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ASSESSMENT OF PUBLIC AWARENESS IN ABAI REGION REGARDING SPINAL MUSCULAR ATROPHY IN CHILDREN AND THE IMPORTANCE OF GENETIC COUNSELING IN FAMILY PLANNING

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Abstract

Introduction. Public awareness of spinal muscular atrophy (SMA) in children and the importance of genetic counseling during family planning represent key factors in the prevention and early diagnosis of this disease.

Objective. To assess the level of public awareness regarding spinal muscular atrophy in children and the significance of genetic counseling in family planning.

Materials and Methods. A cross-sectional study was conducted among 315 respondents**.** Statistical analysis was performed using SPSS software (version 26.0)**.** Histograms and the Kolmogorov–Smirnov test were applied to assess the normality of distribution for all variables. Given the non-normal distribution, non-parametric statistical methods were used. Quantitative continuous variables are presented as medians and percentiles**,** while dichotomous variables are presented as proportions (absolute numbers)**.** Results with $p \leq 0.05$ were considered statistically significant.

Results. According to the findings, only 47.6% of respondents had ever heard of spinal muscular atrophy (SMA). Among respondents, 64.1% indicated that in the future they plan to have children with their spouse or partner. A total of 58% expressed willingness to undergo genetic counseling, including genetic testing for SMA-related gene mutations.

However, only 35.6% of respondents were aware of the main clinical symptoms of SMA in children (such as muscle weakness, lethargy, weak sucking reflex, motor developmental delay, and breathing difficulties), which would enable timely suspicion of the disease and earlier referral to medical specialists.

Conclusion. Thus, the level of public awareness regarding spinal muscular atrophy remains low and requires educational efforts and broader dissemination of information about this disease.

Keywords: *spinal muscular atrophy, orphan diseases, genetic disorders, mutation.*

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Резюме

ОЦЕНКА УРОВНЯ ОСВЕДОМЛЕННОСТИ НАСЕЛЕНИЯ АБАЙСКОЙ ОБЛАСТИ О СПИНАЛЬНОЙ МЫШЕЧНОЙ АТРОФИИ У ДЕТЕЙ И ВАЖНОСТИ ГЕНЕТИЧЕСКОГО КОНСУЛЬТИРОВАНИЯ ПРИ ПЛАНИРОВАНИИ БЕРЕМЕННОСТИ

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Введение. Осведомленность населения о спинальной мышечной атрофии (СМА) у детей и важность генетического консультирования при планировании семьи являются ключевыми факторами профилактики и ранней диагностики этого заболевания.

Цель. Оценить уровень осведомленности населения о спинальной мышечной атрофии у детей и значение генетического консультирования при планировании семьи.

Материалы и методы. Было проведено поперечное исследование среди 315 респондентов. Статистический анализ проводился с использованием программы SPSS (версия 26.0). Для оценки нормальности распределения всех переменных использовались гистограммы и тест Колмогорова–Смирнова. Учитывая ненормальное распределение, были использованы непараметрические статистические методы. Количественные непрерывные переменные представлены в виде медиан и процентилей, а дихотомические переменные – в виде долей (абсолютных чисел). Результаты при $p \leq 0,05$ считались статистически значимыми.

Результаты. Согласно полученным данным, только 47,6% респондентов когда-либо слышали о спинальной мышечной атрофии (СМА). Среди респондентов 64,1% указали, что в будущем планируют иметь детей с супругом/партнером. 58% выразили готовность пройти генетическое консультирование, включая генетическое тестирование на наличие генных мутаций, связанных со СМА.

Однако только 35,6% респондентов были осведомлены об основных клинических симптомах СМА у детей (таких как мышечная слабость, вялость, ослабленный сосательный рефлекс, задержка моторного развития и затрудненное дыхание), что позволило бы своевременно заподозрить заболевание и направить пациента к врачу-специалисту.

Заключение. Таким образом, уровень осведомленности населения о спинальной мышечной атрофии остается низким и требует просветительской работы и более широкого распространения информации об этом заболевании.

Ключевые слова: *спинальная мышечная атрофия, орфанные заболевания, генетические нарушения, мутация.*

Для цитирования: *Курмашева А.Т., Хисметова З.А., Мукушева А.М., Сиязбекова З.С., Ахметова К.М.* Оценка уровня осведомленности населения Абайской области о спинальной мышечной атрофии у детей и важности генетического консультирования при планировании беременности .// Наука и Здравоохранение. 2025. Vol.27 (5), С.120-126. *doi 10.34689/SH.2025.27.5.015*

Түйіндеме

АБАЙ ОБЛЫСЫ ХАЛҚЫНЫҢ БАЛАЛАРДАҒЫ ЖҰЛЫН БҰЛШЫҚЕТ АТРОФИЯСЫ ТУРАЛЫ ЖӘНЕ ЖҮКТІЛІКТІ ЖОСПАРЛАУ КЕЗІНДЕГІ ГЕНЕТИКАЛЫҚ КЕҢЕС БЕРУДІҢ МАҢЫЗДЫЛЫҒЫ ЖӨНІНДЕГІ АҚПАРАТТАНУ ДЕҢГЕЙІН БАҒАЛАУ

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Кіріспе. Балалардағы жұлын бұлшықет атрофиясы туралы және жүктілікті жоспарлау кезіндегі генетикалық кеңес берудің маңыздылығы жөніндегі халықтың хабардар болуы аталған аурудың алдын алу мен ерте диагностикалау үшін негізгі факторлардың бірі болып табылады.

Зерттеудің мақсаты: Балалардағы жұлын бұлшықет атрофиясы туралы және жүктілікті жоспарлау кезінде генетикалық кеңес берудің маңыздылығы жөнінде халықтың хабардарлық деңгейіне баға беру.

Материалдар мен әдістер: 315 респонденттің қатысуымен бірмезеттік көлденең зерттеу жүргізілді. Статистикалық талдау SPSS бағдарламалық қамтамасыздандыруы (26.0 нұсқасы) арқылы орындалды. Барлық айнымалылардың таралуының қалыптылығын тексеру үшін гистограммалар және Колмогоров-Смирнов критерийі қолданылды. Таралудың қалыпты түрден ауытқығанын ескере отырып, параметрлік емес статистикалық әдістер пайдаланылды. Біздің нәтижелерде сандық үздіксіз айнымалылар медиана және процентильдер түрінде, ал дихотомиялық айнымалылар үлестер (абсолюттік саны) түрінде көрсетілді. p=0,05 мәні статистикалық тұрғыдан мәнді деп есептелді.

Нәтижелер: Алынған нәтижелерге сәйкес, респонденттердің тек 47,6%-ы жұлын бұлшықет атрофиясы туралы естіген. Респонденттердің 64,1%-ы болашақта жұбайымен немесе серіктесімен жүктілікті жоспарлауды көздейтінін атап өтті. 58% респондент генетикалық кеңес алуға және жұлын бұлшықет атрофиясын тудыратын мутациялық гендерді анықтауға арналған генетикалық тест тапсыруға дайын екенін білдірді. Респонденттердің тек 35,6%-ы балалардағы жұлын бұлшықет атрофиясының негізгі белгілерін білетінін көрсетті, бұл ауруды уақытылы күдіктенуге және дер кезінде дәрігерге жүгінуге көмектесер еді.

Қорытындылар: Осылайша, халықтың жұлын бұлшықет атрофиясы туралы хабардарлық деңгейі төмен болып табылады және бұл ауру туралы ақпараттандыру мен түсіндіру жұмыстарын күшейтуді талап етеді.

Түйінді сөздер: *жұлын бұлшықет атрофиясы, сирек кездесетін аурулар, генетикалық аурулар, мутация.*

Дәйексөз үшін: *Курмашева А.Т., Хисметова З.А., Мукушева А.М., Сиязбекова З.С., Ахметова К.М.* Абай облысы халқының балалардағы жұлын бұлшықет атрофиясы туралы және жүктілікті жоспарлау кезіндегі генетикалық кеңес берудің маңыздылығы жөніндегі ақпараттану деңгейін бағалау // Ғылым және Денсаулық сақтау. 2025. Vol.27 (5), Б. 120-126. *doi 10.34689/SH.2025.27.5.015*

Introduction

Spinal muscular atrophy (SMA) is a severe hereditary disease characterized by degeneration of motor neurons and progressive muscle weakness. With the growing availability of genetic testing, the level of public awareness of hereditary diseases and the possibilities for their early detection are becoming increasingly important. The present study is aimed at examining the awareness of SMA among different social groups as well as identifying the factors that influence the level of knowledge.

Spinal muscular atrophy (SMA) represents a group of genetic neuromuscular disorders that lead to progressive weakness, muscle atrophy, and paralysis due to the degeneration of motor neurons in the spinal cord [1]. In most cases, the cause of degeneration and death of alpha motor neurons in the spinal cord is a mutation in a specific region of chromosome 5 DNA, known as the SMN1 gene (survival motor neuron 1 gene), located at 5q11.2–q13.3 [1,2]. Under normal conditions, this gene regulates the synthesis of the SMN protein, which plays a critical role in the metabolism of motor neurons. Mutations in this gene result in insufficient SMN protein production, leading to progressive loss of spinal motor neurons and ultimately severe disability [3].

SMA is one of the most common conditions among so-called rare diseases, with a prevalence ranging from 1 in 6,000 to 1 in 10,000 live births worldwide [2]. Approximately 1 in 40 individuals is an asymptomatic carrier of the SMA mutation [2]. This inheritance pattern highlights the importance of genetic counseling as a preventive tool. As Alkuraya (2013) notes, *"genetics and genomic medicine represent essential components of preventive healthcare, and integrating genetic counseling into national health systems is critical, especially in regions with a high prevalence of recessive genetic diseases"* [5].

Moreover, the importance of timely identification is reinforced by clinical evidence. Dangouloff and Servais (2019) demonstrated that *"early treatment of patients with spinal muscular atrophy significantly improves outcomes when therapy is initiated prior to irreversible motor neuron loss"* [4]. This emphasizes the critical role of awareness and early diagnosis.

In addition, population-level approaches are becoming increasingly relevant. As *Nazareth, Lazarin, and Goldberg* (2015) highlight, *"carrier screening has undergone a paradigm shift in recent years, moving from ethnicity-based testing to expanded universal panels, reflecting the increasing need for population-level approaches to genetic disease prevention"* [6]. This perspective underscores the necessity of broad educational initiatives and carrier screening programs to reduce the risk of severe genetic diseases such as SMA.

Objective To assess the level of public awareness of spinal muscular atrophy (SMA) in children and the significance of genetic counseling in family planning.

Materials and Methods

A cross-sectional study was conducted using the online platform Google Forms. The study population consisted of residents of the Abai region. The survey was carried out from February 1, 2025, to March 25, 2025. The study was reviewed and approved by the Ethics Committee of Non-Commercial Joint Stock Company "Semey Medical University" (Protocol No. 2, dated December 12, 2023). All respondents were informed about the aim of the study. Participation was voluntary and anonymous.

The questionnaire consisted of 15 items, including: 6 sociodemographic questions (gender, age, marital status, education, presence and number of children), 4 questions on the importance of genetic counseling during pregnancy, 5 questions assessing awareness of SMA.

Inclusion criterion: residents of the Abai region aged 18 years and older.

Exclusion criterion: minors residing in the Abai region.

Quantitative data were tested for homogeneity of distribution using the Kolmogorov–Smirnov test. Based on the results, quantitative data are presented as medians. Qualitative data are described in absolute numbers and percentages.

For statistical analysis, descriptive statistics and the Pearson χ^2 test (table 3) were applied to examine associations between education and SMA awareness, as well as between gender and awareness. In addition, Cramer's V test (table 2) was used to assess the strength of associations between categorical variables. Data visualization was performed using distribution charts.

Results

A total of 315 respondents participated in the study, of whom 65 (20.6%) were men and 250 (79.4%) were women. The median age was 35–40 years, and the median number of children among respondents was 2. By educational level, 214 respondents (67.9%) reported having higher education. Regarding marital status, 195 (61.9%) were married. In addition, 232 respondents (73.7%) had children. When asked about family and pregnancy planning, 202 respondents (64.1%) stated that they intended to plan pregnancy with their spouse in the future (Table 1).

During family planning and pregnancy, 184 respondents (58.4%) expressed willingness to undergo genetic counseling, including genetic testing for mutations causing spinal muscular atrophy. However, awareness about SMA remained limited: 161 respondents (51.1%) did not know that spinal muscular atrophy is inherited. 182 respondents (57.8%) did not know that a person can be an asymptomatic carrier of the SMA mutation. 173 respondents (54.9%) were unaware of the main symptoms of SMA in children (muscle weakness, lethargy, weak sucking reflex, motor developmental delay, and respiratory difficulties).

Thus, the table shows that none of the sociodemographic factors (gender, age, education, marital status, presence of children) had a statistically significant correlation with the level of awareness of SMA. Statistical analysis using the Pearson χ^2 test showed no significant association between the presence of children and willingness to undergo genetic counseling. This indicates that having children does not influence the decision to participate in genetic counseling.

Discussion

The present study revealed that awareness of spinal muscular atrophy (SMA) among the surveyed population is insufficient, and this lack of knowledge does not significantly depend on gender or education level. These findings highlight that informational gaps are widespread and not confined to any particular demographic or social group.

Table 1.

Knowledge of Spinal Muscular Atrophy and the Role of Genetic Counseling: Evidence from the Abay Region.

		n	%
1. Gender	Male	65	20,6
	Female	250	79,4
Total		**315**	**100,0**
2. Age	18-25 years	62	19,7
	25-30 years	39	12,4
	30-35 years	37	11,7
	35-40 years	45	14,3
	40-45 years	47	14,9
	50 and older	85	27,0
Total		**315**	**100,0**
3. Education	No education	4	1,3
	Higher	214	67,9
	Incomplete higher	31	9,8
	Secondary specialized	27	8,6
	Secondary	39	12,4
Total		**315**	**100,0**
4. Marital status	Marital status	7	2,2
	Married	195	61,9
	Single (never married)	84	26,7
	Divorced	21	6,7
	Widowed	8	2,5
Total		**315**	**100,0**
5. Do you have children?	Yes	232	73,7
	No	83	26,3
Total		**315**	**100,0**
6. If yes, how many children?	None	83	26,3
	One	44	14,0
	Two	79	25,1
	Three	65	20,6
	Four or more	44	14,0
Total		**315**	**100**
7. Do you know what "genetics" and "genetic diseases" are?			
Yes		278	88,3
No		20	6,3
Difficult to answer		17	5,4
Total		**315**	**100,0**
8. Do you think genetic counseling is important in family planning and pregnancy?			
Yes		84	26,7
No		13	4,1
Difficult to answer		218	69,2
Total		**315**	**100,0**

	n	%
9. If you do not yet have children, do you plan pregnancy with your spouse in the future?		
Yes	202	64,1
No	60	19,0
Difficult to answer	53	16,8
Total	**315**	**100**
10. If planning pregnancy, would you undergo genetic counseling, including SMA testing?		
Yes	184	**58,4**
No	63	**20,0**
Difficult to answer	68	**21,6**
Total	**315**	**100,0**
11. Have you ever heard of spinal muscular atrophy (SMA)?		
Yes	150	58,4
No	148	20,0
Difficult to answer	17	21,6
Total	**315**	**100,0**
12. Do you know that SMA is inherited?		
Yes	140	44,4
No	161	51,1
Difficult to answer	**14**	**4,4**
Total	**315**	**100.0**
13. Do you know that a person may be an asymptomatic carrier of the SMA mutation?		
Yes	106	33,7
No	182	57,8
Difficult to answer	27	8,6
Total	**315**	**100,0**
14. Do you know that SMA and carrier status can be diagnosed with a blood test?		
Yes	121	38,4
No	172	54,6
Difficult to answer	22	7,0
Total	**315**	**100,0**
15. Do you know the main symptoms of SMA in children (muscle weakness, lethargy, weak sucking reflex, motor delay, tremor, respiratory problems)?		
Yes	112	35,6
No	173	54,9
Difficult to answer	30	9,5
Total	**315**	**100,0**

One of the key explanations for this outcome may be the limited dissemination of information about rare genetic diseases in public health campaigns, which predominantly focus on more common conditions. Furthermore, the absence of systematic premarital and preconception genetic counseling programs may contribute to low awareness levels, even among highly educated individuals. This underscores the need for nationwide initiatives aimed at increasing public understanding of hereditary disorders, particularly SMA, which is both life-limiting and preventable in terms of risk through carrier screening.

The findings of our study are consistent with international reports indicating insufficient knowledge of SMA and the benefits of genetic counseling in different countries. For example, studies by *Dangouloff and Servais* (2019) emphasize the critical role of early detection and treatment, while *Nazareth et al.* (2015) demonstrated that public awareness and trends in genetic carrier screening remain suboptimal despite technological progress. In contrast, in regions where genetic education and premarital screening are well-established, such as in *Saudi Arabia (Alkuraya, 2013),* awareness and participation in genetic counseling are significantly higher.

Table 2. **Assessment of public awareness in Abai region regarding spinal muscular atrophy in children and the importance of genetic counseling in family planning.**

	11.Have you ever heard of spinal muscular atrophy (SMA)?			
Gender	Yes	No	Difficult to answer	Total
Male	26	34	5	65
Female	124	114	12	250
Total	150	148	17	315
	12. Do you know that SMA is inherited?			
	Yes	No	Difficult to answer	Total
Male	25	37	3	65
Female	115	124	11	250
Total	140	161	14	315
	13. Do you know that a person may be an asymptomatic carrier of the SMA mutation?			
	Yes	No	Difficult to answer	Total
Male	16	42	7	65
Female	90	140	20	250
Total	106	182	27	315
	14. Do you know that SMA and carrier status can be diagnosed with a blood test?			
	Yes	No	Difficult to answer	Total
Male	19	41	5	65
Female	102	131	17	250
Total	121	172	22	315
	15. Do you know the main symptoms of SMA in children (muscle weakness, lethargy, weak sucking reflex, motor delay, tremor, respiratory problems)?			
	Yes	No	Difficult to answer	Total
Male	17	42	6	65
Female	95	131	24	250
Total	112	173	30	315
	11. Have you ever heard of spinal muscular atrophy (SMA)?			
Education	Yes	No	Difficult to answer	Total
No education	3	1	0	4
Higher	104	98	12	214
Incomplete higher	21	9	1	31
Secondary specialized	9	16	2	27
Secondary	13	24	2	39
Total	150	148	17	315
	12. Do you know that SMA is inherited?			
	Yes	No	Difficult to answer	Total
No education	3	1	0	4
Higher	92	111	11	214
Incomplete higher	17	13	1	31
Secondary specialized	15	11	1	27
Secondary	13	25	1	39
Total	140	161	14	315
	13. Do you know that a person may be an asymptomatic carrier of the SMA mutation?			
	Yes	No	Difficult to answer	Total
No education	3	1	0	4
Higher	70	123	21	214
Incomplete higher	14	15	2	31
Secondary specialized	9	18	0	27
Secondary	10	25	4	39
Total	106	182	27	315
	14. Do you know that SMA and carrier status can be diagnosed with a blood test?			
	Yes	No	Difficult to answer	Total
No education	2	2	0	4
Higher	82	116	16	214
Incomplete higher	13	16	2	31
Secondary specialized	10	16	1	27
Secondary	14	22	3	39
Total	121	172	22	315

Continuation of Table 2.

	15. Do you know the main symptoms of SMA in children (muscle weakness, lethargy, weak sucking reflex, motor delay, tremor, respiratory problems)?			
	Yes	No	Difficult to answer	Total
No education	3	1	0	4
Higher	69	122	23	214
Incomplete higher	18	11	2	31
Secondary specialized	11	15	1	27
Secondary	11	24	4	39
Total	112	173	30	315
	11. Have you ever heard of spinal muscular atrophy (SMA)?			
Marital status	Yes	No	Difficult to answer	Total
Marital status	4	2	1	7
Married	83	103	9	195
Single (never married)	50	29	5	84
Divorced	10	9	2	21
Widowed	3	5	0	8
Total	150	148	17	315
	12. Do you know that SMA is inherited?			
	Yes	No	Difficult to answer	Total
Marital status	5	1	1	7
Married	79	109	7	195
Single (never married)	44	37	3	84
Divorced	9	10	2	21
Widowed	3	4	1	8
Total	140	161	14	315
	13. Do you know that a person may be an asymptomatic carrier of the SMA mutation?			
	Yes	No	Difficult to answer	Total
Marital status	4	3	0	7
Married	60	123	12	195
Single (never married)	32	43	9	84
Divorced	8	9	4	21
Widowed	2	4	2	8
Total	106	182	27	315
	14. Do you know that SMA and carrier status can be diagnosed with a blood test?			
	Yes	No	Difficult to answer	Total
Marital status	2	3	2	7
Married	76	111	8	195
Single (never married)	34	41	9	84
Divorced	8	10	3	21
Widowed	1	7	0	8
Total	121	172	22	315
	15. Do you know the main symptoms of SMA in children (muscle weakness, lethargy, weak sucking reflex, motor delay, tremor, respiratory problems)?			
	Yes	No	Difficult to answer	Total
Marital status	3	3	1	7
Married	60	120	15	195
Single (never married)	43	33	8	84
Divorced	4	11	6	21
Widowed	2	6	0	8
Total	112	173	30	315
	11. Have you ever heard of spinal muscular atrophy (SMA)?			
Presence of children	Yes	No	Difficult to answer	Total
Yes	103	120	9	232
No	47	28	8	83
Total	150	148	17	315
	12. Do you know that SMA is inherited?			
	Yes	No	Difficult to answer	Total
Yes	97	126	9	232
No	43	35	5	83
Total	140	161	14	315

Continuation of Table 2.

	13. Do you know that a person may be an asymptomatic carrier of the SMA mutation?			
	Yes	No	Difficult to answer	Total
Yes	74	141	17	232
No	32	41	10	83
Total	106	182	27	315
	14. Do you know that SMA and carrier status can be diagnosed with a blood test?			
	Yes	No	Difficult to answer	Total
Yes	86	132	14	232
No	35	40	8	83
Total	121	172	22	315
	15. Do you know the main symptoms of SMA in children (muscle weakness, lethargy, weak sucking reflex, motor delay, tremor, respiratory problems)?			
	Yes	No	Difficult to answer	Total
Yes	69	142	21	232
No	43	31	9	83
Total	112	173	30	315

Table 3.

Variable (independent)	Dependent variable	χ^2 (df)	p-value	Cramer's V	Interpretation
Gender	Awareness of SMA	2.27 (2)	0.318	0.085	No significant association
Education level	Awareness of SMA	4.81 (4)	0.307	0.092	No significant association
Marital status	Awareness of SMA	3.92 (3)	0.270	0.089	No significant association
Presence of children	Willingness for genetic counseling	2.56 (1)	0.112	0.070	No significant association

These differences suggest that structured national policies and cultural integration of genetic literacy can markedly influence public awareness.

The strengths of this study include its relatively large sample size (315 respondents), which exceeds the required minimum for statistical representativeness, and the inclusion of diverse demographic groups within the Abai region. However, several limitations should be acknowledged. First, the use of an online questionnaire may have introduced selection bias, favoring individuals with higher digital literacy. Second, awareness was measured through self-reported answers, which may not fully reflect objective knowledge. Finally, as this was a cross-sectional study, causal relationships between sociodemographic factors and awareness cannot be established.

Despite these limitations, the study provides important insights into the current state of genetic literacy regarding SMA in Kazakhstan. The results may serve as a basis for designing educational interventions, incorporating genetic counseling into routine reproductive health services, and promoting targeted information campaigns. Future research should explore the effectiveness of such interventions, assess the role of healthcare providers in improving genetic awareness, and examine attitudes toward genetic testing in different cultural and social contexts

Conclusion

The study revealed a low level of public awareness about SMA. Given the high importance of early diagnosis and the possibility of prevention through genetic counseling, government and public initiatives are needed to promote knowledge about hereditary diseases. The introduction of genetic education programs can help reduce the risk of children being born with severe genetic disorders.

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