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PHD THESIS

IMPROVEMENT OF PROFESSIONAL ABILITIES IN THE COMMUNITY MEDICAL NURSES SYSTEM, IN ORDER TO OPTIMIZE CASE MANAGEMENT IN RARE DISEASES

– A B S T R A C T –

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INTRODUCTION

I dedicate these studies and my entire doctoral thesis to people with rare diseases and community nurses. I hope that together they will navigate through the Romanian health system more easily!

The integrated medical-social approach of patients with rare disease is an important topic in Romanian and global health policy. The concept of community care/intervention of the patient, the medical approach of the beneficiary with medical and social vulnerabilities, involves teamwork consisting of at least two professional categories, the medical and social professionals.

The concept of integrated medical-social approach has become more and more common within the activity of the community nurse who, meeting cases of patients with different pathologies, has been able to find a way to help the patients with different diseases. including the rare disease or the suspicion that some patients have symptoms unknown to them, they had to seek information from family doctors and doctors specialised in the management of various diseases, including rare diseases. Community nurses identify and monitor annually a minimum of about 1,200,000 beneficiaries of which about 1,500 patients with rare and genetic diseases, number estimated in 2023 according to the reports existing in the application amcmsr.gov.ro of the Ministry of Health). Since the beginning of 2024, community nurses have applied the case management method in monitoring children, pregnant women and patients with rare disease, the method they will apply to all beneficiaries regulated by the specific legislation, because the application is in an upgrade process to allow the collection of data based on which the case management applied to each beneficiary will emerge.

GENERAL PART

RARE DISEASES

Rare diseases, many of which have a genetic origin, affect a small number of individuals in the population. Despite their individual rarity, rare diseases still have an increased impact, affecting a significant number of people. Globally, it is estimated that about 5% to 7% of the population lives with a rare disease, or about 300 million people, but only 50% of them will have a clear diagnosis.

Of the approximately 7,000 clinically defined rare diseases, 72% are genetic and the majority (70%) are manifest in early childhood. That's why, although rare diseases can be present at any age, about 50% of affected people are children.

Rare diseases (often called "orphan") are conventionally defined as those affecting a very small number of individuals, but may be associated with inadequate management, chronic debilitation, and a dramatic health impact that can lead to death. They are quantified differently in different parts of the world, and there is still no international consensus on this.

For example, in the European Union, a disease is considered rare if it affects no more than 1 person in 2,000, while in the United States, the Orphan Medicines Act defines rare diseases as diseases that affect less than 200,000 people.

Traditional techniques for diagnosing rare diseases were largely based, until recently, on heuristic approaches, coupling clinical experience with medical literature. But many patients remained undiagnosed for years..

In recent years, the development of genetic testing has taken on a special scale and helped identify the molecular cause of many such diseases. These technologies have allowed diagnoses to be elucidated in a considerable proportion of previously undiagnosed patients. Genetic testing, such as exome sequencing and whole genome sequencing, has become one of the most important and powerful methods for detecting a rare disease. The most commonly used genetic test to diagnose rare diseases is whole exome sequencing (WES).

Nearly 95% of rare diseases do not have treatment now. Among the reasons are: the low number of subjects with a particular condition that can be included in clinical trials and subsequently use the obtained treatment, the lack of financial support for therapies, and complicated regulatory requirements for approving drugs in general. However, a number of revolutionary therapies have recently been introduced for rare diseases that previously had

no treatment. Due to the high cost, treatments for rare diseases are often unavailable, especially in low- and middle-income countries, or their availability in these countries is much delayed.

In Romania, the issue of rare diseases is quite complex, the complexity being given by the lack of legislation adapted to the needs of patients with rare diseases, the lack of specialized staff, both in health units and in the vicinity of the home of these patients. At the level of rural territorial administrative units, it is estimated that approximately 200 do not have a family medicine cabinet under contract with the county health insurance house. Also, the community healthcare network is undersized due to insufficient funding.

COMMUNITY HEALTHCARE IN ROMANIA

Home care has old roots in the Romanian healthcare system. The nurses worked in medical dispensaries before transforming them into family medical offices under contract with the county health insurance houses. They were part of the medical dispensary team together with the family doctor, in some cases also a paediatrician, and nurses. The care nurses ensured visits to families where there were pregnant and newborns, thus making the connection between the family medicine cabinet and the households visited, sometimes discovering other medical problems in the members of the visited households, medical problems that they announced to the family doctor who made community visits to sick people.

Community health care is a component of primary health care and is the first specialised medical resource that comes into direct contact with the patient and his/her family. Also, the community nurse is the specialist in the health system who connects the patient with medical service providers, social service providers, educational service providers, between the patient and institutions and / or patient organisations or other non-governmental organisations that can influence for the better the health and socio-economic status of the patient and his caregivers.

The literature notes that a certain eating behaviour of the community nurse can have a certain influence on his health and professional activity. Great emphasis is placed on the importance of promoting attitudes and behaviours that support a healthy lifestyle, including community-based health education initiatives through community-based public health actions and interventions, especially to people belonging to vulnerable groups. It is essential that community nurses implement correct healthy lifestyle practices for their own person to provide already verified information about dietary style analysis.

Empathy is an emotional experience that takes place between an observer and a subject.

In healthcare, empathy refers to the ability to understand and feel what the patient experiences and feels, consequences of his illness. Empathy is therefore the ability to recognize and validate fears, anxieties, pain, worries that the patient has, to gather as much information as possible for a precise diagnosis and personalised treatment.

For community nurses and other medical or social professionals involved in the care of a patient with a rare disease, cultivating and expressing empathy should be both out of concern for the patient and for oneself, becoming a constant of daily practice.

INTEGRATED MANAGEMENT OF RARE DISEASE CASES IN ROMANIA

Currently, Romania does not have a national registry specifically dedicated to rare diseases and we believe that establishing a comprehensive rare disease monitoring system is crucial in facilitating evidence-based health policies.

The need to involve community nurses in the follow-up of patients with rare diseases emerged both from the projects in which community nurses were involved and from the attributions highlighted in the job description, attributions that necessarily include visiting families in the community to identify their medical and medical-social problems.

In Romania there are 1920 community nurses. Since 2019, 137 community nurses have been working alongside social workers from city halls and school counsellors, within integrated community teams, through the POCU 122607 project "Creation and implementation of integrated community services to combat poverty and social exclusion", providing medical-socio-educational services to reduce poverty and facilitate social inclusion. Through case management, the community nurse supports and connects the patient with rare disease with the services he needs, whether they are medical, social and educational services, depending on the identified needs.

The amcmsr.gov.ro application for making the case management report of the rare disease patient is very useful to carry out the effective monitoring of the rare and genetic disease patient. The application is the tool for reporting the activity of the staff in the community health system and creates a database with the samples identified and monitored from the level of each territorial administrative unit that carries out community health activity. Analyses of the impact of piloting CN training in rare disease case management demonstrated that

people living with a rare disease and caregivers who received case management services improved their level of information about the disease and their rights, their knowledge of services available and the ability to self-manage.

SPECIAL PART

OBJECTIVES

Community nurses are nurses who work in the communities of the territorial administrative units they serve. In Romania, the purpose of community nurses' activity is to increase population and vulnerable groups access to quality medical and social services. Because the approach in community healthcare is holistic, community nurses address both the physical and psychosocial aspects of the disease, creating a patient-centered care model that contributes to increasing the quality of life of patients and their caregivers.

People affected by a rare disease are a vulnerable social group in need of access to integrated care and services. The relationship of the community nurse with the patient with rare disease is based on a very high emotional charge due to the interaction between them because of the complexity of the medical problem in these cases and the need for multidisciplinary intervention.

The objective of this project is to assess the impact at national level of community nurses network activity in rare diseases case management development and what factors should be modulated to improve the results of their activity in this field.

Specific aims:

1. To assess how community nurses' empathy is influenced by bio - psyho – social factors.
2. To characterise the health status of the community nurses network members and identify how it can be improved by lifestyle changes.
3. To investigate the role of community nurses network activity in rare diseases case management in Romania.

STUDY 1: THE IMPACT OF THEORY OF MIND, STRESS AND PROFESSIONAL EXPERIENCE ON EMPATHY IN ROMANIAN COMMUNITY NURSES - A CROSS-SECTIONAL STUDY

Empathy, in healthcare, including community nursing, refers to the ability to understand and feel what the patient is experiencing and feeling. It is the ability to recognize and validate fears, anxieties, pain, worries that the patient has, to gather as much information as possible for an accurate diagnosis and personalised treatment. The community nurse has the chance to interact with the patient in his home and thus the degree of empathy can be higher also due to observing his socio-economic situation and interacting with the patient's defenders.

Our purpose was to determine empathy level and to identify which of the socioeconomic status (SES) and psychological factors were able to predict highest empathy levels in a Romanian sample of community nurses. To the best of our knowledge, empathy was not studied in a Romania national sample of community nurses.

Community nurses were invited in January-February 2023 to provide an answer to an online survey, using an advertisement in a professional network. 1580 participants voluntarily agreed to take part in this study, with a response rate of 85.8%. The survey included the Toronto Empathy Questionnaire, the Reading the Mind in the Eyes Test and socio-economic status items. A multivariate model for the prediction of belonging to the highest quartile of empathy as opposed to lowest quartile was constructed using SES and psychological variables as factors.

As required by roles played by these professionals, the mean levels of empathy are increased, with 74.7% of the sample being over the threshold of high empathy level (scoring > = 45), while the mean (SD) reported score was 49.1. A recent study investigating the empathy among senior medical students from Romania has found a similar mean (SD) level of empathy of 48.76. Using the same instrument, when studying nurses that work in psychiatric wards, Alhadidi et al. have identified lower levels of empathy, reporting a mean (SD) of 46.07, along with other studies that showed a decreased level of empathy.

Our results indicate that as experience as a community nurse accumulates, the empathy level increases: nurses with more than 6 years of experience had 56% more chances to be in the higher quartile of empathy, suggesting that job or field retention is a key factor for increased empathy. Observing the tertiles of years of experience, the empathy level increases steadily.

Possible explanations on the differences between our study and studies on nurses working in high demanding positions like in ICU, oncology or psychiatry or while in training, lie

in the job description of community nurses, which are engaged in a diverse and challenging health landscape, providing care is different settings from patient's own houses, nursing homes, family doctor practices, or other types of clinics.

High levels of self-perceived stress are a good predictor of low levels of empathy, both in univariate and multivariate models. Others have also reported negative associations between stress/anxiety and empathy, with stress/ anxiety management having a positive effect on empathy levels.

In the multivariate analysis, predictors of belonging to the highest quartile of TEQ, as opposed to the lowest quartile were: low self-perceived stress level (OR = 2.098, 95%CI 1.362–3.231), higher experience as a community nurse (OR = 1.561, 95%CI 1.120–2.175) and higher levels of the theory of mind (OR = 1.158, 95%CI 1.118–1.199), when controlling for gender, age, relationship status, presence of children in families, education, and income.

Our study did not find significant differences in empathy levels between genders in this population, similar to several studies on nursing students or psychiatric ward nurses or ICU nurses. Also, education level, familial background and income levels did not influence the empathy level in our sample.

Two of the strengths of this study are the large number of participants and the high response rate of 85.8%, leading to the assessment of a representative national sample of community nurses from Romania.

The analysis of empathy in a large sample of Romanian community nurses showed good levels of empathy, since almost three quarters of them scored high empathy levels. The capacity of reading emotions (theory of mind abilities) and higher experience, along with low levels of stress, led to higher levels of empathy. Future early-career training programs targeting to increase emotional competences, reduce levels of stress and encourage personnel retention could promote increased quality of community nursing in Romania.

STUDY 2 - A COMPREHENSIVE ANALYSIS CONCERNING EATING BEHAVIOUR ASSOCIATED WITH CHRONIC DISEASES AMONG ROMANIAN COMMUNITY NURSES

Lifestyle factors, including inadequate eating patterns, emerge as a critical determinant of chronic disease. Beyond their dedicated roles in patient care, nurses should actively participate in monitoring and managing their own health. Nurses must maintain adequate eating behavior for several reasons. Firstly, nurses play a pivotal role in promoting health, and

their own well-being sets an example for others. Secondly, proper nutrition contributes to sustained energy levels, enhancing nurses' ability to meet the physical and mental demands of their profession. Additionally, adequate eating behavior is linked to overall health, reducing the risk of chronic diseases that could affect job performance. Lastly, nurses with healthy eating habits are better equipped to educate and guide patients on lifestyle choices, creating a positive impact on public health

Understanding the intricate relationship between nurses' eating behaviour and managing their own health is crucial for fostering a holistic approach to healthcare, therefore our study aimed to evaluate eating behaviour and demographic factors influencing chronic disease prevalence in a sample of community nurses from Romania.

Between October–November 2023, 1920 community nurses were invited to answer an online survey, using an advertisement in their professional network. Of them, 788 responded. In the survey, which included a semi-quantitative food frequency questionnaire with 53 food items, the Intuitive Eating Survey 2 (IES-2), and demographic items were used. This questionnaire was designed to gather information about food intake over the course of last 30 days. For each item, we investigated the frequency and usual amount of consumption. Additional questions regarding specific items were asked to estimate the quantity of fat or added sugar. We converted the intakes to grams using household scale guidelines and calculated energy and macronutrient intakes for each individual using a specialized computer program. Finally, we transformed macronutrient intakes into a percentage of contribution to the total energy consumed.

A multivariate model was built for the prediction of the association between eating behavior and other factors associated with chronic diseases. The majority of participants were females (95.1%), with the largest age group falling between 40 and 49.9 years (48.2%). Regarding the EFSA criteria for adequate carbohydrate and fat intake, 20.2% of the group have a high intake of carbohydrates, respectively, 43.4% of the group have a high intake of fat. Analysis of chronic diseases indicated that 24.9% of individuals reported at least one diagnosis by a physician. The presence of chronic disease was associated with a low level of perceived health status, with an OR = 3.388, 95%CI (1.684–6.814), compared to those reporting excellent or very good perceived health status. High stress had an OR = 1.483, 95%CI (1.033–2.129). BMI had an OR = 1.069, 95%CI (1.032–1.108), while low carbohydrate diet score had an OR = 0.956, 95%CI (0.920–0.992). Gender and IES-2 did not significantly contribute to the model, but their effect was controlled.

To the best of our knowledge, this is the first study to shed light on the eating habits concerning chronic diseases in community nurses from Romania. This assessment is

important for future programs targeting improvement in eating habits in community nurses, with personal benefits, but also for their patients' benefits, because being able to provide nutrition screening and appropriate nutrition advice is essential to improve healthy eating and subsequent health outcomes of their patients.

According to our study, the predictive model for the association between demographics and eating habits with chronic disease diagnosis found that individuals diagnosed with chronic disease tend to experience higher levels of stress, perceive their health status as poor, have an older age and higher BMI, and consume more carbohydrates compared to those without a diagnosis.

Our research has shown that community nurses with more age and experience tend to have higher intuitive scores. This may be attributed to the fact that as individuals age, they develop a greater understanding of their body and personal preferences, leading to a more refined and knowledgeable approach to intuitive eating. The principles of intuitive eating are in line with mindfulness techniques, which encourage individuals to be fully present and aware of their eating habits.

Our study has certain limitations, such as self-reporting bias and selection bias. The self-reported bias pertains not only to the nutrition evaluation, but also to the reported diagnosis of chronic disease. It is important to note that the design of this study was cross sectional, which means that no causality can be established between eating patterns and the presence of chronic diseases. Instead, the findings can only be interpreted as associations.

Lastly, collaborative efforts between healthcare institutions, policymakers, and researchers are essential to develop and implement tailored programs that prioritize the health and resilience of nurses, thereby contributing to the mitigation of the burden of chronic diseases within healthcare systems. The implementation of such programs would require a concerted effort and the development of strategies that are feasible, sustainable, and evidence-based. Overall, these research endeavors can contribute significantly to the advancement of knowledge in this field and ultimately lead to positive health outcomes for healthcare professionals and patients alike.

This study provides a comprehensive insight into the eating habits and health profiles of community nurses in Romania, uncovering noteworthy associations between dietary patterns, stress levels, and chronic disease diagnoses. The prevalence of chronic diseases among this population, particularly hypertension, underscores the importance of targeted interventions for healthcare professionals. Notably, the study identifies modifiable factors, including stress, BMI, and dietary habits based on high carbohydrate diets, offering potential avenues for personalized health interventions. The findings emphasize the need for holistic

strategies, encompassing stress reduction, healthy weight management, and nutritional education to enhance overall well-being. As healthcare systems struggle with the increasing burden of chronic diseases, these insights contribute to the development of tailored programs that prioritize the health and resilience of healthcare providers.

STUDY 3. COMMUNITY NURSES AND CASE MANAGEMENT FOR RARE DISEASES IN ROMANIA

The study started from the analysis of the national legislation regarding the approach to the problem of rare and genetic diseases in order to achieve case management by the community medical assistants, integrated with the specialists in family medicine, from the centers of expertise in rare diseases, from the centers of medical genetics and others specialist doctors from different pathologies encountered in the approach to the complex medico-social problem of the patient with a rare disease. Prior to the introduction of the rare genetic disease indicators in the specific legislation, the community nurses followed other indicators for all population categories regarding the intervention in the case of different pathologies or different medical and social problems. Community nurses report activity in the amcmsr.gov.ro application starting with 2015 in a pilot phase, and from august 2016 it also received the subdomain registered on REGISTRU.gov.ro being hosted on the STS platform, I am being the administrator of the application from the moment of operation it and until now.

Romania does not have a national registry dedicated to rare diseases. We consider that the establishment of a comprehensive rare disease monitoring system is crucial in facilitating evidence-based health policies and this study aimed to examine the existing landscape of rare diseases in Romania and to identify the challenges associated with the development of a national platform for the coordination of the care of patients with rare diseases.

Through this study, the first and only one of its kind carried out in Romania, we wanted to argue the need to build a pilot system for the management of rare diseases and to conceptualise the creation of a rare diseases ecosystem in Romania that can be used by other health systems.

The amcmsr.gov.ro application, in its initial form, allowed the analysis of cases of rare genetic diseases introduced by CN and their repertory by sex, age, ethnicity, distribution by county of residence, data that demonstrate both the existence of patients with genetic and rare diseases, as well as the involvement of community nurses in providing the necessary medico-social services.

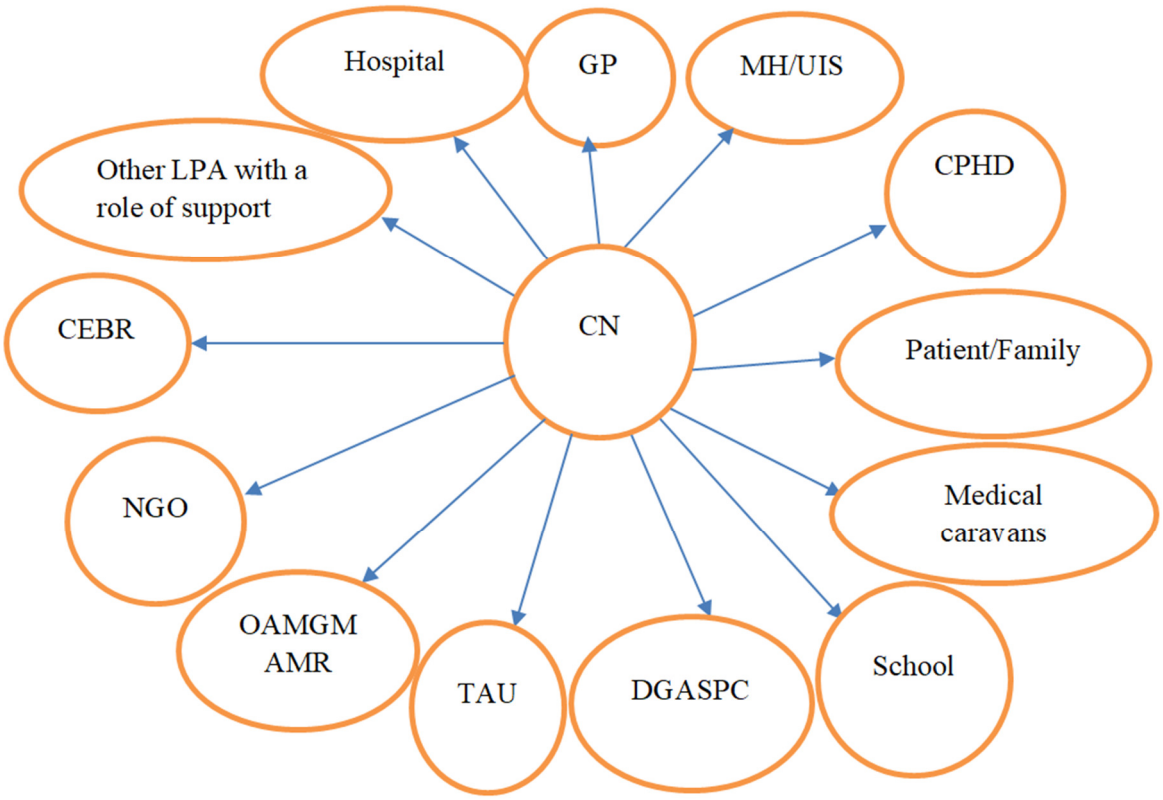
The limited access to information and services for patients with rare diseases, as well as the shortage of specialist medical service providers, led us to include the approach to patients with rare/genetic diseases in the case management system within the ongoing regulatory acts. In order to evaluate the results of the implementation of the community nursing activity, UNICEF carried out a research study for CN from all over the country between June and September 2020. The study, entitled "Community nurses' opinion regarding working conditions, professional activity and training needs", was carried out in collaboration with the Center for Health Policies and Services and with the support of the Ministry of Health, I am being the representative of the Ministry of Health.

I mention that from the multitude of needs identified, I also noticed the CN's request to benefit from special training in the field of rare genetic diseases. Another study in which we collaborated analysed the Romanian experience and the role of political factors in shaping the reform of the health system in Romania. The study concerned primary medical care, the evaluation of medical technologies and the digitization process of the reporting and data collection system.

The Romanian Ministry of Health recognizes the importance of an integrated care approach and significant progress has already been made in organising the activities of community nurses and in primary care. Community healthcare is considered a fundamental component of primary health care, with specific objectives outlined in legislation, such as article 5 of OUG 18/2017. For the effective implementation of case management and the definition of the role of community nurses, several legal and organisational measures were needed. In 2017, OUG 18/2017 was approved, replacing OUG 162/2008.

For the first time, the intervention of community nurses is recognized in the context of rare diseases. The identification of beneficiaries of community medical assistance, in collaboration with the public social assistance service, aims to address medical and social challenges in the community, especially among vulnerable groups. Through this normative act, the Ministry of Health aims to facilitate access to health services and social services for the population, especially for those belonging to vulnerable groups. In addition, it emphasises the importance of promoting attitudes and behaviours that support a healthy lifestyle, including health education initiatives within communities [103]. Participation in the implementation of public health programs, projects, actions and interventions should be adapted to the needs of the community, especially those belonging to vulnerable groups. It is essential that the provision of health services is carried out by health personnel with legally recognized professional competences and with the assignments established for those who provide community medical assistance services.

The figure below briefly presents the relational picture of the community nurse's work with beneficiaries in the process of identification, intervention and monitoring:



Legend

CEBR - Center for Expertise in Rare Diseases
CN – Community Nurse
CPHD - County Public Health Directorate
DGASPC- General Directorate of Social Assistance and Child Protection
GP- General Practitioner
LPA - Local Public Authority
MH- Ministry Health
NGO - non-governmental organization
OAMGMAMR- The Order of General Medical Assistants, Midwives and Medical Assistants from Romania
TAU- Territorial Administrative Unit
UIS - Social Inclusion Unit

FINAL CONCLUSIONS AND PERSONAL CONTRIBUTIONS

The concept of integrated care has become increasingly used lately due to the need to address the individual and his problems to identify the most reliable solutions. Thus, the integrated approach to the medical-social issues of the individual and communities is a concept in full ascension.

Through the methods used in the first two research studies, the degree of empathy of the community nurse in relation to beneficiaries and his health status in relation to his eating behaviour were analysed, starting from the high degree of overload by interacting with beneficiaries who have complex medical-social problems, with emphasis on interaction with the patient in general, including the one with the rare disease.

Analysis of empathy in a large sample of community nurses in Romania showed good levels of empathy, as nearly three-quarters of them achieved high levels of empathy.

Future training programs at the beginning of the career aimed at increasing emotional competences, would lead to reducing stress levels and encouraging staff activity, which could promote the increase of the quality of community healthcare in Romania.

The eating behaviour analysis provides a detailed examination of the eating habits and health profiles of community health nurses in Romania, uncovering significant links between eating patterns, stress levels and chronic disease diagnosis.

The findings underscore the need for holistic strategies that include stress reduction, healthy weight management, and nutrition education to improve overall well-being.

Analysis of the complex interplay between nutrition, lifestyle and health outcomes in the community health care cohort provides valuable insights for the design of targeted interventions and personalized support programs. These programs aim to improve the well-being of community nurses and, consequently, the patients they serve through integrated health care.

The amcmsr.gov.ro application for making the case management report of the rare disease patient is very useful to carry out the effective monitoring of the rare and genetic disease patient. The application is the tool for reporting the activity of the staff in the community health system and creates a database with the samples identified and monitored from the level of each territorial administrative unit that carries out community health activity.

Analyses of the impact of piloting CN training in rare disease case management demonstrated that people living with a rare disease and caregivers who received case management services improved their level of information about the disease and their rights, their knowledge of services available and the ability to self-manage.

A good monitoring and intervention in the case of patients with rare diseases, the integration of community support networks, collaboration with centers of expertise will greatly contribute to increasing the quality of medical and social services and will allow:

1. Improved access to healthcare
2. Improved care coordination
3. Personalised support and education
4. Advocacy and empowerment.
5. Continuity of care.
6. Psychosocial support.

CNs work closely with multidisciplinary teams and collaborate with health care providers, social workers, educators, and community leaders to address the unique health care needs of the populations they serve. They play a crucial role in promoting health equity, preventing disease and improving access to healthcare services. The use of case management for patients with rare diseases aims to regulate multidisciplinary and inter-institutional interventions, in an organised, standardised, coherent and efficient manner in implementation for the patient, family/legal representative and other important persons for the patient.