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NURSING PERSPECTIVES ON INHERITED METABOLIC DISORDERS: EXPERIENCE AND EDUCATIONAL NEEDS IN AN ITALIAN HOSPITAL SETTING

S. GIGANTE, K. POZZI, V.B. SISTI, A. CERIZZA, R. PRETESE, V. CRESCITELLI

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Department of Pediatrics, Fondazione IRCCS San Gerardo dei Tintori, Monza, Italy

Corresponding Author
Silvia Gigante, MD; e-mail: silvia.gigante@irccs-sangerardo.it

***ABSTRACT* – Objective:** The aim of this study was to assess the clinical experience and training needs of nursing staff involved in the care of patients with inherited metabolic disorders (IMDs), a group of rare diseases that require specialized, multidisciplinary management and are currently underrepresented in nursing education.

Subjects and Methods: A cross-sectional, single-center survey was conducted at Fondazione IRCCS San Gerardo dei Tintori (Monza, Italy). An 18-item online questionnaire was distributed to 500 nurses across 23 hospital units. The survey addressed demographics, clinical experience, and educational exposure related to IMD care. Descriptive statistics were used to analyze the responses.

Results: A total of 104 nurses (21%) completed the survey. Among them, 87% had assisted at least one IMD patient, and 82% perceived the nursing care as highly complex. While routine procedures such as blood glucose monitoring and enteral nutrition were commonly performed, IMD-specific interventions (e.g., intravenous arginine administration or enzyme replacement therapy) were rarely carried out. Emotional responses to IMD care included uncertainty (53%), concern (50%), and a sense of inadequacy (46%). Formal training was limited: 54% had not received IMD-related education during their studies, and 48% had never attended relevant courses. Notably, 92% expressed a desire for further training in this area.

Conclusions: Despite frequent clinical exposure to IMD patients, most nurses reported limited formal training and low procedural confidence. These findings underscore the need for structured, IMD-focused educational initiatives to enhance nursing competence and support high-quality, multidisciplinary care for rare disease populations.

Keywords: Nursing, Inherited metabolic disorders, Training, Expertise, Education.

Abbreviations: IMD: inherited metabolic disorder; EN: enteral nutrition.

INTRODUCTION

Inherited metabolic disorders (IMDs) account for approximately 15% of rare diseases, with an overall incidence ranging from 1 in 500 to 1 in 4,000 live births[1]. Inherited metabolic disorders encompass a heterogeneous group of conditions that impair metabolic pathways responsible for the breakdown or storage of carbohydrates, fatty acids, and proteins[2].

The implementation of Newborn Screening Programs has enabled early diagnosis for many IMDs, reducing the risk of irreversible damage and allowing prompt intervention[3-6]. Dietary management remains central to IMD treatment, involving scheduled meals to prevent fasting and nutritional supplementation to avoid deficiencies[7].

The use of food for special medical purposes is often lifelong and essential in many IMDs to ensure adequate caloric intake, support growth, maintain satiety, and provide dietary variety. In addition to early diagnosis, advances in genetics and biochemistry have enhanced clinicians' understanding of the pathophysiology and treatment of IMDs, thereby increasing the life expectancy of affected individuals[8,9].

Given the growing attention to the quality of life of patients with IMDs and their families, alongside the complexity of these conditions, appropriate clinical management by a multidisciplinary team, including medical doctors, biochemists, nurses, and other healthcare professionals, is crucial to ensure comprehensive care[10-12].

Nurses play a vital role within the therapeutic team, assisting physicians in the diagnostic and treatment process, as well as providing direct patient care, including nursing procedures, medication administration, and injections[13].

As nurses are typically the first healthcare professionals to interact with patients and often spend extended periods with them, patients tend to expect not only medical knowledge but also safety, trust, and effective communication from them[13].

To foster awareness and build specific expertise in IMDs among all professionals involved in their complex management, including nurses, educational programs, and training initiatives have been developed within the European Reference Network[14].

However, the role of nurses in IMD care remains poorly defined, and the extent of their training and expertise in the specific management of IMD patients has not been thoroughly examined, particularly in the Italian context.

This survey aims to investigate, within a single-center cohort, the role of nurses in the management of IMDs, assess their level of expertise and training, and identify emerging needs to be addressed through targeted future strategies.

SUBJECTS AND METHODS

An online survey comprising 18 single- or multiple-choice questions (Supplementary Material) was developed by an expert panel composed of nurses from the pediatric day hospital and pediatric hospitalization departments, pediatric metabolist doctors and dietitians of the simple operating unit of inherited metabolic diseases. These professionals provide care for both pediatric and adult patients with IMDs. The survey was designed with Microsoft Forms, which enabled us to share it directly *via* institutional Outlook email addresses. The survey was distributed electronically to nurses from 23 Operating Units of Fondazione IRCCS San Gerardo dei Tintori between April and May 2024 *via* their respective clinical center contacts, using QR codes or direct links.

The non-validated questionnaire, developed in Italian, was structured into three sections: personal information, clinical experience, and training needs.

Ethical approval was not required, as the survey did not constitute a clinical study and did not involve clinical outcomes or patient-specific data, but was instead conducted among healthcare professionals.

Statistical Analysis

Survey responses were analyzed using descriptive statistics, with results reported as percentages (%) and absolute numbers (n). As the survey included multiple-choice questions, participants could select more than one response; in such cases, total percentages may exceed 100%.

RESULTS

Personal Information

Of the 500 nurses who received the survey, 21% (104/500) responded. Among respondents, 90% (94/104) were female.

Regarding age distribution, 43% (45/104) were under 36 years old, 27% (28/104) were aged 36-50 years, and 30% (31/104) were over 50 years.

Most participants (88/104, 85%) identified as general nurses, while 15% (16/104) reported working as pediatric nurses. With respect to the highest educational qualification, 72% (75/104) held a diploma or Bachelor's degree, 26% (27/104) had a Master's or first-level degree, and 2% (2/104) reported holding a second-level Master's degree.

The majority of respondents, about 51% (53/104), work in pediatric areas: Pediatric Unit (16/104, 15.3% of the total respondents), Pediatric Hematology Unit (15/104, 14.4%), Neonatal Intensive Care

(8/104, 7.6%), Pedatric Day Hospital (4/104, 3.8%), Pediatric Ematology Day Hospital (7/104, 6.7%) and Neonatal Clinic (3/104, 2.8%). 20% of nurses belong to critical care areas: 9.7% (10/104) in the emergency room, 4.9% (5/104) in Operating Rooms and 5.8% (6/104) in intensive care (general and neuro-intensive care). Additionally, 7.8% (8/104) were based in diagnostic departments (endoscopy and radiology).

Responses were also received from nephrology (5/104), emergency medicine (10/104), psychiatry (3/104), otorhinolaryngology (2/104), orthopedics (1/104), general medicine (5/104), and the day hospital of medicine (4/104). No responses were received from departments of the emergency medicine swab department (activated following the COVID-19 emergency), child neuropsychiatry, neurosurgery and the general critical care area.

Regarding professional experience, 14% (15/104) of respondents reported less than 5 years of total and departmental experience. A total of 24% (25/104) had 5-10 years of general experience, with 10 of these (10/25) having worked in their current department for fewer than 5 years. Among 26% (27/104) of respondents, general experience ranged from 10 to 25 years; of these, fourteen had fewer than 5 years, four had 5-9 years, and nine had 10-25 years of experience in their current department.

Finally, 36% (37/104) reported more than 25 years of overall experience. Among them, eight had spent the entire period in the same department, 14 had worked 10-25 years, eight for 5-9 years, and seven for less than 5 years in their current department.

Clinical Experience

Of all respondents, 25% (26/104) reported having cared for pediatric patients (<18 years), 20 participants reported having assisted only adult patients, and 58 respondents (56%) indicated experience with both pediatric and adult patients during their working experience in their current Unit.

When asked how frequently patients with IMDs are admitted to their unit, 12% (12/104) of participants reported daily admissions, 21% (22/104) reported approximately once a week, 24% (25/104) nearly once a month, and 25% (26/104) about once a year. Notably, 18% (19/104) stated they had never encountered IMD patients in their unit (Figure 1).

Participants were then asked to report the frequency with which they performed specific clinical tasks and activities during their professional experience with both general and IMD cases (Figure 2).

Central venous access management and capillary blood sugar control (digital therapeutics, DTX) monitoring were frequently performed by 77.9% (81/104) and 73.1% (76/104) of respondents, respectively. A majority also reported frequent administration of enteral nutrition (EN) (57.7%, 60/104) and nasogastric tube placement (56.7%, 59/104). In contrast, preparing EN mixtures was less commonly performed, with 47.1% (49/104) reporting frequent involvement.

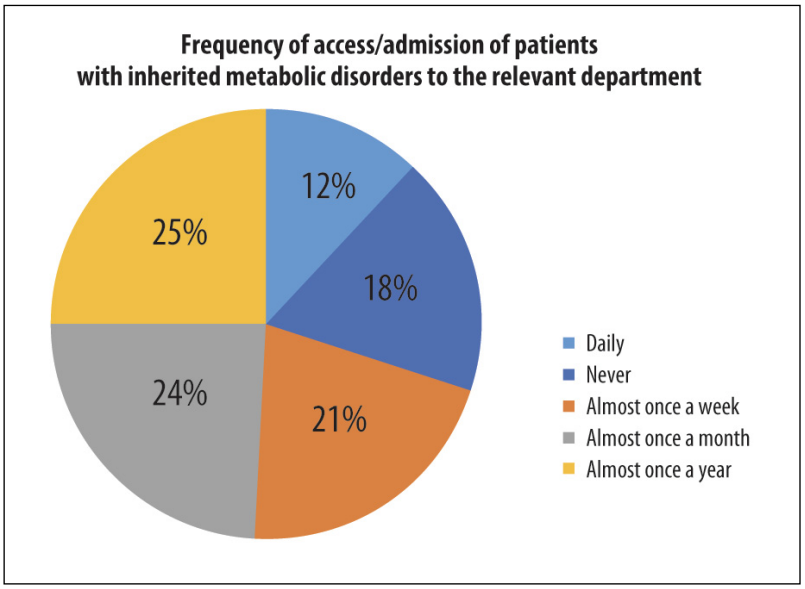

Figure 1. Frequency of access/admission of patients with inherited metabolic disorders to the relevant department.

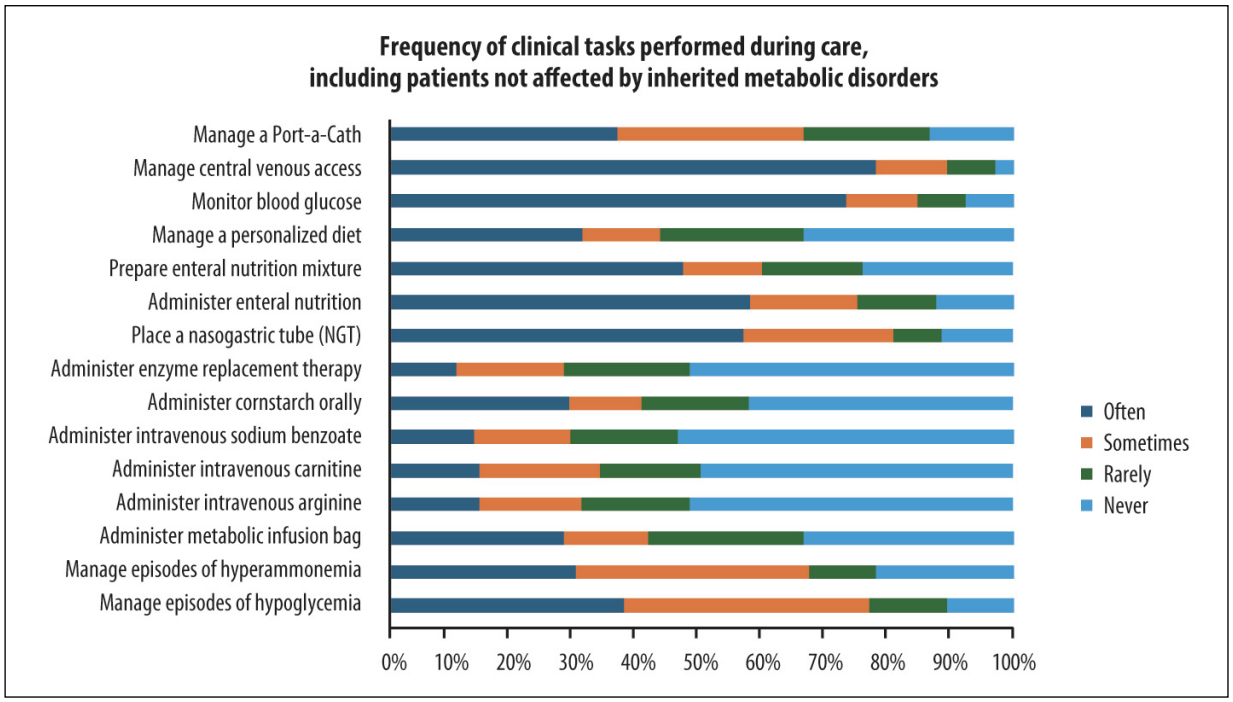

Figure 2. Frequency of clinical tasks performed during care, including patients not affected by inherited metabolic disorders.

30.8% of nurses reported having managed a diet *ad personam* (i.e., checking the delivery of the meal, the correspondence with the prescription, the time of consumption, monitoring the amount of food consumed, and managing potential substitutions), while certain IMD-specific interventions were less common. Notably, 42.3% (44/104) had never administered oral cornstarch. Specialized procedures such as intravenous administration of arginine (51.9%, 54/104), carnitine (50.0%, 52/104), sodium benzoate (53.8%, 56/104), and enzyme replacement therapy (51.9%, 54/104) were also reported as never performed by more than half of the respondents. Similarly, 33.7% (35/104) had never administered metabolic infusion bags, and 24.0% (25/104) had never prepared EN mixtures.

Conversely, episodes of hyperammonemia and hypoglycemia were more frequently managed, with 29.8% (31/104) and 37.5% (39/104) of participants, respectively, indicating frequent involvement.

Considering the subgroup of the Pediatric Area, specific interventions and therapies in management of IMD patients are performed more frequently: for istance, the management of *ad personam* diet – which consists of checking that the meal corresponds to the dietician's prescription, that the correct quantity is consumed, monitoring compliance with the timetables, and administering any substitute foods – happens "often" in 25/53 cases and "sometimes" in 12/53.

The same occurs with the administration of some specific therapies, such as Enzyme Replacement Therapy (ERT), reported "often" and "sometimes" by 41% of nurses working in the Pediatric Area, while in the remaining areas it takes place "often" in only 4 cases (all belonging to Adult Day Hosptal) and "sometimes" in three cases, for a total of 23% of the 30 nurses who do not work in the pediatric area and who have assisted a patient with IMD at least once.

A total of 90 nurses (87%) reported having cared for at least one patient with an IMD during their professional experience. Among these respondents (n=90), 82% rated the complexity of nursing care for IMD patients as "high," while 18% perceived it as "medium." Notably, none of the participants reported the level of care complexity as "low."

In addition, when assisting patients with IMDs, respondents reported experiencing a range of emotional responses (Figure 3), including uncertainty (48/90), concern (45/90), a sense of inadequacy (41/90), anxiety (20/90), personal gratification (17/90), and confidence (6/90).

When assisting patients with IMDs in non-urgent situations that are complex to manage independently, participants reported primarily seeking support from colleagues (76%, 71/104) or nurses from other units (46%, 41/104). Additionally, 59% (54/104) indicated they may consult a physician from their department, while 49% (45/104) reported contacting the designated metabolist within their center.

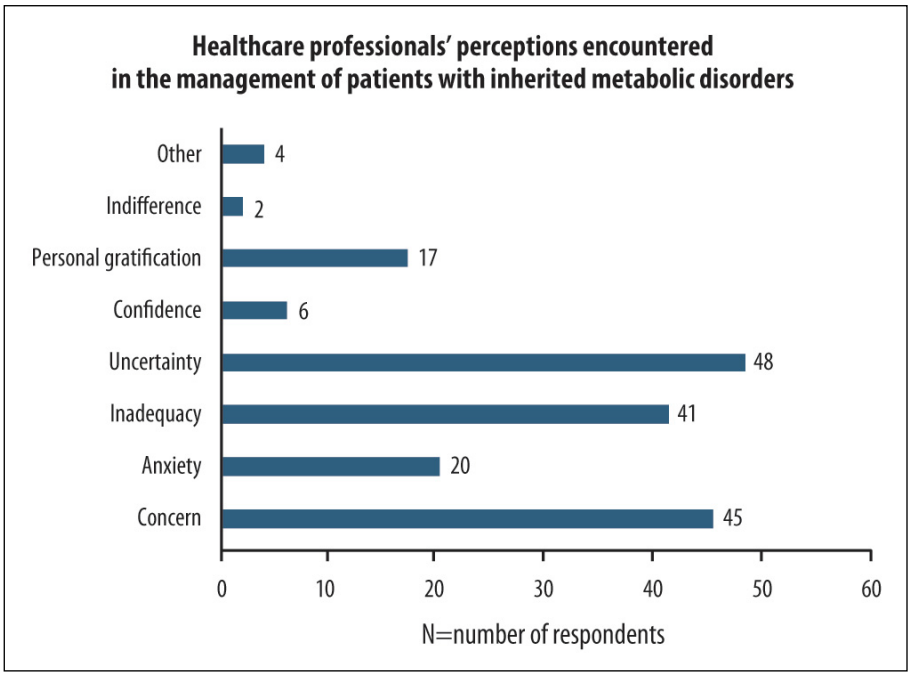

Figure 3. Healthcare professionals' perceptions encountered in the management of patients with inherited metabolic disorders.

Approximately half of the respondents (47/104) reported referring to specific protocols, and 19 participants (18%, 19/104) indicated consulting online resources such as medical books.

Formation and Training Needs

In 54% of cases (56/104), respondents reported that IMD topics were not included in their formal education. Additionally, 48% (51/104) stated they had never attended a training course or conference on IMDs.

A large majority (92%, 96/104) of nurses expressed a need for more information on IMDs and reported a desire to receive further training on the topic.

DISCUSSION

This single-center survey assessed the clinical experience and educational needs of nurses caring for patients with inherited metabolic diseases (IMDs), a growing yet underrepresented field in nursing education.

Despite follow-up reminders, the response rate was low (21%; 104/500), potentially reflecting limited awareness or interest in IMDs among nurses. Nonetheless, 87% of total respondents reported having managed at least one IMD patient in their career, with particularly high exposure among pediatric nurses (among the 53 nurses belonging to the pediatric area, 52 gave an affirmative response).

A large majority (82%) perceived IMD-related nursing care as highly complex, indicating the clinical challenges these patients pose. Consistent with this, nurses frequently performed procedures applicable to other categories of patients, such as blood glucose monitoring, enteral feeding, and central venous access management, while IMD-specific interventions (e.g., intravenous administration of arginine, sodium benzoate, or enzyme replacement therapy) were less common. This likely reflects the rarity and specialized nature of such treatments.

Pediatric departments demonstrated greater experience with IMD-specific procedures due to the higher admission rates of affected patients in these units. The example of ERT administration is significant: while in non-pediatric settings this mostly occurs in outpatient care, the administration of ERT often takes place in both pediatric departments and pediatric hematology departments, as patients with lysosomal storage diseases undergoing transplantation (Mucopolysaccharidosis patients) can be hospitalized in these settings.

Emotional responses such as uncertainty, concern, and a sense of inadequacy were commonly reported. These findings highlight the need for structured training and standardized protocols to improve preparation and professional confidence among nurses.

Significant educational gaps emerged: 54% of respondents had not received IMD-related instruction during their undergraduate education, and 48% had never attended any relevant course or conference. Notably, among the youngest age group (20-35 years), comprising 43% of respondents, only 29% had addressed IMDs during their degree program. In contrast, 92% expressed a clear need for further training in this field.

This discrepancy between the perceived complexity of IMD care and the limited basic nursing education on this subject underscores the urgency of targeted educational interventions. As supported by the literature[13,14], continuing professional development enhances diagnostic confidence, procedural competence, and communication skills in rare disease care. Nurses play a central role within multidisciplinary teams, providing both routine and emergency care for IMD patients, and directly influencing their quality of life[12].

Adequate training would allow nurses to effectively respond to patients' clinical, therapeutic and psychological needs, significantly impacting their quality of life, and would enable them to provide comprehensive health education to patients and caregivers. Informed patients and caregivers, educated in therapy administration, device use, and emergency management, can reduce hospitalizations and emergency visits. This is especially important given the high treatment burden that already disrupts daily life. Moreover, comprehensive nursing care supports daily functioning, ensures adherence to therapy and diet, manages pain, and helps prevent hypoglycemia.

A key limitation of this study is the reliance on subjective perceptions to assess the complexity of nursing care in the absence of standardized indicators, which may compromise the objectivity of the findings[15]. Additionally, being a single-center study with limited departmental participation and a predominance of pediatric responses, the generalizability of findings is restricted.

Future multicenter studies involving diverse clinical settings are needed to confirm these results and support reforms in undergraduate and continuing nursing education. Dedicated training should focus on nursing roles in metabolic emergencies, especially the preparation and administration of frequently used therapies.

Following the revision of the survey data, a SWOT analysis was conducted (Figure 4). The strengths identified include the extensive experience nurses have in the care of IMD patients working in pediatric settings. This expertise could be shared with colleagues from other departments through residency courses and the development of protocols and quality documents.

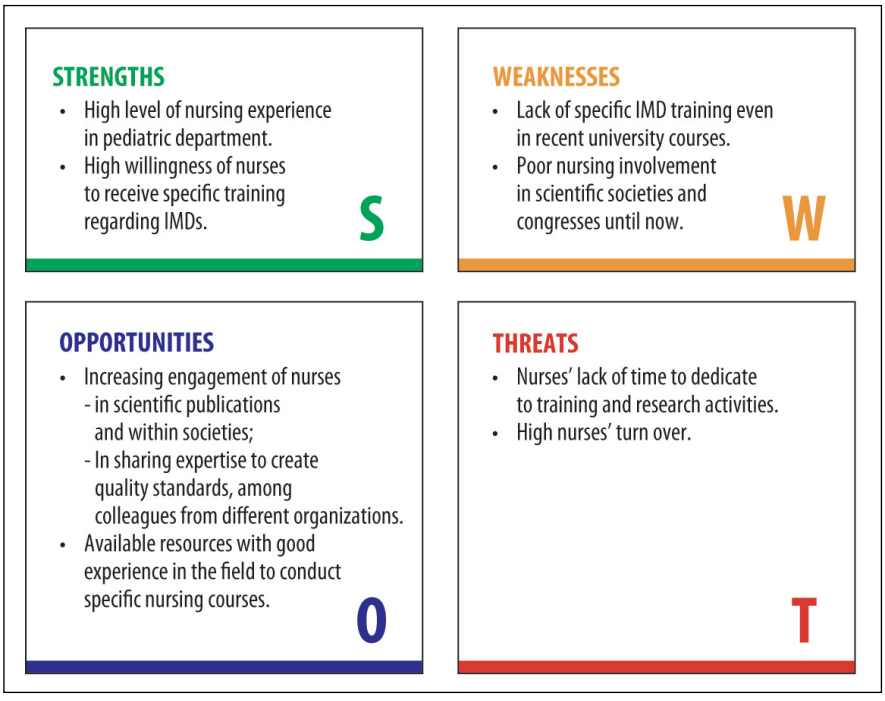

Figure 4. SWOT analysis, based on survey findings.

The lack of specific IMD training, especially in recent University curricula, as highlighted by the survey, is a significant weakness, along with the limited involvement of nurses in scientific societies and congresses up to now.

The increasing engagement of nurses in this field and scientific publications represents a valuable opportunity for enhancing nursing expertise and management of IMDs and eventually improving the quality of nursing care.

This knowledge can also be shared and expanded at both national and international levels. To achieve this, it is essential to provide experienced IMD nurses with opportunities to engage in scientific and research activities, allowing them to dedicate adequate time while balancing these commitments with their clinical duties.

Reducing staff turnover is also of fundamental importance, as it enables the development of specialized skills within specific areas, thereby improving the quality of nursing care provided.

Following this analysis shared with the working team, a dedicated residential course on IMDs is already active at Fondazione IRCCS San Gerardo dei Tintori. This course has been implemented and integrated with a nursing section focused particularly on the management of metabolic emergencies, as well as the preparation and administration of specific therapies in such cases.

Our institution, Fondazione IRCCS San Gerardo dei Tintori, has revised protocols and manuals and is developing a comprehensive handbook for managing metabolic emergencies. In parallel, the University of Milano-Bicocca has launched a Master's program in Intensive Care Nursing Sciences that includes a specific module on IMD emergency management. The nursing component addresses therapy preparation and administration and includes case-based learning.

Finally, establishing a Nursing Work Group within the Italian Society for the Study of Inherited Metabolic Diseases and Neonatal Screening (SIMMESN) could foster the exchange of expertise among national reference centers and promote clinical standardization. The present survey could serve as a foundation for a broader, multicenter investigation across Italy.

CONCLUSIONS

This single-center survey reveals substantial gaps in the training and expertise of nurses involved in the care of patients with IMDs. Despite frequent clinical encounters and a high perceived complexity of care, most respondents reported limited formal education and hands-on experience in IMD management.

These findings underscore the urgent need to better define the role of nurses in IMD care and to implement targeted training programs that address the unique, specific challenges associated with managing rare metabolic conditions.

Broader, multicenter studies are warranted to validate these results and to guide the integration of IMD-specific content into nursing curricula and continuing professional development pathways.

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Study conception and design: all; collection and interpretation of data: S. Gigante, K. Pozzi, V. Sisti, A. Cerizza; statistical analysis: S. Gigante, K. Pozzi, V. Sisti, A. Cerizza; manuscript drafting: all; manuscript editing: all; approval to submit: all.

Availability of Data and Material:
The datasets generated during and/or analyzed during the current study are available from the corresponding author upon reasonable request.

Conflicts of Interest:
The authors declare that they have no conflicts of interest to disclose.

Ethics Approval:

Ethical approval was not required, as this survey was not classified as a clinical study, according to the most recent national regulatory framework outlined in Regulation No. 425/2024. Specifically, the survey did not involve clinical outcomes or patient-specific data but was exclusively conducted among healthcare professionals, aligning with the definition established in the current legislative reference.

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Informed Consent:

Participants were clearly informed about the study's purpose, their voluntary involvement, and data confidentiality. Consent is considered implicit, as participation was entirely voluntary and free from coercion. Responses were anonymous or de-identified, and the study did not involve interventions or pose any risk to participants.

ORCID ID:

Roberta Pretese: 0009-0006-6713-8030
Katia Pozzi: 0009-0000-7244-7272
Silvia Gigante: 0009-0007-4035-9017
Alessia Cerizza: 0009-0007-7902-7968

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