and effective PWS management approach.

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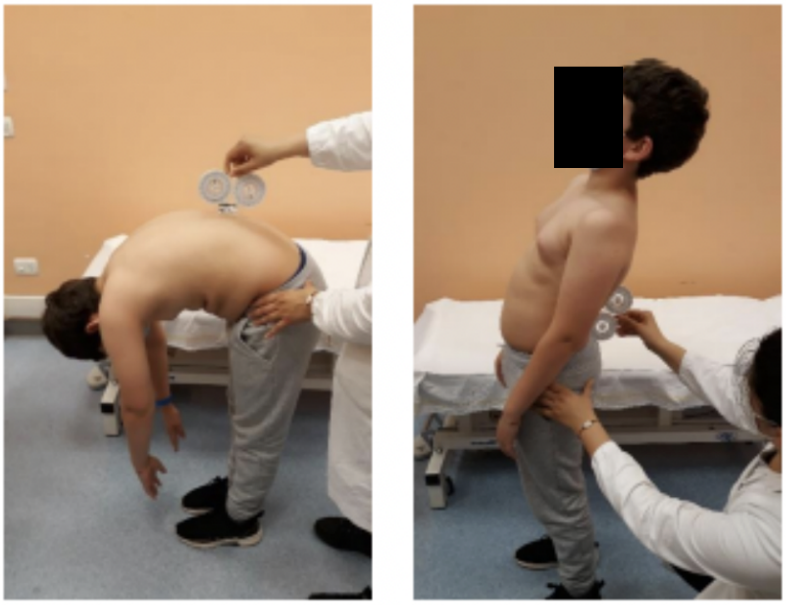

Figure 1. Measurement of the range of motion (ROM) of lumbar flexion and extension.

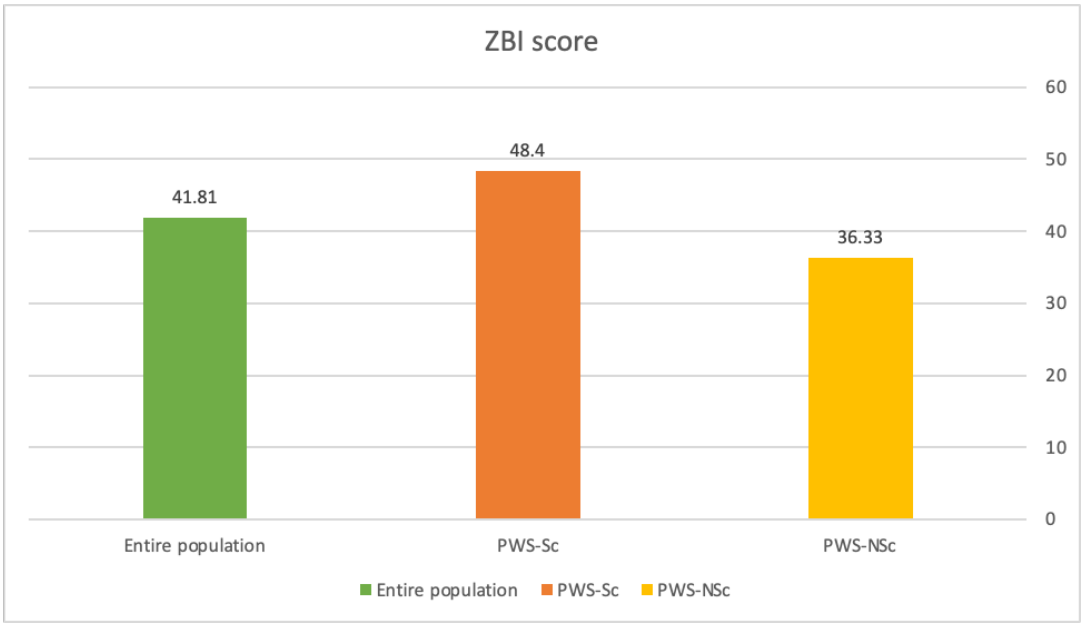

Figure 2. The graph shows the findings related to Zarit Burden Interview (ZBI) scores in the study population. The mean ZBI score for the entire population indicates a caregiving burden

ranging from moderate to severe. Within the PWS-Sc group, the score reflects a mild to moderate caregiving burden, while the PWS-Sc group shows a higher score (p= 0.07).

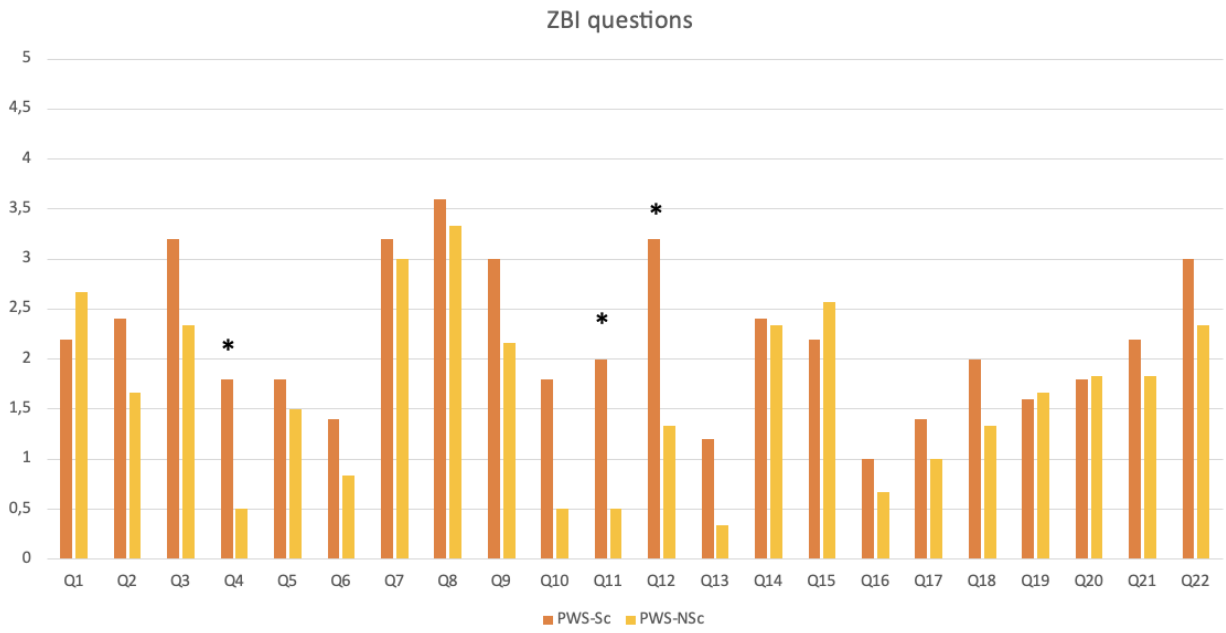

Figure 3. The graph represents the average values obtained for each question in the group of patients with Prader-Willi syndrome (PWS) and scoliosis (PWS-Sc) and in the group with PWS but without scoliosis (PWS-NSc). The asterisk (*) denotes a statistically significant difference ($p < 0.05$).

Milestone	**Mean age in months (standard deviation)**			**p-value**
	Entire population	**PWS-NSc group**	**PWS-Sc group**	
Head control	7.6 (±4.2)	7.1 (±4.2)	8.6 (± 4.2)	0.11
Independent sitting	11.2 (± 4.2)	10.1 (± 4.2)	13.5 (± 4.2)	0.07
Standing onset	18.5 (±6.5)	17.5 (±6.5)	19 (± 6.5)	0.15

Crawling	16 (±4.4)	13 (±4.4)	20 (± 4.4)	0.01*
Independent walking	21.5 (±5.9)	20.4 (±5.9)	23.6 (± 5.9)	0.18

Table 1. The table summarizes developmental milestones, including the mean age (in months) for achieving head control, independent sitting, standing onset, crawling, and independent walking among our population. The values are presented for the entire group, distinguished by those with Prader-Willi syndrome (PWS) without scoliosis (PWS-NSc) and those with both PWS and scoliosis (PWS-Sc). The symbol (±) indicates the standard deviation. The asterisk (*) denotes a statistically significant difference ($p < 0.05$).

	PWS-Sc	**PWS-NSc**	**p-value**
Right inclination	19.2°	21.25°	0.59
Left inclination	21.6°	22.5°	0.83
Lumbar flexion	52.4°	58°	0.66
Lumbar extension	5.5°	22.5°	0.04*

Table 2. The table includes measurements of Range of Motion (ROM) in different planes of movement. The p-value column indicates the p-values for the statistical analysis of the differences between PWS-Sc and PWS-Nsc patients. The asterisk (*) denotes a statistically significant difference ($p < 0.05$), with PWS-Sc having less ROM than PWS-Nsc in the lumbar extension measurement.