

Paramedics' knowledge of the symptoms and prehospital care of patients with Prader-Willi and Angelman syndromes

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Abstract

Introduction: Rare genetic disorders are conditions in which the emergency medical team's knowledge of characteristic clinical and phenotypic symptoms can significantly improve the quality of the provided medical care.

Aim of the research: This study focuses on assessing paramedics' knowledge of characteristic symptoms that may influence the emergency procedures in patients with rare Prader-Willi and Angelman genetic syndromes. This topic was chosen due to the lack of guidelines for emergency care of patients with genetic disorders, posing a major challenge in medical practice.

Material and methods: The survey was conducted among 100 paramedics. The research took place at the Medical University of Warsaw in Poland. An author's survey questionnaire consisting of closed-ended single-choice and multiple-choice questions was used. Statistical analyses were performed using descriptive statistics and correlation analysis.

Results: Paramedics' overall knowledge of Prader-Willi and Angelman syndromes and related emergency procedures was low (around the 50th percentile in knowledge score statistics), and the results indicated a statistically significant deviation from the normal distribution (Shapiro-Wilk test, $p < 0.001$). Individuals of varying ages, work experience, workplace settings, educational backgrounds, and genders did not differ in their knowledge of the symptoms and emergency procedures related to these selected genetic disorders (Spearman's rank correlation, Mann-Whitney U test, Kruskal-Wallis test). The rising prevalence of genetic disorders highlights the growing need to address this issue in training and practice.

Conclusions: There is a need for education and continuous professional training for paramedics regarding general knowledge and emergency medical procedures for individuals with Prader-Willi syndrome and Angelman syndrome as rare genetic disorders.

Introduction

Adapting emergency medical procedures to the individual needs of the patient and the current conditions at the scene of the event is a key element in effective pre-hospital care. During the provision of assistance to the injured person, it is crucial to consider their specific anatomical and physiological characteristics, which may affect the course and effectiveness of the medical care and emergency actions taken.

Prader-Willi syndrome (PWS) results from disruptions in the 15q11–q13 region of chromosome 15 and may have the following genetic origins: paternal chromosome 15 microdeletion, maternal uniparental disomy of chromosome 15, and genomic imprinting defects. The prevalence of Prader-Willi syndrome in the population of the United States is 1 : 15,000/1 : 30,000 live births [1, 2]. The most observed clinical symptoms include muscle hypotonia, psychomotor developmental delay, a lack of satiety leading to hyperphagia (abnormally strong, incessant sensation of hunger or desire

to eat often leading to overeating), and, consequently, overweight and obesity. Additionally, hypogonadotropic hypogonadism is present, leading to infertility. In male patients, features such as a small penis, hypoplastic, non-wrinkled scrotum with low pigmentation, and cryptorchidism (unilateral or bilateral) are often observed, with cryptorchidism present in about 80% of cases. Structural changes in the central nervous system include ventricular system enlargement, shallowing of the Sylvian fissure, incomplete closure of the insula, and a decrease in the volume of the parietal and occipital lobes. The leading cause of death in PWS patients remains complications related to obesity. Psychiatric problems play an important role in the health status and quality of life of these patients. In the study of Xia *et al.* the medication groups of PWS that were used most frequently included psychotropics (29.41% of patients) [3]. Nevertheless, medical advancements have considerably extended the expected life span of individuals affected by this syndrome [1, 2, 4–6].

Angelman syndrome (AS) is most commonly caused by a microdeletion in the 15q11–q13 region on the maternally inherited allele, paternal disomy of chromosome 15, imprinting defects in the 15q11–q13 region, or mutations in the UBE3A gene, inherited from the mother. The prevalence of Angelman syndrome is 1 : 15,000 live births [7]. Clinical symptoms typically manifest before the age of 3 years and include ataxia, profound intellectual disability, significant speech delay, general psychomotor developmental delay, and epilepsy. Characteristic phenotypic traits include a puppet-like gait, inappropriate laughter, microcephaly, and dysmorphic facial features such as widely spaced teeth and tongue protrusion. Common symptoms include microcephaly, early onset of seizures (before the third year of life), and abnormal EEG findings observed in over 80% of patients, usually in the first 2 years of life and often unrelated to epileptic seizures [7–11].

A comprehensive analysis of existing literature enabled the formulation of key emergency response – important factors for paramedics managing patients with the genetic disorders PWS and AS. Among the most essential recommendations are the implementation of the SAMPLE history protocol, clinical assessment utilising the Glasgow Coma Scale and the AVPU scale, blood pressure evaluation, initiation of airway management procedures including intubation and suctioning, and appropriate response to epileptic manifestations (Tables 1 and 2).

The SAMPLE interview is a medical interview commonly used by paramedics, which addresses the following issues: symptoms, allergies, medicines, medical history, last oral intake, and events leading to the present illness [12]. The lack of adequate knowledge of PWS and AS syndromes and their symptoms in the paramedic may disturb the correct assessment of the patient's condition. In individuals with Angelman syndrome, factors such as developmental delay,

intellectual disability, and speech impairments may significantly hinder the collection of a comprehensive SAMPLE history [9, 13, 14]. Similarly, in the case of children with Prader-Willi syndrome, paramedics should anticipate difficulties due to speech delays, aggressive behaviour, mood instability, and stubbornness, all of which may negatively impact the accuracy and completeness of the SAMPLE interview. In adolescents and adults with PWS, articulation disorders are commonly observed, further complicating the process of obtaining reliable medical history [15–17].

The Glasgow Coma Scale focuses on the assessment of 3 parameters on a point scale: eye opening, verbal response, and motor response. Dynamic changes in the score may suggest a deterioration of the patient's condition in the case of a decrease in points, while an increase may suggest an improvement in the patient's condition. In the context of medical rescue activities, obtaining 8 or fewer points is an indication to consider endotracheal intubation of the patient. The scales can be used in all patients over 4 years of age. In the case of paediatric patients up to 4 years of age, we use the paediatric Glasgow Coma Scale to assess the level of consciousness [12, 18]. In children with PWS, developmental delay should be taken into consideration when using the Glasgow Coma Scale (GCS) because confusion or inappropriate verbal responses may be inherent to the syndrome rather than indicative of acute pathology. Individuals with PWS often exhibit a high pain threshold, which may affect both problems with pain localisation and withdrawal responses [1, 6, 12, 18]. In AS, profound developmental delay and speech impairment, including minimal or absent verbal output, can significantly influence verbal assessment within the GCS. Furthermore, ataxia and motor coordination disturbances, frequently observed in AS, may alter the interpretation of motor responses within the scale [8–10].

Table 1. Symptoms of Prader-Willi syndrome and their relevance in Paramedic assessment and intervention

Symptoms	Impact on the work of the Paramedic
• Delays in speech development • Aggressive behaviors • Moodiness • Stubbornness	• May affect and disrupt the collection of valuable SAMPLE history
• Developmental delay • High pain threshold	• May lead to inaccurate assessment of the patient's condition using the Glasgow Coma Scale
• High pain threshold	• May lead to inaccurate assessment of the patient's condition using the AVPU scale
• Coexisting overweight and obesity	• Routinely measure blood pressure
• Triangular and small mouth with downward-turned corners • Thick saliva • Obesity	• Increased difficulty in endotracheal intubation
• Thick saliva	• Difficulty in oral suctioning

Table 2. Symptoms of Angelman syndrome and their relevance in Paramedic assessment and intervention

Symptoms	Impact on the work of the Paramedic
• Developmental delay • Speech disorders • Intellectual disability	• May affect and disrupt the collection of valuable SAMPLE history
• Ataxia • Movement and balance disorders • Developmental delay • Speech disorders • Intellectual disability	• May lead to inaccurate assessment of the patient's condition using the Glasgow Coma Scale
• Severe intellectual disability • Speech disorders • Lack of willingness or inability to interact with the paramedic performing the assessment	• May lead to inaccurate assessment of the patient's condition using the AVPU scale
• Microcephaly, Protruding tongue • Widely spaced teeth • Excessive salivation	• Increased difficulty in endotracheal intubation
• Large mouth • Pronounced protrusion of the jaw	• Reduced difficulty in endotracheal intubation
• Excessive salivation	• Difficulty in oral suctioning
• Seizures	• Application of appropriate rescue pharmacotherapy (Diazepam at a dose of 0.2–0.5 mg/kg; if ineffective, Midazolam at a dose of 0.1–0.5 mg/kg)

The AVPU scale is one of the basic ways to assess the level of consciousness in patients, which is routinely used by paramedics in the initial assessment of the patient. The letters in the acronym AVPU stand for Alert, Verbal, Pain, and Unresponsive [12, 18]. The high pain threshold associated with PWS, which typically manifests in childhood or adolescence and persists into adulthood, may significantly affect the "P" (Pain) component of the AVPU scale [6, 12, 18]. Additionally, due to profound intellectual disability, speech impairments, and either a lack of willingness or inability to interact with medical personnel, the "A" (Alert) and "V" (Voice) responses may be misinterpreted in both PWS and AS patients [1, 6, 9, 10, 12, 13].

In individuals with PWS, the impaired sensation of satiety leads to hyperphagia, often resulting in overweight and obesity. Obesity is a well-established risk factor for arterial hypertension [6, 12]. Therefore, routine blood pressure monitoring in patients with PWS is recommended to detect potential hypertensive disorders [1, 19, 20].

Endotracheal intubation is a procedure for advanced airway management of the patient, consisting of placing a special tube with a sealing balloon in the patient's trachea under visual control. The tube itself can be inserted through the nose or mouth. In PWS patients, anatomical features such as small, triangular-shaped mouths with downturned corners, thick saliva, and obesity may present significant challenges during endotracheal intubation [1, 6, 12, 21–23]. In individuals with AS, microcephaly and craniofacial

dysmorphisms, including a protruding tongue, wide interdental spaces, and excessive salivation, may further complicate the intubation process [7, 8, 10, 22, 24]. However, in some cases, features such as a large oral opening or a prominently protruding mandible may facilitate the procedure [12, 22, 24].

If the patient is unable to expectorate, spit out, or swallow secretions from the oral cavity on their own, and the airway is not yet secured with an endotracheal tube or other supraglottic device, there is a significant risk of the patient choking on secretions. To prevent this, secretions from the oral cavity should be suctioned out. Patients with PWS often produce abnormally thick saliva, which can obstruct the suction catheter, particularly if its diameter is too narrow [12, 18, 22, 23]. Individuals with AS also demonstrate a tendency toward excessive salivation [24].

An important symptom of AS in the context of the work of a paramedic are epileptic seizures present in this genetic syndrome. Seizures are a prominent feature of AS, typically emerging before the age of 3 years, peaking in frequency during early childhood, and gradually decreasing over time [10, 11]. To terminate a seizure episode in a rescue situation in a patient with AS, emergency medical personnel should initially administer benzodiazepines, the most used are diazepam, midazolam, clonazepam, or lorazepam. Diazepam at a dose of 0.3–0.5 mg/kg body weight rectal (approved for 2 years old and older) or intramuscular diazepam/midazolam at a dose of 0.15–0.2 mg/kg of body weight or intravenously (approved

for 6 years old and above) or intervention with midazolam at a dose of 0.1–0.5 mg/kg body weight intranasal (approved for 12 years old and above) or midazolam in buccal form. Emergency medical personnel should administer intravenous diazepam not exceeding a single dose of 10 mg, and for rectal form not exceeding 20 mg per single dose. Midazolam can be used at a dose of 10 mg for individuals weighing over 40 kg, and 5 mg for those weighing between 13 and 40 kg. The dose may be repeated once [11, 12, 25–30]. Paramedics in Poland are not authorised to administer the other medications listed in the guidelines for seizure epilepsy; therefore, the description of those drugs has been omitted.

Aim of the research

This paper assesses the level of knowledge of paramedics regarding general information about PWS and AS, as well as their knowledge of adapting emergency medical procedures to the specifics of these genetic syndromes. The choice of this topic stems from the lack of dedicated guidelines regarding emergency procedures for patients with genetic disorders, which can present a significant challenge in medical rescue practice.

Material and methods

As part of the preparation for this study, a survey was conducted involving 100 paramedics. The study was quantitative and took the form of an environmental questionnaire. The Bioethics Committee at the Medical University of Warsaw declared that the study complies with the principles of research ethics in accordance with the statement (no. AKBE/45/2023). The survey questionnaires were distributed electronically. Participation in the study was entirely voluntary, and the responses remained anonymous. The research method used was a diagnostic survey, and the research tool was an author-designed questionnaire. The questionnaire was developed based on the current state of knowledge regarding general symptoms and emergency medical procedures applied to patients with selected genetic disorders. The questionnaire consisted of a total of 21 questions. The first 5 questions of the questionnaire were the personal information section, including questions about the respondents' gender, age, work experience, place of employment, and level of education. The main part of the questionnaire consisted of 16 closed-ended single-choice (4) and multiple-choice (12, allowed for the collection of supplementary material) questions. These questions covered both general aspects of PWS and AS as well as questions on emergency procedures for patients with these genetic diseases. The method used made it possible to assess the respondents' level of knowledge on the issues studied.

Statistical analysis

Statistical analyses were conducted based on the data obtained from the survey. The analyses were performed using IBM SPSS Statistics 26 software (licensed to the Medical University of Warsaw). To assess the overall knowledge level of participants, descriptive statistics of the knowledge scores were calculated. Descriptive statistics were calculated, including the Shapiro-Wilk test for normality. Additionally, Spearman's rho correlation, the Mann-Whitney *U* test, and the Kruskal-Wallis test were applied where appropriate. The level of statistical significance was set at $\alpha = 0.05$.

Results

Characteristics of the study group

The study included 100 respondents, of whom 28% were women and 72% were men. The most represented age group consisted of individuals aged 23–25 years, accounting for 66% of the total sample. The least represented group was those aged 19–22 years, comprising only 4% of participants. The remaining age distribution included 23% of respondents aged 26–40 years and 7% aged 41–60 years. Many respondents (76%) had less than 5 years of professional experience as paramedics. Participants with 5–10 years of experience represented 16% of the sample, while those with 10–20 years of experience accounted for 5%. The smallest group, comprising 3% of the sample, consisted of individuals with over 20 years of experience in the profession. In terms of educational background, the dominant group were bachelor's degree holders, who constituted 82% of respondents. Individuals with secondary education accounted for 6%, while 12% of participants held a master's degree. At the time of the study, there were no master's programs specifically in emergency medical services for paramedics in Poland. Respondents with a master's degree (12 paramedics: 7 women and 5 men) had a university degree in public health (100%), which was closely related to the field of health sciences. The largest proportion of respondents ($N = 66$; 48.88%) worked in hospital emergency departments (ED) and admission rooms. Emergency Medical Services (EMS) teams employed 52 respondents (38.51%). The least representative group were medical transport personnel – 14 (10.37%) individuals. At the time of the survey, 3 (2.22%) participants were not professionally active as paramedics. A detailed distribution of sociodemographic characteristics is presented in Table 3.

Analysis of Paramedics' knowledge level regarding patients with selected genetic disorders

The second part of the questionnaire included items assessing respondents' knowledge of emergency medical procedures applied to individuals with

PWS and AS, as well as general familiarity with these genetically determined conditions. In the first question, participants were asked to subjectively assess their own level of knowledge regarding emergency interventions for patients with syndromes. The largest group (35%) rated their knowledge as "rather poor", followed by 30% who responded with "hard to say". A total of 27% assessed their knowledge as "poor". Only 8% considered their knowledge "rather good", and no respondent selected "good" as an answer (Figure 1). Among respondents with a master's degree, 50% answered "hard to say", 25% "poor", 17% "rather poor", and 8% "rather good", indicating greater uncertainty than in the overall group.

In summary, respondents had the greatest knowledge (correct answers) in the field of examination in PWS patients (69%), seizure epilepsy rescue medication administration (38%), and SAMPLE in PWS (32%). Respondents' knowledge related to the fewest correct answers concerned questions about symptoms of PWS (0%) and of AS (4%), difficulties that may occur during the intubation process of AS (0%) and of PWS (4%), and about the type of seizure to expect in AS (0%) (Table 4).

Figures 2 and 3 present the detailed answers of the respondents for the multiple- and single-choice questions about PWS and AS, respectively. Most responses (19% in total) correctly identified obesity as a characteristic symptom of Prader-Willi syndrome. Other frequently selected correct responses included excessive appetite (14%), high pain threshold (12%), muscle hypotonia (12%), short stature (11%), thick saliva (7%), and speech strabismus (33). Incorrect responses included speech impairment (8%), frequent smiling (7%), and epilepsy (6%). The key features of P-W syndrome to consider during the SAMPLE history, whose omission may affect clinical assessment correct responses, included delayed speech development (39%) and speech articulation disorders (27%). With regard the Glasgow Coma Scale for PWS, correctly indicated responses included speech delay (63%) and high pain threshold (37%). In the AVPU scale, the most accurately indicated response for PWS

Table 3. Sociodemographic data of the study group

The group studied <i>n</i> = 100	<i>n</i> (%)
Gender	
Women	28 (28)
Men	72 (72)
Age	
19–22	4 (4)
23–25	66 (66)
26–40	23 (23)
40–60	7 (7)
> 60	0
Education	
Secondary school	6 (6)
Bachelor`s degree	82 (82)
Master`s degree	12 (12)
Doctoral school	0
Work experience	
Less than 5 years	76 (76)
5 to 10 years	16 (16)
10 to 20 years	5 (5)
More than 20 years	3 (3)
Workplace*	
Emergency Medical Services	52 (39)
Emergency Department	66 (49)
Medical Transport	14 (10)
Doesn't in any profession	3 (2)

*Possibility to choose more than one place of employment.

was a high pain threshold (38%). In the question about recommended examinations for PWS patients, 69% of responses correctly indicated blood pressure measurement. Regarding difficulties with intubation for PWS, the most frequently and correctly indicated

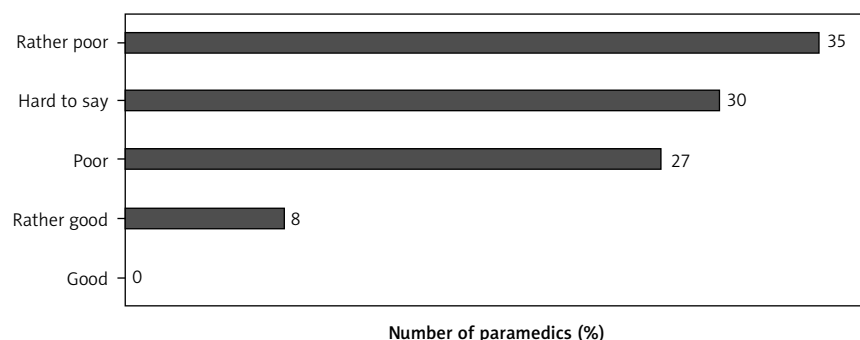


Figure 1. Self-assessment of respondents' knowledge about emergency medicine procedures used in people with Prader-Willi and Angelman syndromes

Table 4. Summary of respondents' answers to questions

Question	Fully correct answer (% of respondents)
Summary of respondents' answers to multiple-choice questions	
What symptoms are characteristic of Angelman syndrome? (10 options in total, including 6 correct and 4 incorrect answers)	1
What symptoms are characteristic of Prader-Willi syndrome? (10 options total, including 7 correct and 3 incorrect answers)	0
Which characteristics of Angelman syndrome should be considered during the SAMPLE history, and whose omission could distort the assessment of the patient's condition? (5 options in total, including 3 correct and 2 incorrect answers)	6
Which characteristics of Prader-Willi syndrome should be considered during the SAMPLE history, and whose omission could distort the assessment of the patient's condition? (4 options in total, including 2 correct and 2 incorrect answers)	32
What should you expect when assessing a person with Prader-Willi Syndrome using the Glasgow Coma Scale (GCS)? (2 options in total, including 2 correct and 0 incorrect answers)	25
What should you expect when assessing a person with Angelman Syndrome using the Glasgow Coma Scale (GCS)? (3 options in total, including 3 correct and 0 incorrect answers)	5
Which symptoms of Prader-Willi Syndrome may affect assessment using the AVPU scale (Alert, Verbal, Pain, Unresponsive)? (3 options in total, including 1 correct and 2 incorrect answers)	13
Which symptoms of Angelman Syndrome may affect assessment using the AVPU scale (Alert, Verbal, Pain, Unresponsive)? (4 options in total, including 2 correct and 2 incorrect answers)	28
What difficulties may occur during endotracheal intubation in a person with Prader-Willi Syndrome? (5 options in total, including 3 correct and 2 incorrect answers)	4
What difficulties may occur during endotracheal intubation in a person with Angelman Syndrome? (7 options in total, including 4 correct and 3 incorrect answers)	0
What should be expected during oral suctioning in a person with Prader-Willi Syndrome? (4 options in total, including 2 correct and 2 incorrect answers)	7
What type of seizure would you expect in a person with Angelman Syndrome? (8 options in total, including 5 correct and 3 incorrect answers)	0
Summary of respondents' answers to single-choice questions	
Which examination should always be performed in a patient with Prader-Willi syndrome?	69
What should be expected during oral suctioning in a patient with Angelman syndrome?	11
Which medication would you administer to stop a seizure in a person with Angelman syndrome?	38

responses were triangular-shaped mouth (29%) and obesity (23%). In the question about secretion suctioning in PWS, 32% of responses correctly indicated the risk of vomiting, and 30% cleft palate.

Regarding the symptoms characteristic of AS, 16% of responses correctly identified frequent smiling as a symptom. However, 14% of responses incorrectly indicated a high pain threshold, and 12% indicated severe developmental delay. Microcephaly, ataxia, and epilepsy were each selected in 11% of answers. Muscle hypotonia, incorrectly associated with AS, was marked in 10% of responses, excessive appetite in

4%, and overweight/obesity in 3%. For AS features relevant during a SAMPLE history, the most commonly and correctly selected responses were intellectual disability (24%), developmental delay (21%), and speech disorders (20%). For AS, the most frequently correct selected response for the Glasgow Scale was inability to speak (50%). Regarding difficulties with intubation for AS, the most frequently and correctly indicated responses were excessive salivation (17%), protruding tongue (16%), microcephaly (15%), and widely spaced teeth (14%). In the AVPU scale, the most accurately indicated responses for AS were speech disorders

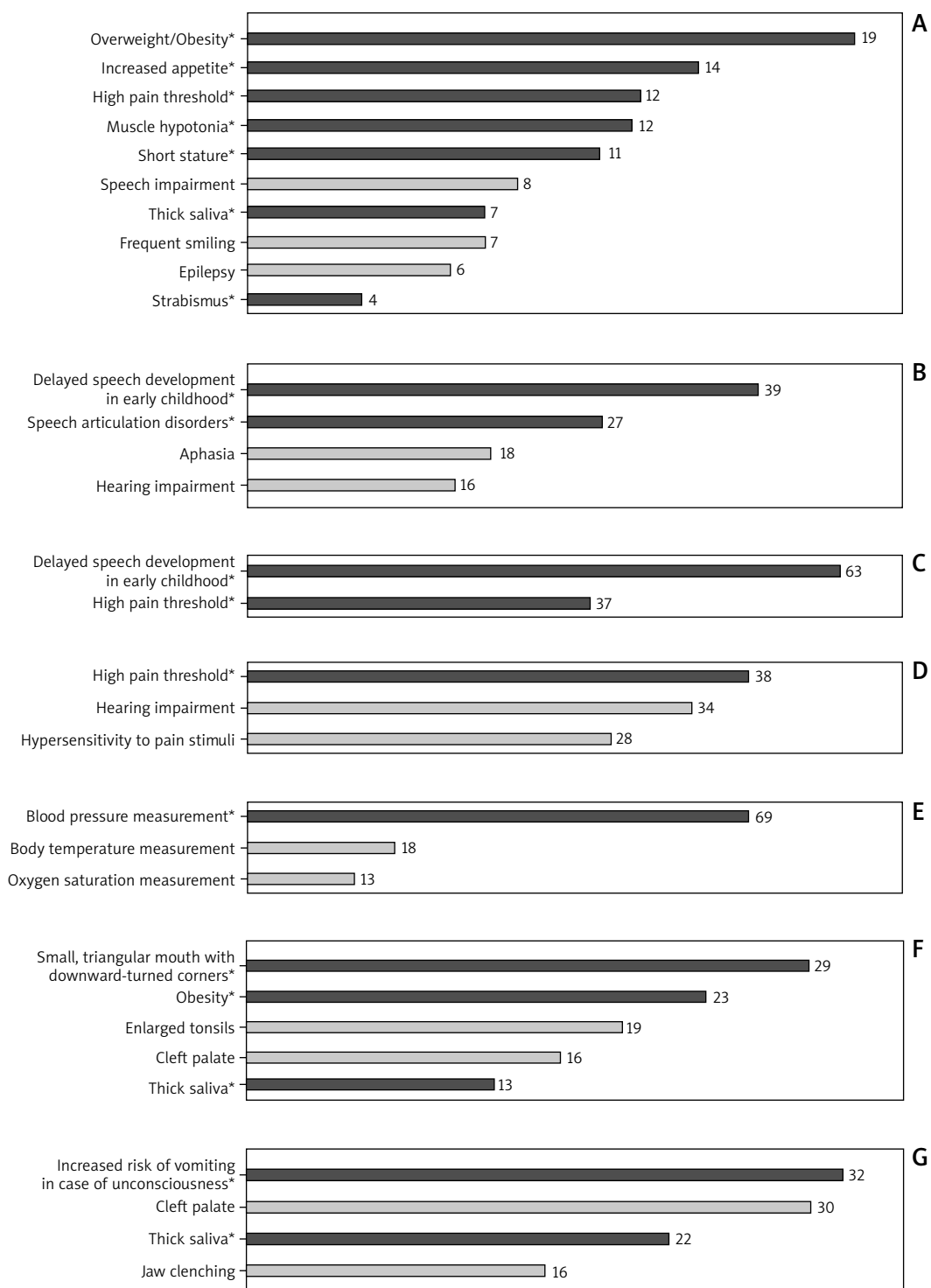


Figure 2. Prader-Willi syndrome number of paramedics selecting answers (%) in questions about: **A** – Symptoms; **B** – Key features to consider during the SAMPLE history, whose omission may affect clinical assessment; **C** – Anticipated findings during GCS assessment; **D** – Symptoms that may influence AVPU assessment; **E** – Recommended routine examination; **F** – Anticipated difficulties during endotracheal intubation; **G** – Expected complications during oral secretion suctioning. Dark grey bars with * indicate correct responses, while light grey bars represent incorrect answers.

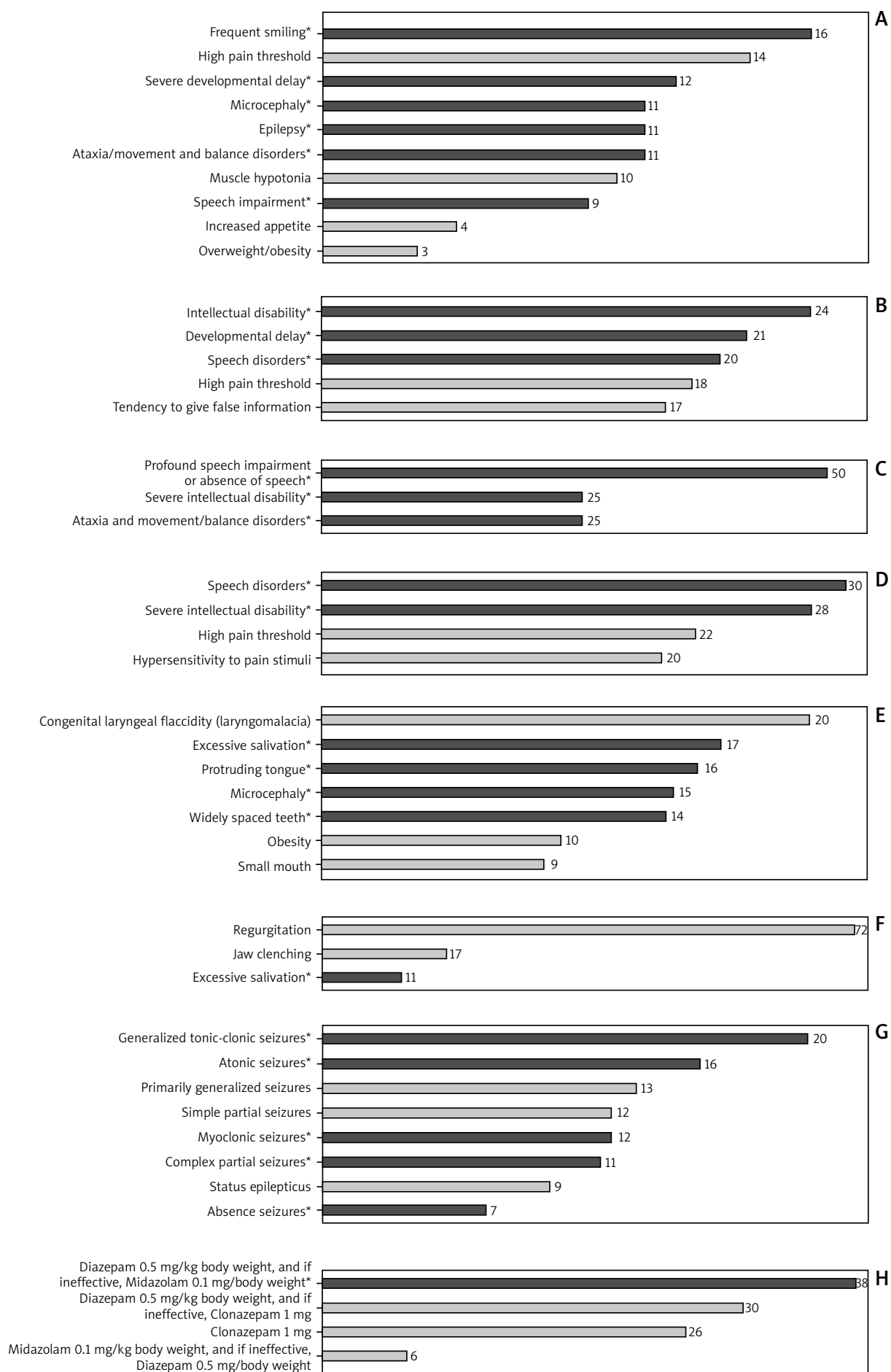


Figure 3. Angelman syndrome number of paramedics selecting answers (%) in questions about: **A** – Symptoms; **B** – Key features to consider during the SAMPLE history, whose omission may affect clinical assessment; **C** – Anticipated findings during GCS assessment; **D** – Symptoms that may influence AVPU assessment; **E** – Anticipated difficulties during endotracheal intubation; **F** – Expected complications during oral secretion suctioning; **G** – Seizure types commonly observed; **H** – Medications typically used to interrupt a seizure

Dark grey bars with * indicate correct responses, while light grey bars represent incorrect answers.

(30%) and intellectual disability (28%). In the question about secretion suctioning in AS, 72% of responses incorrectly indicated regurgitation as a symptom. On the topic of seizures in AS, the most frequently and correctly selected responses were tonic-clonic seizures (20%) and atonic seizures (16%); however, no participant answered this question fully correctly.

Descriptive statistics

To evaluate the knowledge level of paramedics concerning emergency management of patients with PWS and AS, 1 point was awarded for each correct answer, and 0 points for each incorrect one. In multiple-choice questions, selecting all options resulted in 0 points. Based on this scoring system, a knowledge index was calculated as the sum of all points obtained in the survey questionnaire. The low average score (20.60) indicated a generally low level of knowledge among paramedics regarding the topic around the 50th percentile (pct). The scale was as follows: 25th pct 18–20.5, 50th pct 20.5–23, and 75th pct ≥ 23 .

The distribution of scores was analysed using descriptive statistics, and the Shapiro-Wilk test was used to assess normality. The results are presented in Figure 4. The Shapiro-Wilk test showed a statistically significant deviation from a normal distribution ($p < 0.001$), and the presence of outliers further justified the use of non-parametric correlation tests.

Correlation between knowledge level and selected sociodemographic variables

The relationship between the overall knowledge score and selected sociodemographic variables – age,

years of professional experience, education level, self-assessment of knowledge, gender, and place of employment – was examined using Spearman's rank correlation (for ordinal variables) and the Mann-Whitney U test or Kruskal-Wallis test (for nominal variables). The analyses revealed no statistically significant relationships (Figure 5). This indicates that the level of knowledge did not significantly differ based on any of the analysed variables, including gender, place of employment, age, professional experience, and level of education, also in relation to paramedics with a master's degree.

Discussion

Both globally and in Poland, the number of newborns and adults with Prader-Willi syndrome and Angelman syndrome is steadily increasing. Research by Abate *et al.* indicates that the mortality rate in young patients is higher in those with PWS than in those with AS. However, patients with PWS are very often hospitalised, starting from a very young age [31]. It is estimated that in the group of patients aged 1–4 years, the hospitalisation rate is 93.2%, and in those aged 5–9 years, 79.6%. In the case of Angelman syndrome, this rate in children aged 5–9 years is 79.9% [31]. Furthermore, analysed groups of patients struggle with numerous clinical problems throughout their lives. For example, 94% of AS patients develop epilepsy, with episodes lasting several minutes in children and even over an hour in adults [32, 33]. In addition to conditions requiring rapid intervention, adult patients with AS develop chronic diseases such as scoliosis (50% of patients) and osteoporosis (20%), which impact

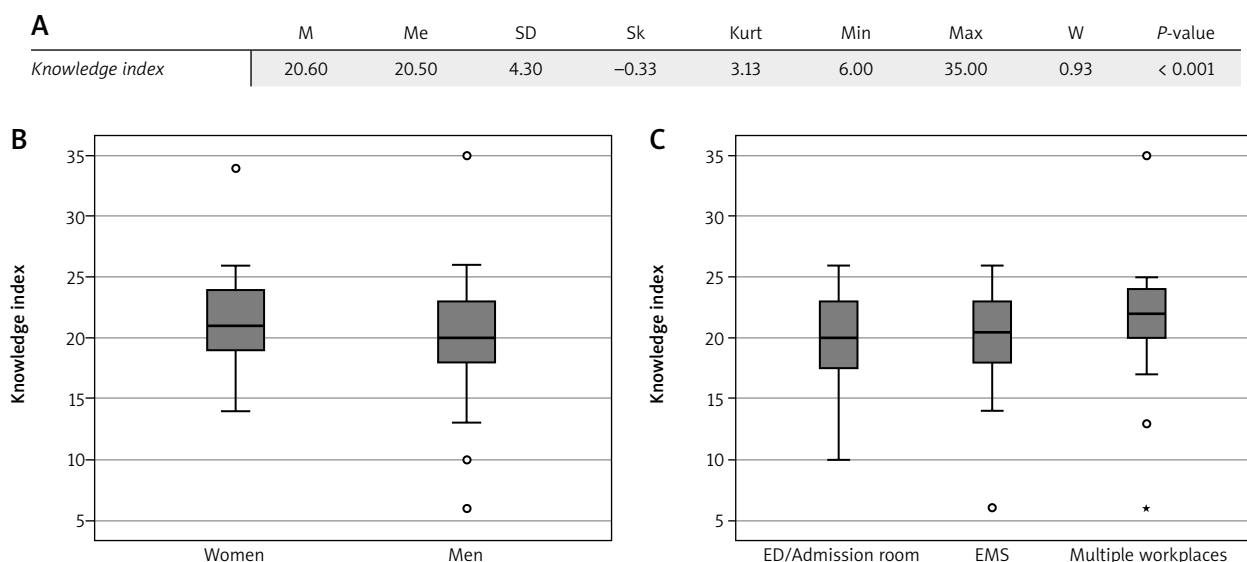


Figure 4. **A** – Knowledge index of Paramedics Shapiro-Wilk Test; **B** – box plot showing the distribution of knowledge scores by gender; **C** – box plot showing the distribution of knowledge scores by workplace

M – mean, Me – median, SD – standard deviation, Sk – Skewness, Kurt – Kurtosis, Min – minimum, Max – maximum, W – Shapiro-Wilk test statistic, *statistically significant, ED – Emergency Department, EMS – Emergency Medical Services.

A	Knowledge index	Women (n = 28)			Men (n = 72)			Z	P-value	r
		Mean rank	Mdn	IQR	Mean rank	Mdn	IQR			
		55.25	21.00	5.00	48.65	20.00	5.00	-1.03	0.305	0.10

B	Knowledge index				H(2)	P-value	η^2
		Mean rank	Mdn	IQR			
	Emergency department (n = 40)	42.96	20.00	5.75	5.29	0.071	0.04
	Emergency medical service (n = 22)	43.86	20.50	5.25			
	Two or more workplace (n = 33)	56.86	22.00	4.00			

C		Knowledge index	
Age	rho Spearman		0.04
	Significance		0.701
Professional experience	rho Spearman		0.11
	Significance		0.269
Level of education	rho Spearman		< 0.01
	Significance		0.978
Self-assessment of knowledge	rho Spearman		0.16
	Significance		0.107

Figure 5. Correlation statistics of paramedics' knowledge scores (**A**) result of the Mann-Whitney test comparing the knowledge index depending on gender; **B** – result of the Kruskal-Wallis test comparing the knowledge index depending on the place of work; **C** – Spearman correlation between the level of knowledge and age, professional experience, level of education, and self-assessment of knowledge

Mdn – median, IQR – interquartile range, Z – Z statistic, r – Correlation coefficient, H(2) – Kruskal-Wallis H statistic with 2 degrees of freedom, η^2 – eta squared, rho – Spearman's rho.

their functioning [32, 34], and depression is seen in 18% of PWS patients [35]. Many patients with PWS are treated with several daily medications, the necessity for each of which should be periodically re-evaluated [36]. Due to their low lean body mass and, often, increased sensitivity to drugs, a low starting dose is suggested, especially for psychotropic drugs and antihistamines. The use of antidepressants may also result in many respiratory complications in these patients [37, 38]. All of this contributes to low life satisfaction rates in these patients. The specificity, extensiveness, and chronic nature of the diseases requires medical interventions from childhood to adulthood, which also burdens the healthcare system [39, 40].

Rapid diagnosis is difficult for frontline physicians and other medical professionals, including paramedics, because these patients are few in number. However, it is essential for recognising the patient's clinical condition and implementing appropriate treatment, resulting in long-term survival [41]. Diagnostic difficulties and access to healthcare services are common, and the lack of knowledge about these conditions leads to unnecessary medical interventions, hospitalisations, and therapies [42, 43]. Currently, various social groups are paying greater attention to patients with genetic syndromes.

The greatest knowledge of a given genetic syndrome is given by specialists who report direct contact with these patients. This does not change the fact that medical personnel should possess fundamental/basic knowledge of specific factors that may hinder first-aid care in emergency situations. This study can therefore be considered preliminary and exploratory regarding this group of medics – paramedics, who most often have a direct impact on saving the lives of out-of-hospital patients. The presented topic requires further comparative analyses, which will identify knowledge gaps in individual medical professions, which could contribute to positive changes in medical education programming, also with regard to the increasingly reported genetically determined conditions. Medical professionals from various groups report a lack of evidence-based literature on care practices in these syndromes and emphasise the importance of further training for medical professionals from various medical fields in this area [25].

There is similar low access to knowledge in the field of PWS and AS. In Poland, it is difficult to estimate the total number of patients with both PWS and AS. There is also a lack of data on patients' health status and medical procedures in diagnostic and treatment management. It is worth noting that Suleja *et al.* col-

lected clinical descriptions of 75 Polish patients with AS, a significant contribution to the Polish cohort of patients with the condition. The results of Suleja *et al.* emphasise that primary care physicians have limited knowledge in the field in question, so it is difficult to find a specialist in diagnosis and treatment [44]. Other professionals who provide interdisciplinary care to patients with PWS and AS, including teachers and therapists, also point to knowledge gaps and the need for additional training and a holistic approach [45].

The findings of this project indicate that paramedics in Poland demonstrated insufficient knowledge to accurately identify the clinical manifestations associated with the analysed genetically determined disorders. This relationship is similar to that in other medical fields. However, in many situations, paramedics will be the first group of medical professionals to respond to emergency clinical conditions in patients with PWS and AS. This professional group was selected for the research project because it appears to be crucial in the interdisciplinary team's treatment of patients with genetic syndromes.

Despite the overall low level of knowledge observed in the study domain, it is worth noting that the vast majority of respondents were able to give some correct answers to the questions posed. Among the multiple-choice questions, respondents performed best on the item concerning the SAMPLE history and the components that should be considered when collecting this information from a person with Prader-Willi syndrome. There are no studies in the available literature that specifically address paramedics' knowledge of PWS and AS. To our knowledge, ours is the first study.

The limited knowledge of paramedics regarding emergency medical procedures for individuals with genetically determined disorders may be attributed to the insufficient coverage of this subject within the standard paramedic education curriculum. Furthermore, the relatively low prevalence of individuals with PWS and AS in Poland means that paramedics rarely encounter such patients in their routine practice. Consequently, this contributes to a low level of practical knowledge and limited familiarity in this area. On the other hand, the prevalence of individuals with genetically determined disorders is steadily increasing, which underscores the growing relevance and urgency of addressing this issue in both training and practice. This perceived knowledge gap may contribute to challenges in the effective execution of emergency medical interventions. Developing the knowledge and skills of paramedics (targeted education) is essential to improve emergency care for people with Prader-Willi and Angelman syndromes and other rare genetic diseases. In Poland, the registry of rare diseases is under development, and there is a list of rare diseases [46], as well as Polish Orphanet, an online database of rare diseases and orphan drugs,

including facilities providing care for such patients [47]. Poland also has the Polish Registry of Congenital Malformations [48], which since 1998 has collected data on all congenital malformations, which constitute a significant group (20–25%) of rare diseases.

The efficiency and quality of medical, paramedical, and even family prehospital care are primary indicators of life expectancy in patients with genetic diseases as well. This care should be based on recommendations relating to modern deep learning algorithms for disorder detection or molecular genotype-phenotype diagnostics [49–51]. The process of medical interventions can be influenced by all the disorders that co-occur in the genetic diseases in question.

Disorders associated with genetic diseases may affect the process of medical interventions in emergencies. Since the level of knowledge of paramedics about the characteristic symptoms of the patients with Prader-Willi and Angelman syndromes is low, they may use inappropriate emergency procedures that will affect patient survival. These results highlight that the topic of symptoms and medical procedures used in emergency situations in patients with rare genetic syndromes requires expansion during the education of paramedics, both at the undergraduate and graduate levels, and it could be included in continuing professional training programs and recommendations for paramedics.

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Ethical approval

Ethics Committee of the Medical University of Warsaw, Poland (AKBE/45/2023).

Conflict of interest

The authors declare no conflict of interest.

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