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Psychological and psychiatric service use among family caregivers of individuals with Angelman Syndrome: A cross-sectional study

Jan Domaradzki[1], Karolina Pospieszyńska-Martysiuk[2], Hanna Dianow[2], Joanna Węgrzyn[2], Dariusz Walkowiak[3]

[1] Department of Social Sciences and Humanities, Poznan University of Medical Sciences

[2] FAST POLAND – Foundation for Angelman Syndrome Therapeutics Poland, Al. Grunwaldzka 5, 80-236 Gdansk

[3] Department of Organization and Management in Health Care, Poznan University of Medical Sciences

Summary

Aim. This study aims to assess the perceived impact of caregiving for persons with Angelman Syndrome (AS) on the health of family caregivers and the extent and costs of using psychological and psychiatric services.

Material and methods. An anonymous, computer-assisted online survey among 84 family caregivers was conducted in collaboration with the Foundation for Angelman Syndrome Therapeutics Poland.

Results. 86.9% of caregivers experienced a negative impact of long-term caregiving on their physical and mental health. Many parents neglected their own health needs, including undergoing medical examinations (81%), taking prescribed medications (41.7%), engaging in physical activity (81%), and maintaining healthy dietary habits (53.5%). 63.1% of parents reported seeking specialist consultations or participating in health-related interventions, while 33.3% used professional psychological and psychiatric care, including pharmacotherapy (15.5%). A correlation was observed between the use of psychological and psychiatric services and both the age of the individual with AS and the occurrence of epileptic seizures in the past year. Most of the costs of the parents' psychological and psychiatric care incurred in 2024 were covered by private funds. While the total expenditure for such care amounted to 32,604.50 Euros, 96.2% of this sum was paid exclusively out of pocket.

Conclusions. To alleviate caregiver stress and burden, there is an urgent need to develop effective interventions and emotional support systems for caregivers. It is necessary to adopt a comprehensive approach to the mental health of families of individuals with AS, incorporating

professional psychological and psychiatric care. Additionally, adjustments in the reimbursement system for psychiatric services provided to caregivers are needed.

Key words: Angelman syndrome, family caregivers, health expenditure

Introduction

Angelman syndrome (AS) (ICD-10: Q93.51, ICD-11: LD90.0, ORPHA: 72, OMIM: 105830) is a rare, genetically determined neurodevelopmental disorder [1] that results from several genetic mechanisms: 65–75% of cases arise from a deletion in the maternal chromosome 15q11–q13 region, approximately 10–15% are caused by a point mutation in the maternal copy of the *UBE3A* gene, 3–4% result from an imprinting center defect (ICD), and 3–5% from paternal uniparental disomy (UPD) [2–4]. Additionally, about 1% of cases involve mosaicism. The genetic basis of AS influences the severity and range of symptoms, as well as the level of care required and the burden experienced by caregivers [5]. The prevalence of AS is estimated at 1 in 10,000-25,000 live births, with approximately 500,000 individuals affected worldwide [6]. As of now, 141 individuals from Poland are registered in the Global Angelman Syndrome Registry, the largest international database on this disorder [7].

Although AS presents with a wide spectrum of symptoms, the most common include severe to profound intellectual disability, motor impairment, balance disorders, minimal or absent speech development, epilepsy, and sleep disturbances [8–11]. Characteristic facial features include microcephaly, mandibular prognathism, macroglossia, a wide mouth with widely spaced teeth, and deep-set eyes [8, 12, 13]. Individuals with AS also exhibit a distinctive behavioral phenotype that includes a happy demeanor, inappropriate bouts of laughter accompanied by hand-flapping, limb tremors, difficulties with concentration, hyperactivity, eating and swallowing disorders, excessive drooling, and aggressive or self-injurious behaviors such as head-banging, biting, scratching, hair-pulling, and loud vocalizations. Additionally, they often display a fascination with water [3, 8, 12]. Common comorbidities include scoliosis, recurrent vomiting or nausea, and autism spectrum disorders [10, 14].

Due to its complex clinical presentation, AS poses a diagnostic challenge and may be mistaken for other genetic syndromes, leading to a prolonged diagnostic odyssey [9, 14–18]. Genetic testing is the only definitive diagnostic method for AS, including methylation testing, MS-MLPA, microarray analysis, fluorescence in situ hybridization (FISH), and *UBE3A* gene sequencing in cases of point mutations [13, 19]. Currently, no approved curative treatment exists for AS; however, preclinical studies as well as Phase I, II, and III clinical trials are ongoing to develop transformative therapies. At present, treatment is primarily symptomatic and includes pharmacotherapy (e.g., antiepileptic drugs), physical rehabilitation, occupational therapy, psychological therapy, manual therapy, sensory integration, speech therapy, and augmentative and alternative communication (AAC) training [20]. Despite the lack of a causal treatment, the life expectancy of individuals with AS is comparable to that of the general population [21], although it may be reduced by 10–15 years due to sudden unexpected death in epilepsy (SUDEP) or sudden unexpected death in sleep (SUDS) [22].

Individuals with AS require continuous, intensive, multidisciplinary care at home, frequent rehabilitation, and, in many cases, hospitalizations [23]. Long-term caregiving results in significant burdens for parents, negatively affecting their physical health (e.g., sleep deprivation, chronic fatigue, spinal problems, injuries from aggressive behaviors) and mental health (e.g., increased anxiety, depression, emotional instability, and a sense of helplessness), as well as their social well-being (e.g., family life disruption, social isolation, loneliness) [24–28]. These challenges are compounded by substantial financial costs, including direct expenses for medical care and diagnostics [9, 29, 30] and indirect costs associated with reduced or abandoned employment, most often by mothers [31]. Consequently, caregivers experience high levels of stress, a feeling of burden, and reduced quality of life [5, 9, 25, 27, 28, 30, 32]. Previous studies indicate that caregivers of individuals with AS report a greater caregiving burden than parents of children with other neurodevelopmental disorders and rare genetic syndromes [30, 33–35] and are particularly vulnerable to mental health crises, often requiring psychiatric care [9, 14–18].

Although previous studies have shown that AS care in Poland aligns with European and global standards, families face limited access to genetic diagnostics. A further challenge is the insufficient knowledge of rare diseases among primary care physicians. Additionally, there is a lack of coordinated care and a comprehensive support system for families [9]. However, the extent and costs of psychiatric care utilization among caregivers of individuals with AS remain unknown. Thus, as part of a broader project investigating the economic costs of caregiving for individuals with AS, this study aims to assess the perceived impact of caregiving on: (1) caregivers' health, (2) the extent of psychological and psychiatric care utilization, and (3) the associated costs for caregivers.

Materials and methods

Study design

This study was designed as patient-oriented research [36, 37], meaning that its development and implementation were carried out in close collaboration with the patient organization Foundation for Angelman Syndrome Therapeutics Poland – FAST Poland (https://www.cureangelman.pl). Given the study's objectives, involving caregivers as research partners was essential, allowing the research to focus on priorities defined by parents. As an equal partner in the research team, FAST Poland was actively engaged in developing the research tool and data collection. Together with the foundation, an anonymous, computer-assisted online self-report survey was designed to assess the scope and costs of psychological and psychiatric care utilization among caregivers of individuals with AS.

The study was approved by the Bioethics Committee of Poznan University of Medical Sciences (KB – 660/24, issued on October 9, 2024). Additionally, it was endorsed by the board of FAST Poland. In accordance with the Helsinki Declaration, all participants provided written informed consent before completing the survey [38].

Study participants

Due to the rarity of AS and the absence of a national patient registry in Poland, participant recruitment was conducted with the assistance of FAST Poland, which invited caregivers through internal communication channels, including its website, Facebook page, and internal mailing list.

The inclusion criteria for participation were: (1) age of at least 18 years, (2) being the primary family caregiver of a person with AS, (3) a molecularly confirmed AS diagnosis, (4) proficiency in Polish, (5) access to the Internet Patient Account (*Internetowe Konto Pacjenta* – IKP) system, (6) ability to use electronic devices for online participation, and (7) providing written informed consent to participate.

Research tool

Based on a literature review on the costs associated with caring for individuals with AS, an original questionnaire was developed to assess caregivers' perceived health impacts and psychiatric care utilization. The questionnaire underwent pilot testing with five parents. The survey retrospectively asked caregivers whether providing care for an individual with AS had affected: (1) their health, (2) their medical and health-related behaviors, (3) their utilization of healthcare and mental well-being support, and (4) their expenses for psychological and psychiatric care. Additionally, the questionnaire included demographic questions about the caregiver and the individual with AS.

Participants completed the questionnaire retrospectively, evaluating the impact of caregiving over the past 12 months (from January 1 to December 31, 2024) using a five-point Likert scale or by entering data from the Internet Patient Account (IKP) system.

Procedure

The study was conducted over a two-month period, from January to February 2025, via the FAST Poland website, social media platforms, and internal mailing lists. In the first stage, participants received an invitation letter describing the research team, study objectives, methodology, and plans for data presentation. They were also informed that participation was anonymous, voluntary, and confidential. FAST Poland then provided all eligible participants with instructions for completing the questionnaire and using the IKP system. Participants were informed about the study scope in advance, allowing them to prepare responses regarding care-related expenses. For privately funded procedures, respondents had time to locate relevant documents, notes, or other records. After signing an electronic informed consent form, caregivers gained access to the online questionnaire. No financial compensation was provided for participation.

Statistical Analysis

Descriptive statistics were used for data analysis. Questions regarding the use of various forms of specialist care were analyzed using logistic regression in a dichoto-

mous format ("used" vs. "not used"). Logistic regression was applied to sociodemographic variables, with the age of the individual with AS included as a covariate. Examined factors included educational level, caregiver age, gender, employment status, and the occurrence of epileptic seizures in the past year. Statistical analyses were conducted using JASP software (version 0.18.3).

Results

A total of 84 primary caregivers participated in the study, including 79 mothers (94%) and 5 fathers (6%) (Table 1). The age of the surveyed parents ranged from 29 to 55 years, with a mean of 39.4 years. While 45.2% of caregivers were professionally active, more than half reported being unemployed due to caregiving responsibilities (54.8%).

Table 1. **Socio-demographic characteristics of AS caregivers**

Characteristics	*N* (%)
Sex	
female	79 (94)
male	5 (6)
Age (in years)	
range	29-55
mean (*SD*)	39.4 (6.3)
median	39
Professional activity	
yes	38 (45.2)
no	46 (54.8)

Among individuals with AS, the proportion of males and females was equal, with ages ranging from 1 to 26 years (mean: 8.8 years) (Table 2). While the majority had a genotype resulting from a deletion in the 15q11–q13 region (81%), 8.3% had a mutation in the *UBE3A* gene, 4.8% had paternal uniparental disomy, and 2.4% had an imprinting center defect. More than half of individuals with AS experienced epileptic seizures in the past year (51.2%).

Table 2. **AS patients' characteristics**

Characteristics	*N* (%)
Sex	
female	42 (50)
male	42 (50)

table continued on the next page

Age (in years)	
range	1-26
mean (*SD*)	8.8 (5.4)
median	8
Genotype determined at diagnosis	
deletion of the 15q11–q13 region	68 (81)
UBE3A gene mutation	7 (8.3)
uniparental disomy (UPD)	4 (4.8)
imprinting center defect	2 (2.4)
other	3 (3.6)
Did the patient have seizures in the past year?	
yes	43 (51.2)
no	41 (48.8)

A total of 86.9% of caregivers reported that providing care for a person with AS had a negative impact on their health (Table 3). Additionally, 81% of parents admitted that they lacked the time or financial resources for regular medical check-ups, and 41.7% stated that they did not purchase medications for themselves. Caring for a child with AS led 81% of parents to give up physical activity. Moreover, 42.9% of respondents indicated that they did not take vitamin supplements, and 53.5% reported a decline in the quality of their diet.

Table 3. **Impact of caregiving for a person with AS on caregiver's health**

	Definitely not *n* (%)	Rather not *n* (%)	I do not know *n* (%)	Rather yes *n* (%)	Definitely yes *n* (%)
Has caring for a person with AS had a negative impact on your health?					
	1 (1.2)	3 (3.6)	7 (8.3)	31 (36.9)	42 (50)
I don't have the time and/or resources to attend regular medical check-ups					
	1 (1.2)	12 (14.3)	3 (3.6)	23 (27.4)	45 (53.6)
I do not purchase medications for myself					
	18 (21.4)	24 (28.6)	7 (8.3)	14 (16.7)	21 (25)
I have given up physical activity					
	7 (8.3)	6 (7.1)	3 (3.6)	32 (38.1)	36 (42.9)
I do not take vitamin supplements					
	14 (16.7)	31 (36.9)	3 (3.6)	22 (26.2)	14 (16.7)
The quality of my diet has declined					
	12 (14.3)	24 (28.6)	3 (3.6)	17 (20.2)	28 (33.3)

63.1% of caregivers reported that due to their caregiving responsibilities, they attended individual specialist visits or participated in physical activities aimed at improving their health, with 29.8% utilizing services funded by mandatory health insurance contributions (Table 4). Additionally, 33.3% of parents reported participating in individual or group therapy sessions with a psychologist, psychiatrist or other mental health specialist, while 15.5% were taking psychiatric medications.

Table 4. **Healthcare utilization and well-being support among caregivers**

Characteristics	*N* (%)
Have you attended individual visits with specialists or participated in physical exercise classes (e.g., yoga or "healthy spine" classes) aimed at improving your health in connection with caregiving?	
yes	53 (63.1)
no	31 (36.9)
Including specialist visits funded by mandatory health insurance contributions	
yes	25 (29.8)
no	59 (70.2)
Have you attended individual visits or group therapy sessions with a psychiatrist, psychologist, or another mental health specialist to improve your well-being in connection with caregiving?	
yes	28 (33.3)
no	56 (66.7)
Have you used psychiatric pharmacological therapy in connection with caregiving?	
yes	13 (15.5)
no	71 (84.5)

The results of the logistic regression analysis across different models indicated that caregivers' use of medical services, including psychological and psychiatric support, was significantly associated with the age of the individual with AS and the occurrence of epileptic seizures in the past year (Table 5). A positive association was observed between the age of the individual with AS and the odds of caregivers accessing specialist consultations covered by mandatory health insurance (OR = 1.101). This suggests that the older the child, the greater the odds that the family will use this type of care. In addition, the occurrence of seizures significantly increased the odds that caregivers would engage in individual or group therapy sessions (OR = 2.727) and use psychiatric pharmacotherapy (OR = 6.187).

Table 5. **Stepwise logistic regression analysis of factors associated with healthcare utilization and well-being support among caregivers**

Regression parameters	*Have you attended individual visits with specialists or participated in physical exercise classes (e.g., yoga or "healthy spine" classes) aimed at improving your health in connection with caregiving?*	*Including specialist visits funded by mandatory health insurance contributions*	*Have you attended individual visits or group therapy sessions with a psychiatrist, psychologist, or another mental health specialist to improve your well-being in connection with caregiving?*	*Have you used psychiatric pharmacological therapy in connection with caregiving?*
	OR (95% CI)	OR (95% CI)	OR (95% CI)	OR (95% CI)
Intercept		0.179*** (0.062-0.468)	0.290** (0.138-0.610)	0.191*** (0.013-0.231)
Patients' age		1.101* (1.006-1.206)		
Caregivers' age				
Education				
Working vs. nonworking				
Mother vs. father				
Patients' seizure in the past year vs. no seizure			2.727* (1.049-7.090)	6.187* (1.274-30.042)
Genotype				
R^2 Nagelkerke	0	0.078	0.072	0.137
p-value for model	0	0.03	0.04	0.01

Notes: * p-value < 0.05; ** p-value < 0.01; *** p-value < 0.001

Among the 28 caregivers who used psychological and psychiatric care, 100% paid for individual or group therapy sessions with a mental health specialist using private funds, while 53.6% also received support from the National Health Fund (NFZ) (Table 6). The total cost of these therapies amounted to 23 869.72 Euros, of which caregivers covered 97.8% from private funds, with only 2.2% funded by the NFZ. Additionally, among the 13 parents who reported using psychiatric pharmacological treatment, all covered the costs privately, while 69.2% also benefited from NFZ re-

imbursement. Although the total cost of psychiatric pharmacological treatment was 8 734.78 Euros, caregivers covered 91.8% of this amount from private funds. Overall, the total cost of psychological and psychiatric care amounted to 32 604.50 Euros, with only 3.8% of all expenses covered by public funds. Of this, 532.35 Euros (1.6%) was allocated to therapy sessions, and 715.37 Euros (2.2%) to pharmacological treatment.

Table 6. **Costs and utilization of psychiatric and psychological care among caregivers**

	Number of caregivers *n* = 28	Total cost EURO* (%)
Due to caregiving, participation in individual visits or group therapy sessions with a psychiatrist, psychologist, or other mental health specialist to improve personal health:		
private funds	28 (100.0)	23 337.37(97.8)
public funds (NFZ)	15 (53.6)	532.35 (2.2)
total cost of individual or group therapy sessions		23 869.72(100)
	n=13	EURO (%)
Due to caregiving, psychiatric pharmacological treatment was used		
private funds	13 (100)	8 019.41 (91.8)
public funds (NFZ)	9 (69.2)	715.37 (8.2)
total cost of psychiatric pharmacological treatment		8 734.78 (100)
Total cost of psychiatric and psychological care		32 604.50 (100)
total care costs from private funds		31 356.79 (96.2)
total care costs from public funds		1 247.71 (3.8)

* The conversion of Polish zloty to Euros was based on the exchange rate from Table No. 252/A/NBP/2024 of the Polish National Bank, dated December 31, 2024. (1 EURO = 4.2730 PLN)

Discussion

The findings highlight the complex impact of providing long-term care to individuals with AS on parents' health and psychological well-being, emphasizing the urgent need for systemic support tailored to their specific needs. Many parents experience significant emotional and psychological strain, underscoring the necessity of creating a comprehensive support system and effective interventions that address their actual needs [25, 27]. This is particularly important given that the current *Rare Disease Plan* (2024–2025) focuses on clinical aspects of patient care while overlooking the psychosocial situation of caregivers. Meanwhile, recent studies show that parents—especially mothers—of individuals with AS experience reduced quality of life, high levels of stress and parental burden, and considerable financial strain [28]. Furthermore, existing

research suggests that caregiving demands associated with AS result in greater caregiver burden compared to parents of children with other neurodevelopmental disorders [30, 33–35], and these parents are also at higher risk for mental health crises and psychiatric service use [9, 14–18]. Therefore, it is crucial to implement measures that support the mental well-being of entire families affected by AS, including access to psychological and psychiatric care, emotional support, and reimbursement for these services.

The study has led to several significant findings, which highlight that: (1) the majority of caregivers of individuals with AS experience a negative impact on their own health due to long-term caregiving; (2) the challenges associated with caregiving cause parents to neglect their own health needs; and (3) although one-third of parents used professional psychological and psychiatric care, including pharmacological therapy, the majority of the associated costs are covered by private funds.

Our study confirms findings from other researchers who have demonstrated that the demands of caring for an individual with AS result in severe, prolonged stress and a feeling of burden, negatively affecting various aspects of caregivers' functioning, including their physical, mental, and emotional health [25, 27, 28]. Although caring for an individual with AS can be a source of personal satisfaction [24, 39], parents experience many negative emotions, including anxiety, uncertainty, fear, depression, loss of control, helplessness, sadness, anger, and guilt [25–27]. It has also been shown that, due to typical AS-related problems such as sleep disturbances and discontinuity and nocturnal awakenings, parents experience chronic fatigue, report high levels of exhaustion, and suffer from reduced quality of life [5, 40, 41].

These findings are particularly significant, as Pelentsov et al. [42] have shown that, regardless of the specific rare disease affecting the patient, caregivers experience physical exhaustion, sleep disturbances, fatigue, loss of appetite and weight, headaches, and frequent colds. Existing studies also suggest that parents of children with AS face more severe stressors than parents of children with other neurodevelopmental disorders or healthy children and are more likely to suffer from depression and anxiety. For example, Adams et al. [34, 35] demonstrated that while anxiety, depression, and a feeling of burden are common among parents of children with rare genetic syndromes, these issues are particularly pronounced among caregivers of children with AS due to the severity and chronic nature of challenging behaviors in these children. Similarly, Griffith et al. [24] observed higher levels of clinical anxiety and depression in parents of children with AS than in caregivers of children with other rare genetic syndromes, autism spectrum disorder, and healthy children. A recently published comparative study of Polish caregivers also showed that, due to seizures, sleep problems, and mobility deficits, parents of children with AS reported significantly lower quality of life and higher perceived burden compared to parents of children with 22q11 deletion syndrome and Williams syndrome [33].

The findings also confirm that due to the time and energy required for caregiving tasks such as feeding, bathing, dressing, hygiene, and ensuring appropriate medical care, many caregivers rarely prioritize their own health. Lacking time for rest and self-care, parents prioritize their child's needs over their own, often neglecting regular medical check-ups, limiting their own medication intake, reducing physical activity, and

disregarding their dietary habits [43–45]. This aligns with observations that caregivers, even when ill themselves, rarely seek medical help, leading to deteriorating health and well-being [46]. As a result, parents of individuals with rare diseases report inadequate sleep, forgetting to take medications, and neglecting medical visits; they often postpone or forget their own health needs to remain at home and care for their loved ones [47, 48]. Previous studies have shown that parents and other family caregivers frequently neglect their sleep, emotional well-being, personal happiness, and pursuit of personal goals and interests. Consequently, more than half of parents report worsening physical, mental, and social health, with the most common complaints including chronic stress, sleep problems, fatigue, lack of energy, and depression [49–51].

Finally, this study reveals that although many caregivers seek professional psychological and psychiatric care, including pharmacological therapy, the vast majority of associated costs are covered by private funds. This finding aligns with previous findings showing that while many family caregivers of individuals with rare diseases experience depression, anxiety disorders, and other negative emotional states, they lack adequate emotional and social support systems [52]. Numerous existing studies indicate that comprehensive psychological support for families caring for children with AS is lacking, and many families report difficulty accessing emotional support, psychological counseling, medication therapy, and respite care [9, 53]. Although psychological support is recommended for the entire family at the time of diagnosis, our study shows that nearly all parents must seek such support independently and bear the high costs of psychological and psychiatric therapy. Similarly, Bourrat et al. [54] found that among the 31% of caregivers who opted for psychological therapy, 80% had to cover the costs themselves, and nearly half of the parents who did not use such care declared that its cost was the main barrier. This aligns with findings from a previous study of Polish caregivers of children with rare diseases, which indicated that despite experiencing health problems, many parents feel overlooked by the healthcare system and believe that their own needs, including psychological and emotional support, remain unmet [53]. Consequently, caregivers of sick family members are significantly more vulnerable to depressive and anxiety disorders, declining physical health, lack of additional health insurance, and reduced access to healthcare services compared to non-caregivers [55].

Study limitations

Although this is the first attempt in Poland to estimate the self-reported impact of caring for a person with AS on the health of family caregivers, as well as the extent of psychological and psychiatric care utilization and its associated costs, our study has several limitations. First, the study involved a relatively small cohort of respondents. However, given the rarity of the disease and the absence of a rare disease registry in Poland, the exact number of AS patients and families in the country is unknown. Moreover, since the data were collected from households, the 84 responses should be considered a high number. Second, the study was retrospective, and some data were based on caregivers' subjective assessments without the possibility of precise

verification. However, it should be emphasized that data regarding the patient, the use of psychological and psychiatric care, and its costs reimbursed by the NFZ were obtained by respondents from the Internet Patient Account (IKP). Third, the reported use of psychological and psychiatric care and its privately funded costs may be subject to errors due to the lack of detailed records among some caregivers or memory bias. Fourth, the study was conducted via an online survey, which may have influenced the recruitment process. However, given that AS is a rare disease and Poland lacks a patient registry, participant recruitment was carried out through the patient organization FAST Poland, whose primary communication tools include its website, Facebook profile, and internal mailing list. A further limitation may be that, like any system, the IKP may contain incorrect information. Another limitation of the study is the significant gender imbalance among respondents – the vast majority were women, with only five male caregivers participating. This may have introduced a gender bias and limits the generalizability of findings across different caregiving experiences. Future research should aim to include a more balanced representation of male and female caregivers to better understand potential gender-specific challenges and coping strategies.

Conclusions

The findings highlight several important implications for improving healthcare support for parents of children with AS. Since many parents report that the burdens of caregiving lead to emotional and mental health problems, there is an urgent need to develop effective interventions and support systems that address their needs. Particular attention should be paid to the experience of various negative emotional states, which may threaten the psychological well-being of family caregivers who juggle multiple roles without institutional or social support. Since the *Rare Disease Plan for 2024-2025*, adopted last year, focuses on clinical aspects and the medical needs of patients but overlooks the psychosocial needs of caregivers, it is necessary to develop psychological interventions that can help reduce caregiving-related stress, burden, and feelings of isolation experienced by many caregivers. A holistic approach to the mental health of families affected by AS should be emphasized, incorporating emotional support, professional psychological and psychiatric care for the entire family, and reimbursement for psychological and psychiatric services.

List of abbreviations: *IKP: Internet Patient Account; NFZ: National Health Fund, AS: Angelman syndrome*

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Address: Jan Domaradzki
Department of Social Sciences and Humanities
Poznan University of Medical Sciences
60-806 Poznań, Rokietnicka 7 Street
e-mail: jandomar@ump.edu.pl