Knowledge, Attitudes and Practices of Mothers regarding their Children with Spinal Muscular Atrophy

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Abstract

Background: Spinal Muscular Atrophy (SMA) is a genetic disease affecting the central nervous system and voluntary muscle movement (skeletal muscle). **This study aimed:** Was to assess knowledge, attitudes and practices of mothers regarding their children with spinal muscular atrophy**. Research design:** A descriptive research design was used in carrying out this study. **Setting:** The study was conducted at Hassan Awad Health Insurance Clinic in Banha City. **Sample:** Purposive sample of 100 mothers and their children with SMA. **Tools:** Three tools were **used. Tool 1:** Consisted two parts: **Part (1):** An interviewing questionnaire consisted to assess socio-demographic data of studied sample, and personal characteristics of preschool children with spinal muscular atrophy. **Part (2)** Mothers` knowledge regarding SMA. **Tool (II)** Mothers` attitude likert scale**. Tool (III):** Reported practices of mother regarding their children with SMA**. Results:** 36.0% of the children with SMA were aged from 1< 3 years . 93.0% of studied children had motor problems of difficult standing. 60.0 % of mothers had poor total level of knowledge regarding spinal muscular atrophy,66.0 % of them had negative attitude and 53.0 % of studied mothers had unsatisfactory total reported practices. **Conclusion:** Most of studied children had motor problems of difficulty standing, There were statistically significant correlation between studied mother`s total knowledge, total attitude and total reported practices. **Recommendations:** Health education program should be given for mothers about SMA to enhance their knowledge, attitude and practices regarding disease.

Keywords: children, Mothers` knowledge, Mothers` attitude, Mothers` practices, Spinal Muscular Atrophy (SMA).

Introduction

Childhood is a time when developmental changes are happening that can have profound and lasting consequences for a child's future. Children are the backbone of a nation as on children' health and prosperity depend on the health of a nation. Children need the care, protection and guidance that are normally provided by mothers, especially during the early years when they are most dependent **(Haraldstad et al., 2019).**

Spinal Muscular Atrophy (SMA) is a common autosomal recessive disorder that causes degeneration of anterior horn cells in the human spinal cord and subsequent loss of motor neurons. SMA is clinically divided into five subtypes; type 0 (the most severe form with onset in the prenatal period(severe respiratory problems after birth), type I (Werdnig–Hoffmann disease, a severe form with onset before 6 months of age; the inability to sit unsupported), type II

(Dubowitz disease; an intermediate form with onset before 18 months of age; the ability to sit unaided, but not to stand or walk), type III (Kugelberg–Welander disease; a mild form with onset after 18 months of age; the ability to stand and walk unaided), and type IV (the mildest form with onset after 30 years of age) **(Kimizu et al., 2021).**

Anciently, SMA is a rare disease, and the incidence of SMA varies among countries. Reports of SMA incidence were based on clinical symptoms and were mainly based on genetic testing. Currently, the number of SMA incidence based on newborn screening is increasing. The incidence of SMA continues to change owing to many factors, including awareness about the disease among the population. It should be noted that the current newborn screening program does not include detection of an intragenic mutation in Survival Motor Neuron gene (SMN1) because the occurrence of intragenic mutation in SMN1 is very rare. Among these subtypes, SMA type I accounts for half of all SMA patients and is a leading inherited cause of infant mortality. Many children with SMA type I die of respiratory insufficiency by the age of 2 when respiratory support is not available. Meanwhile, children with types II, III, and IV are able to survive for longer times with limited motor function. .Spinal muscular atrophy is the second fatal neuromuscular disease after cystic fibrosis. Globally, the incidence of SMA is about 1 in 10,000 live births and a carrier frequency of 1/60. The prevalence is approximately 1–2 per 100,000 persons so, SMA is a relatively rare condition **(Baranello et al., 2021; Nicolau et al., 2021).**

Spinal Muscular Atrophy causes deterioration in the functioning of multiple tissues including those found in skeletal muscle, heart, autonomic and enteric nervous systems, metabolic/endocrine system, lymphatic systems, and reproductive system. Therefore, dysfunction of multiple organ systems may occur in a SMA children as the disease progresses. SMA children may need medical care and nursing support for daily activities as well as for long-term management of several medical devices **(Weaver et al., 2020).**

Current treatment options in SMA include ventilatory support including noninvasive and invasive ventilation, management of secretions including inhalational therapies and section, feeding support including nasal feeding or placement of a percutaneous endoscopic gastrostomy and supply with medical devices to increase rate of improvements regarding survival and quality of life of SMA children. In pediatric neurology, attentions mostly has focused on developing novel technologies and pharmaceuticals for children with SMA. It focused on investigating the impacts of the clinical treatments on the quality of life of children and caregivers **(Strauss et al., 2022).**

Mother plays an essential role in child health control not only during infancy and childhood but also during pregnancy in the embryo stage. Maternal health and adequate antenatal follow up have an impact on early detection of any congenital problems in the fetus. Mothers who have children with age less than 3 years should increase their awareness and knowledge about spinal muscular atrophy, and to understand the physiological changes and nutritional need during each stage of child growth **(Cremers et al., 2019).**

Community health Nurses should demonstrate actions through skill development, activities and providing support that enhances self-efficacy of for successful behavior changes. These actions represent

key elements of the health belief model and can be used to design or adapt health promotion or disease prevention programs. The nurse should communicate to the mothers the steps that are involved in taking the recommended action and highlighting the benefits to action. Providing assistance in identifying and reducing barriers to action **(Motta & Paulo2020).**

Community health Nurse also has a relevant role in health care for children population mainly physical, psychological preparation of children and control of the specific adverse reactions of spinal muscular atrophy. Nurses play an important role in the prevention of SMA through health education. Nurses should take urgent steps through motivation and to create awareness about SMA. Health education raising mothers' awareness about SMA, its complication and adverse effects not only on children but also on family as a whole **(Fernandes et al., 2022).**

Significance of the study:

Egypt had an efforts to treat children suffering from muscle atrophy. It cost up to 3 million per child and Egypt is determined to secure it although many other countries cannot afford it. Account to help there are 204 muscle atrophy cases have been detected in Egypt, of which more than 32 can be treated. The Ministry of Health affirmed that Egypt will start the treatment of 10 patients with muscular atrophy out of 32 at Nasser Institute and Ain Shams Hospital **(Muscular Dystrophy Association, 2021).**

Spinal Muscle Atrophy is one of the most common neurodegenerative diseases of genetic background the child's age. Regardless of the form, it is essential to enhance mother' knowledge and attitude that will help in accepting disabilities as well as in the preservation of preschool children -care. So the investigator found urgent to assess the knowledge and attitudes of mothers regarding their preschool children with spinal muscular atrophy.

Aim of the study

The aim of this study was to assess knowledge, attitudes and practices of mothers regarding their children with spinal muscular atrophy.

Research questions:

1-What are the health problems of children with spinal muscular atrophy?

2-What are the mothers` knowledge regarding the spinal muscular atrophy?

3-What are the mothers` attitude regarding their children with spinal muscular atrophy?

4-What are the reported practices of mothers regarding spinal muscular atrophy?

5- Is the correlation between the mothers` knowledge, attitude and reported practices regarding their children with spinal muscular atrophy?

Subject and Methods

Research design:

Descriptive research design was utilized to conduct this study. Descriptive research design is described accurately and systematically a population, situation or phenomenon. It can answer what, where, when and how questions, but not why questions. A descriptive research design could use a wide variety of research methods to investigate one or more variables.

Setting:

This study was carried out at Hassan Awed Health Insurance Clinic in Benha City, which consists of three floors and there is a spinal muscular atrophy clinic on the second floor. The examination takes place every Monday per week.

Sample:

Purposive sample of mothers and their children with Spinal Muscular Atrophy from previously mentioned setting. All mothers accompanied their children attending at

Hassan A wad Health Insurance Clinic in Banha City.It included 100 mothers take their children were regarding of their characteristics and willing to participate in the study.

Tools of data collection:-

Three tools were used in this study to collect the data.

Tool 1: A structured interviewing Questionnaire: It was developed by the investigator and revised by supervisor staff, based on reviewing related literatures and it was written in a simple clear Arabic language. It consisted of two parts to assess the following:

Part I: It consisted of two parts to assess the following:-

(A):-It was concerned with Socio-demographic data of mothers involved in the study. It included 6 questions (age, level of education, social status, occupation, residence and monthly income).

(B):-It was concerned with personal characteristics of preschool children with spinal muscular atrophy. It included 3 closed ended questions (child age, gender and child ranking in the family).

Part II:- It was concerned with the mother`s knowledge regarding spinal muscular atrophy .It included 14 questions (meaning, causes, types of Spinal Muscular Atrophy, meaning of wording-Hoffmann's, meaning of chronic neonatal muscular dystrophy, meaning of Gilliberg-wilander disease), symptoms, tests and examination, diagnoses, complications, treatment, surgical treatment, pharmacological treatment, supportive devices, way to prevent Spinal Muscular Atrophy and source of information about Spinal Muscular Atrophy).

**Scoring system of Mother`s knowledge:** -It was calculated as following 2 for correct and complete answer, while 1 for correct and incomplete answer and 0 for don`t know .These scores converted into a percent and classified as the following.

N.B-Source of information about spinal muscular atrophy not included in the scoring system.

Total scores of knowledge= 28 points.

- **Good:** If the total score of knowledge was ≥ 75% (21 points).
- **Average:** If the total score was 50 < 75% (14<21 points).
- **Poor:** If the total score was < 50 % (<14 points).

Tool II:- Mother`s attitude likert scale adopted from **(Hjorth**, **2020)** was used to assess mother`s attitude regarding their children with Spinal Muscular Atrophy.

It included (believing that diagnosis of Spinal Muscular Atrophy is essential to prevent progression of the disease, thinking that early diagnosis is the better for the child health, thinking that necessary to educate parents about the health problem, thinking that important from the early stage to develop the muscle strength of the child and maintain the flexibility of the rib cage, believing that physical therapy helps to take care of the muscles and decrease deterioration, thinking that spinal muscular atrophy can be treated, feeling the interest of the state and government institutions of children with spinal muscular atrophy, feeling the interest of the state and government institutions for children with spinal muscular atrophy, feeling that taking care of the child physically cause psychological burden, thinking that the psychological stress of caring for a child with SMA is results in a family conflict- feeling afraid of losing the child, thinking that spinal muscular atrophy is a serious disease and its complications are serious).

Scoring system for mother`s attitude:

The scoring system for mother`s attitude was graded as 2 for always,1 for sometimes and 0 for never.

The total attitude scores were categorized into levels as following:

Total scores of attitude = 22 points.

- **Positive:** If the total score of attitude was ≥ 60 % (≥13 points).
- **Negative:** If the total score of attitude was < 60 % (<13 points).

Tool III:-It was concerned with the reported practices of mothers regarding their children with Spinal Muscular Atrophy. It was divided into 4 main items included; nutritional practices, activity practices, psychological practices, follow-up and treatment practices. These categories consisted of 25 items classified as follow:

Nutritional which included 7 items (give the child food in vitamin D (salmon-eggs-cod liver oil), increase vegetables and fruits in child`s diet (orange-spinach), supply the child with food rich in energy (bananas-potatoes-apples-dark chocolate), give the child light and regular meals , give the child food rich in calcium (mozzarella cheese- yogurt-milk-oats-oranges), give the child food rich in iron (yolk-red meat-legumes-sea food) and use-lollipops and support the child during feeding.

Activity which included 5 items (observe the child during daily activities, monitor the child`s sleep periods, monitoring for any signs of lethargy on the child, doing exercises for the child constantly to improve muscle strength and put the child in the appropriate positions for comfortable breathing.

Psychological which included 7 items (provide the child with psychological and spiritual support, participate the child in some activities that do not require physical activities, involve the child in making decisions about his affairs, try not to appear sad and cry in front of the child so that it does not worse mental state, encourage child to swim and exercise to maintain muscle strength as much as possible, involve child in going out to the club to integrate with the community and encourage child to make friends with children who practice physical therapy with him.

Follow-up and treatment which included 6 items (stick to the follow-up appointments determined by the doctor treating the child1s condition, commit to using physical therapy, commit to using the supportive aids designated to help movement, commit to using the supportive aids designated to help breathing, notice any complications for your child and report them to the doctor and stick to the treatment dates and medications prescribed by the child physician.

Scoring system of total reported practices mothers of studied regarding their children with Spinal Muscular Atrophy was calculated as follow:

1 for done and 0 for not done. These scores of the items were summed-up and the total divided by the number of the items, giving a mean score. As well as total reported practices score was classified as the following:

Total scores of reported practices = 25 points

- **Satisfactory** when the total score was ≥ 60% (≥ 15 points).
- **Unsatisfactory** when the total score was < 60 % .(< 15 points).

Content validity of the tools:

The tools validity was done by five members Faculty`s Staff Nursing-Benha University Experts from the Community Health Nursing Specialties who reviewed the tools for clarity, relevance, comprehensiveness ,applicability and easiness for implementation and according to their opinion minor modifications were carried out.

Reliability of the tools :

The reliability was done by Cronbach`s Alpha coefficient test that developed by Lee Cronbach in 1951 which revealed that each of the three tools consisted of relatively homogeneous items as indicated by the moderate to high reliability of each tool. The internal consistency of knowledge was 0.711, attitude was 0.825 and the internal consistency of reported practice was 0.783.

Ethical considerations:

The investigator clarified the aim of the study to the mothers included in the study. Mothers` oral consent were obtained from them before their participation in the study .They were also reassured that all information gathered would be confidential and used only for the purpose of the study. They were also informed about their right to withdraw at any time from the study without giving any reasons.

Pilot study:

The pilot study was carried out in the mid of March 2022 to ascertain the clarity and applicability of the study tools. Data about it included Preparatory phase and data collection phase through two weeks were included in pilot study. It has also served in estimating the time needed for filling questionnaires. It ranged between30-45 minutes to assess knowledge, attitudes and practices of mothers regarding their children with Spinal Muscular Atrophy. No modifications were done, so the pilot study included in the study main subjects.

Field work:

Data collections were carried out in the period from the beginning of April 2022 to the end of September 2022 covered 6 months. The investigator was available in the study setting one day every week (Monday) to collect data. The average number of interviewed mothers were 4-5 mothers per day At the beginning of interview; the investigator welcomed each mother and introduce herself to them. The title, objectives, tools and the study technique were illustrated for each mother to obtain their approval and cooperation which was needed for conducting this study. Each mother was individually interviewed with a questionnaire. The time needed for filling out the tools was around 30-45 minutes depending on the responses of the interviewers. Samples reward such as (simple candy) was giving to children to encourage them to participate in the study.

Statistical analysis: -

All data collected were organized, tabulated and analyzed by using the Statistical Package for The Social Science (SPSS version 21), which was used frequencies and percentages for qualitative descriptive data and chi-square coefficient x^2 was used for relation testes and mean standard deviation was used for quantitative data.

The observation difference and associations were considered as the following: (p-value)

- Highly statistically significant P<0.001**
- Statisticaly significant p<0.05*
- Not –significant p > 0.05

Results:

Table (1): Shows that; 41.0% of the studied mother`s aged from 30<40 years old with mean and standard deviation was 30.65±4.21. 35.0% of them had intermediate education and 91.0% of them were married. 75.0% of them did not work. 62.0% of them from rural areas, 56 .0% of them their monthly income did not enough.

Table (2): Shows that; 36.0% of the studied children with SMA aged from 1<3 years old with Mean ±SD 3.21±2.14 whenever, 56.0%

of them were male and 42.0% of them were the first child in the family

Figure (1): Shows that; 60.0% of mothers had poor total level of knowledge regarding spinal muscular atrophy while 27.0% of them had average total level of knowledge and 13.0% of them had a good total knowledge level regarding spinal muscular atrophy.

Figure (2): Illustrates that; 66.0% of mothers had negative attitude toward spinal muscular atrophy and 34.0% of them had positive attitude.

Figure (3): Illustrates that; 53.0% of studied mothers had unsatisfactory total reported practices regarding their children with spinal muscular atrophy and 47.0% of them had satisfactory total reported practices regarding their children with spinal muscular atrophy.

Table (3): Clarifies that; there were statistically significant correlation between studied mothers total knowledge, total attitude and total practices $p < 0.05$**.**

Table (1): Distribution of the studied mothers regarding their socio-demographic data (n=100).

Socio-demographic data	%
Age \ years old	
20 < 30	40.0
30 < 40	**41.0**
≥ 40	19.0
Mean ±SD 30.65±4.21	
Level of education	
Can't read or write	17.0
Basic education	17.0
Intermediate education	**35.0**
University education	31.0
Marital status	
Married	**91.0**
Widow	5.0
Divorced	4.0
Occupation	
Work	25.0
Not work	**75.0**
Residence	
Rural	**62.0**
Urban	38.0
Monthly income	
Not enough	**56.0**
Enough	39.0
Enough and save	5.0

Table (2): Distribution of the studied children with SMA regarding their personal characteristics (n=100).

Personal characteristics of preschool children	%
Age\ years	
< 1	21.0
1<3	**36.0**
3<6	32.0
≥6	21.0
Mean ±SD 3.21±2.14	
Gender	
Boy	**56.0**
Girl	44.0
Ranking of the child in the family	
The first child	**42.0**
The second child	34.0
The third or more	18.0
The only child	6.0

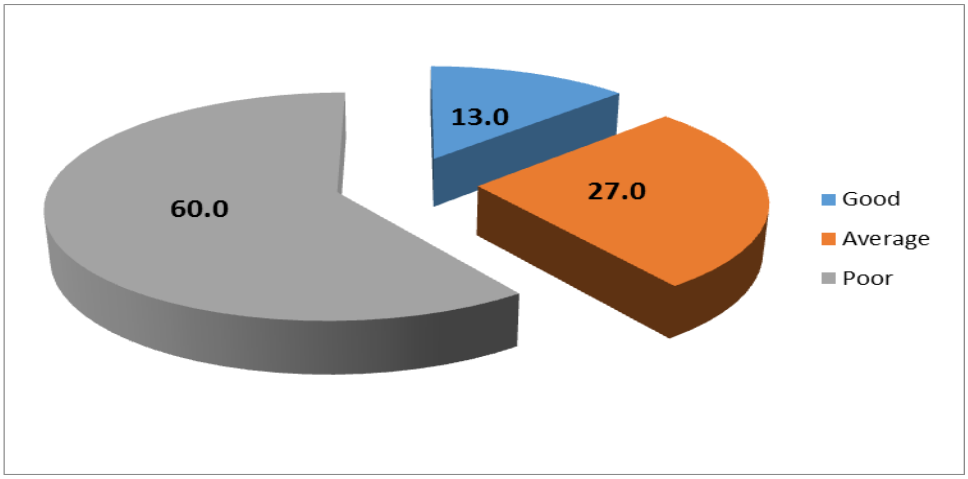

Figure (1): Distribution of studied mothers` total knowledge level regarding SMA (n=100).

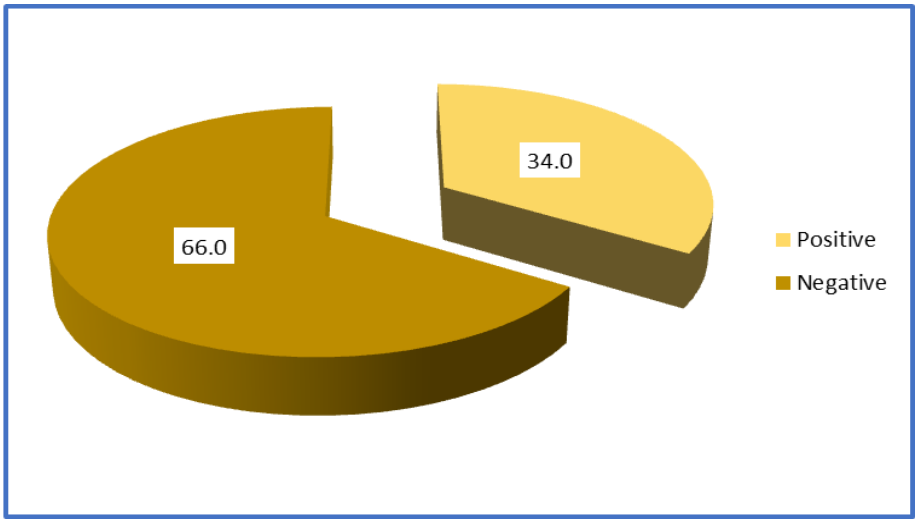

Figure (2): Distribution of studied mothers total attitude level SMA (n=100).

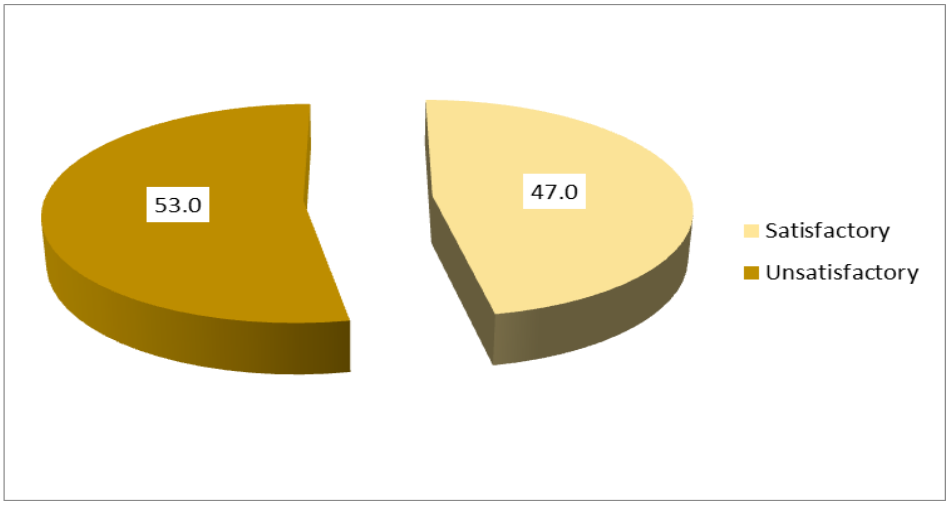

Figure (3): Distribution of studied mothers` total practices level regarding their children with SMA (n=100).

Table (3): Correlation matrix between studied mothers` total knowledge, total attitude and total practices level (n=100).

		Total knowledge	Total attitude	Total practices
Total knowledge	**r**	**1**	**.424**	**.858**
	p-value		.011*	.018*
	n	100	100	100
Total attitude	r	.424	1	.891
	p-value	.011*		.014*
	n	100	100	100
Total practices	r	.858	.891	1
	p-value	.018*	.014*	
	n	100	100	100

Discussion

Spinal muscular atrophy (SMA) is a rare, genetic, neurodegenerative, and disabling disease, whose symptoms can appear from early childhood to early adulthood. It results in the dysfunction and death of lower motor neurons, leading to decreases in appendicular, axial, and medullary strength, culminating in difficulty in swallowing, speaking, breathing, and maintaining cognitive integrity and sensory and sensorial functions **(Nurputra et al., 2022).**

Spinal Muscular Atrophy of children is a severe disease that has an impact on the whole family. Even though the disease is severe and families face a challenging trajectory with progressive losses, anticipatory grief, and ethical dilemmas regarding treatment, knowledge of how families experience their child's care and everyday life is limited. Respecting autonomy and adequate communication of the diagnosis and prognosis of neurodegenerative and disabling diseases, such as SMA, impact the relationships among the physicians, patients, and family caregivers; the mental health of those involved and the therapeutic follow-up **(Anestis et al., 2020).**

Regarding socio- demographic characteristics of the studied mothers, the

current study revealed that more than one third of the studied mothers aged from 30 to less than 40 years old had a mean & standard deviation 30.65 ± 4.21 years old and less than two thirds of the studied mothers were living in rural areas .This could be due to less accessibility to medical services in rural areas so the mothers come to hospitals. Regarding educational level, the current study showed that more than one third of the studied mothers had intermediate education

Regarding personal characteristics of the studied children, the current study showed that less than two fifths of children aged from 1 to less than 3years old. These findings agreed with **Ovgren et al., (2016),** who found that more than one third of the studied children aged from 1to less than 3years old or more. This might be due to the age stage of children for walking and activity.

Regarding gender of the studied children, the current study revealed that more than half of children were male. This result was disagreed with **Metcalfe et al., (2018),** who conduct study of ‘‘Family communication between children and their parents about inherited genetic conditions’’ in UK (n=100) and found that more than half of the studied children were female. This might be due to gender differences were not fully understood but probably reflect differences in exposure to environmental risk factors and endogenous hormones, as well as complex interactions between these influences.

Regarding total mothers’ knowledge level about spinal muscular atrophy, the present study showed that less than two thirds of the studied mothers had poor total level of knowledge. This finding was paralleled with **Vankruijsbergen et al., (2021)** who studied “Parents’ perspectives on nusinersen treatment for children with spinal muscular atrophy” in German (n= 75)and found that the more than half of the studied subject had poor total knowledge level about spinal muscular atrophy. This might be due to shortage of campaigns that educate mothers’ sufficiently regarding causes, types and treatment of disease and significant association between mothers’ level of knowledge and education level.

Regarding mothers total attitude regarding spinal muscular atrophy, the current study showed that, less than two thirds of the studied mothers had negative attitude toward spinal muscular atrophy. This result was agreed with **Kocova, et al., (2019)** who conduct of “Health-related quality of life in children and adolescents with spinal muscular atrophy in the Czech Republic” in Czech Republic (n=120) and found that two thirds of the studied subjects had negative attitude toward spinal muscular atrophy. This might be due to poor level of knowledge related to spinal muscular atrophy which reflected negatively in mothers' attitude about this disease.

On assessing mothers’ total practices regarding their children with spinal muscular atrophy, the current study showed that, more than half of studied mothers had unsatisfactory total practices regarding their children with spinal muscular atrophy. This finding was paralleled with **Fernandes et al., (2022)** and found that two thirds of the studied mothers had unsatisfactory total practices regarding their children with spinal muscular atrophy**.** This could be due to the inadequate mothers’ knowledge and the residence had an adverse effect on proper practice toward children with SMA disease

Concerning correlation between mothers` total knowledge, total attitudes and total practices level studied. There were statistically significant correlation between studied mothers total knowledge, total

attitudes and total practices. This result was consistent with **Yeo et al. (2021)** who found that there was positive correlation between total knowledge, total attitude and total practices. This might be due to mothers' knowledge, attitude directly influenced by each other during caring of children. Moreover, knowledge and practice was considered the baseline that helping in achieving best outcome regarding prevention of SMA in children.

Conclusion

Most of studied preschool children with spinal muscular atrophy had motor problems of difficulty standing and waking, and majority of them had gastrointestinal problems of inability to swallow, and three fifths of studied mothers had poor total knowledge regarding spinal muscular atrophy, also more than two thirds of them had negative attitude regarding spinal muscular atrophy, while slightly more than half of studied mothers had unsatisfactory total practices regarding their children with spinal muscular atrophy, while there were statistically significant correlation between studied mother`s total knowledge, total attitude and total reported practices.

Recommendations

- Health education programs should be developed and implemented for mothers to improve their knowledge, attitudes and practices about spinal muscular atrophy.
- A periodic, comprehensive health campaigns should be carried continuously to aware community about spinal muscular atrophy disease and the reflection of early diagnosis and proper management in minimizing complication of spinal muscular atrophy on children health condition.
- **Further research:**

1-Evaluate quality of life of children with spinal muscular atrophy and asses burden of care for their caregivers.

2-Psychological training programs for mothers to deal positively with society and cope with their children with spinal muscular atrophy.

3- Replication of the study on a larger probability sample in other different settings is highly recommended to achieve generalizable results.

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