

# ACCESS TO LIFE-SAVING FOODS FOR SPECIAL MEDICAL PURPOSES FOR PATIENTS WITH INHERITED METABOLIC DISORDERS: A NATIONAL SURVEY OF ITALIAN DIETITIANS

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**ABSTRACT – Objective:** The aim of this study was to investigate access to Food for Special Medical Purposes (FSMPs) for patients with Inherited Metabolic Disorders (IMDs) across Italian regions, with a focus on the role of dietitians and the challenges faced in prescription and supply processes.

**Subjects and Methods:** An online survey of 32 multiple-choice questions was distributed to 38 dietitians affiliated with the Italian Society for the Study of Hereditary Metabolic Diseases and Newborn Screening (SIMMESN). Responses were analyzed using descriptive statistics.

**Results:** 29 dietitians across 12 regions responded to the survey. Only 48.3% of respondents believe that a standardized regional procedure for FSMP distribution exists, while 51.7% cited district-level variations. Most respondents (86.2%) reported that patients sometimes need to purchase FSMPs independently. Prescription methods varied, with both paper and electronic systems in use. Dietitians played a central role in FSMP selection but had limited authority in administering prescription plans. Bureaucratic delays were common, with over 34% of respondents reporting delivery times exceeding two weeks. Budget limitations and procurement issues further restricted access.

**Conclusions:** Despite national coverage under essential care levels, disparities in FSMP access persist due to regional policy fragmentation. Enhancing national harmonization, formalizing the role of dietitians, and improving administrative efficiency are critical to ensuring equitable and timely dietary support for IMD patients in Italy.

**KEYWORDS:** Food for special medical purposes, Inherited metabolic disorders, Italian National Health Service, Dietitian.

**ABBREVIATIONS:** ASL/USL/ASUR/AST: Regional/local health units in Italy (terminology varies by region); ESTAR: Ente di Supporto Tecnico Amministrativo della Sanità Toscana (Technical Administrative Support Entity of Tuscany Healthcare); FSMP: Food for Special Medical Purposes; GP: General Practitioner; IMDs: Inherited Metabolic Disorders; SIMMESN: Italian Society for the Study of Hereditary Metabolic Diseases and Newborn Screening (Società Italiana per lo Studio delle Malattie Metaboliche Ereditarie e lo Screening Neonatale).

## INTRODUCTION

### The role of Food for Special Medical Purposes in Inherited Metabolic Disorders

The management of inherited metabolic disorders (IMDs), including certain disorders of protein, lipid, and carbohydrate metabolism, necessitates specialized dietary interventions that are critical for preventing potentially irreversible damage<sup>1,2</sup>.

In certain conditions, such as aminoacidopathies, urea cycle defects, fatty acid oxidation defects and glycogen storage diseases, specific dietary products – such as protein-free foods, protein substitutes, modular formulations (e.g., glucolipid mixtures), medium-chain triglyceride (MCT)-based formulas and ketogenic diet products – are required<sup>3</sup>.

In individuals with severe phenylketonuria who have a natural protein tolerance of less than 10 g/day, these dietary products may contribute up to 50% of total energy intake<sup>4,5</sup>.

Unlike other conditions, the use of FSMPs in many IMDs is lifelong and essential to ensure adequate caloric intake for supporting growth, maintaining satiety, and providing dietary variety. These products help mimic certain aspects of a normal diet and improve long-term adherence<sup>6</sup>. Both factors are fundamental in preventing catabolism and ensuring optimal metabolic control<sup>1,2</sup>.

### Italian Regulatory and Distribution Framework

In Italy, the use of dietary products, defined as Foods for Special Medical Purposes (FSMPs) according to Ministry of Health guidelines<sup>7</sup>, has been included in the Essential Levels of Care, as established by the Prime Minister's Decree of January 12, 2017<sup>8</sup>. Article 14 states that individuals affected by congenital metabolic diseases are entitled to receive FSMPs through the National Health Service, with distribution based on the National Special Medical Foods Registry<sup>8,9</sup>.

Established under Article 7 of the Ministerial Decree of June 8, 2001, and amended by the Ministerial Decree of May 17, 2016, this registry is organized into three sections: foods for special medical purposes (FSMPs), gluten-free foods, and infant formulas. Each section is further categorized by product and company. It lists FSMPs that are reimbursable by the National Health Service for patients with specific medical conditions, including IMDs. Annex 7 of the January 12, 2017, decree provides the list of rare diseases exempt from cost-sharing.

Nevertheless, despite growing awareness of the importance of FSMPs in dietary management for specific IMDs, patient access to these products may be limited by disparities in reimbursement and availability<sup>10</sup>. Furthermore, product reimbursement is recognized as a critical factor influencing dietary adherence, including the use of FSMPs, and is, therefore, essential in preventing potential clinical complications<sup>6,11</sup>.

In Italy, the responsibility for defining the distribution pathway of FSMPs is delegated to individual regions. This results in significant variability in the distribution process, which may involve various stakeholders, including prescribing clinical centers, regional coordination centers for rare diseases, local health districts, hospital pharmacies, private territorial pharmacies, general practitioners or pediatricians, patients, and FSMP manufacturers.

Despite this complexity, patient access to FSMPs for IMDs has not been comprehensively analyzed in the Italian context.

This study aims to investigate patient access to FSMPs for IMDs across different Italian regions, focusing on the role of dietitians in the distribution process, assessing the difficulties reported by patients, and identifying key challenges related to FSMP access and supply.

## SUBJECTS AND METHODS

An online survey consisting of 32 multiple-choice questions (see [Supplementary Material](#)) was developed by an expert panel of six dietitians, all members of the Nutrition and Dietetic Working Group of the Italian Society for the Study of Hereditary Metabolic Diseases and Newborn Screening (SIMMESN). These professionals work with both pediatric and adult patients across various centers in Italy. The survey was distributed online to members of this working group ( $n=38$ ) between 1 July and 31 August 2024.

The non-validated questionnaire, developed in Italian, was divided into a general section and two specific parts: one on protein substitutes or modulars, and the other on low-protein or ketogenic foods.

Participants were instructed to respond based solely on patients residing in the same region as their clinical center to provide an accurate representation of the regional context.

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



When asked to reflect on how FSMPs are provided in their region, 14 respondents (48.3%) believed that a standardized process was established by the regional authority and applied uniformly across the territory. In contrast, 15 participants (51.7%) thought that different procedures were used within the same region, depending on the patient’s local health district of residence.

Regarding the prescription format, 19 participants (65.5%) reported that FSMPs – both protein substitutes and low-protein foods – were prescribed within a single prescription plan document, while 10 respondents (34.5%) indicated that separate documents were used.

The validity of the FSMP prescription plan was reported to be predominantly 12 months (21/29, 72.4%), with shorter durations described as less common.

Many dietitians (75.9%) indicated that a declaration of irreplaceability/non-substitutability is required for FSMPs listed in the national registry. This declaration certifies that a specific FSMP is essential for the patient and cannot be replaced with alternative products, thereby allowing access to the most suitable formulation. In contrast, 24.1% of respondents stated that such a declaration is never required.

Perceptions of procedural authorizations varied widely. Approval of prescription plans by local health authorities or hospital pharmacies was reported by the majority (23/29, 79.3%). However, only nine respondents (31.0%) believed that authorization by the Regional Rare Disease Coordination Centre is always required.

Regarding the frequency of monthly prescription renewals specifying product type and quantity by the general practitioner (GP) or pediatrician, four respondents (13.8%) believed that such renewals always occur, while 12 participants (41.4%) stated that this never occurs.

Finally, authorization of monthly prescription renewals by local health authorities was believed to occur never or rarely by most respondents, 15 (52%) and 7 (24%), respectively.

Further details are presented in Figure 2.

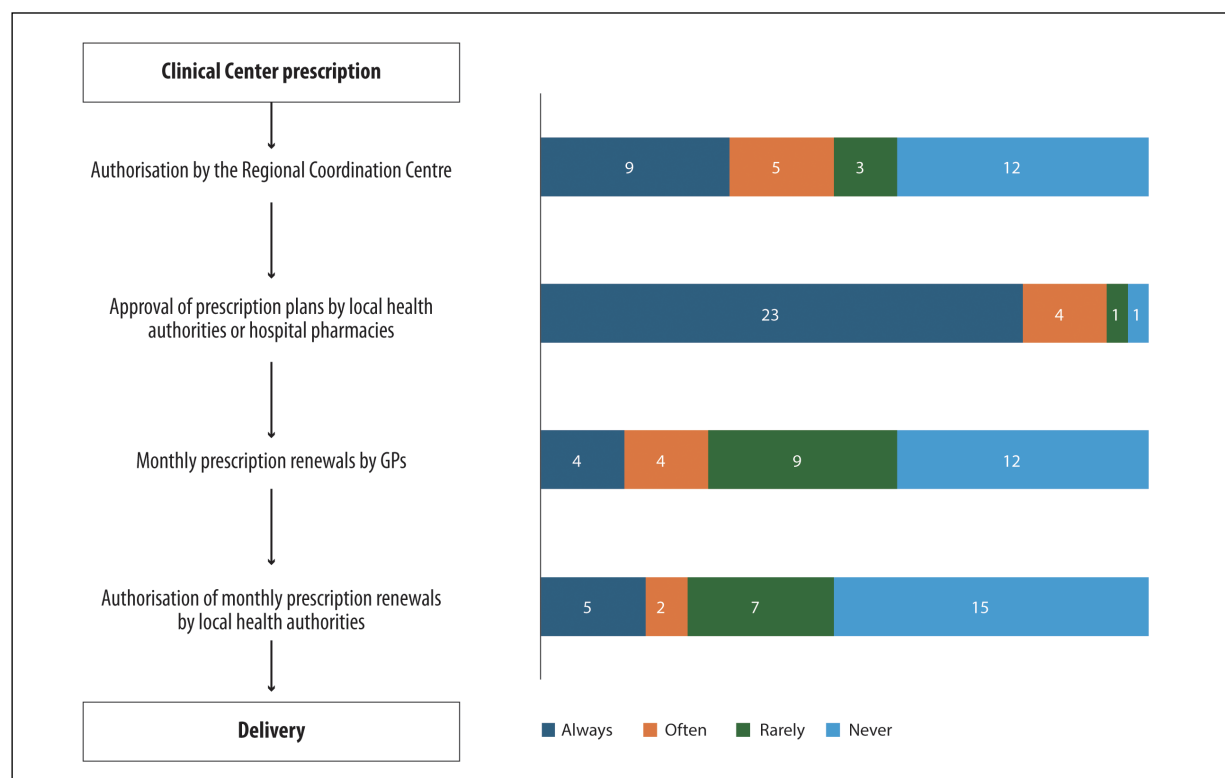


Figure 2. Procedural authorizations of prescription plans.

Home delivery of FSMPs was reported to be rarely available (4/29, 13.8%).

Most respondents (25/29, 86.2%) indicated that patients sometimes have to purchase FSMPs at their own expense. Dietitians noted that patients frequently purchase low-protein or ketogenic FSMPs independently due to various factors, including product unavailability at pharmacies (22/29, 75.9%), lengthy wait times for free supplies (9/29), exclusion of certain products from regional formularies (4/29), or insufficient regional budget allocations (6/29).

Regarding delivery times, 10 respondents (34.5%) estimated that the time from prescription to delivery often exceeds 2 weeks, while three participants (10.3%) reported delays exceeding 4 weeks.

FSMP delivery was predominantly reported to occur on a monthly basis (27/29, 93.1%), with only two respondents indicating a bimonthly schedule.

When asked whether pharmacies in their regions primarily source FSMPs from manufacturers or wholesalers, 11 respondents (37.9%) believed that wholesalers were the main supply route, while an equal number (37.9%) were unable to provide precise information.

According to most respondents (22/29, 75.9%), when a patient works or studies in a different region, FSMPs are typically supplied by the patient's region of residence. Only four respondents (13.8%) indicated that access is dependent on a formal change in healthcare residency status.

### Section 1 – Protein Substitutes or Modulars

Regarding protein substitutes or modulars, 14 out of 29 participants (48.3%) stated that dietitians primarily make product selections. However, only six respondents (20.7%) reported that the prescription plan is signed jointly by the dietitian and physician. The dietitian's signature was never reported to occur independently of the physician's (Figure 3).

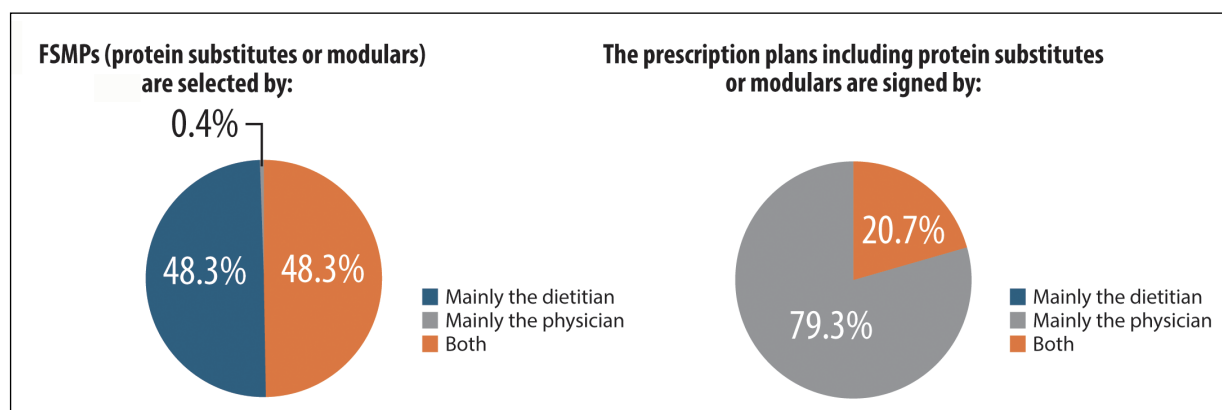


Figure 3. FSMPs (protein substitutes or modulars) selection and prescription plan signature.

FSMP prescriptions were most commonly reported to be electronic (16/29, 55.2%), although paper prescriptions were still in use, according to nine respondents (31.0%).

Hospital and territorial pharmacies were identified by 17 participants (58.6%) as the primary distribution channels for protein substitutes and modular products.

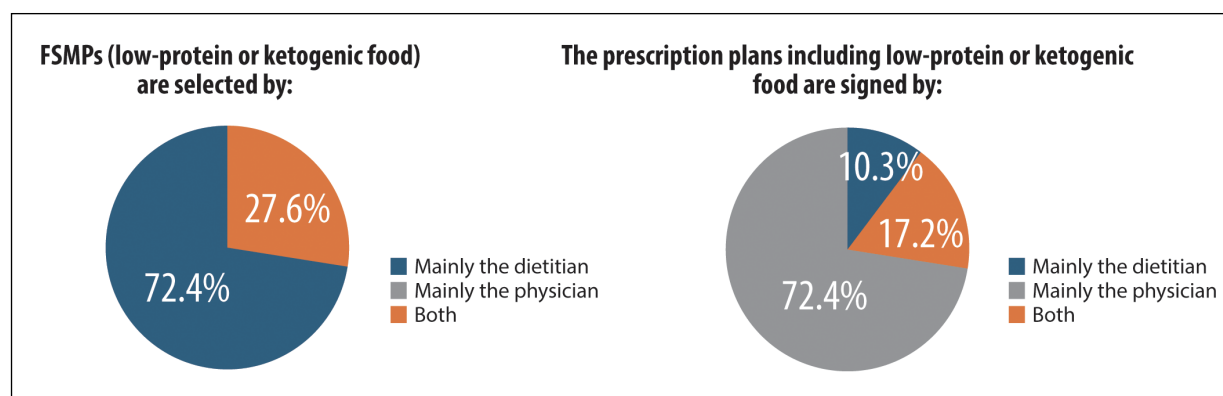
### Section 2 – Low-Protein or Ketogenic Food

A predefined monthly spending limit for low-protein FSMPs was reported by 10 respondents (34.5%). Among these, issues included an insufficient budget for adult patients (2/10) and the failure to adjust the budget in response to rising product prices (3/10).

Additionally, the presence of a regional procurement procedure for low-protein FSMPs was reported by five dietitians, specifically from Abruzzo, Calabria, and Tuscany. A key concern highlighted was the difficulty in accessing products not included in the regional procurement system.

Regarding ketogenic and low-protein foods, 21 participants (72.4%) stated that dietitians primarily make product selection. In 17% of responses, the prescription plan was reported to be signed jointly by the dietitian and the physician, while in 10% of cases, the dietitian was reported to have signed the plan independently (Figure 4).

Prescription of these FSMPs was mainly reported to be digital (14/29, 48.3%), although paper prescriptions were still used according to 10 participants (34.5%).



**Figure 4.** FSMPs (low-protein or ketogenic food) selection and prescription plan signature.

Most respondents (25/29, 86.2%) reported that prescription plans for ketogenic and low-protein foods typically include the commercial name, product categories, and/or quantities. The responses indicate that the selection of specific brands falls on patients (11/29, 37.9%), dietitians (5/29, 17.2%), or a combination of both (6/29, 20.7%), while in a few cases, this role was attributed to the physician (4/29, 13.8%).

When vitamin and mineral supplements – classified as non-FSMPs under current legislation – were required, 17 participants (58.6%) indicated that they were still included in the prescription plan. However, reimbursement for these products was reported to be inconsistent, with only eight respondents (27.6%) stating that they are reimbursed without issues.

For ketogenic and low-protein foods, dietitians reported that private pharmacies represent the primary distribution channel for FSMPs (20/29, 69.0%).

## DISCUSSION

The collection of data from 29 dietitians across 60% of Italian regions offers a valuable overview of current practices related to the prescription and supply of FSMPs in the context of IMDs. The variety of responses highlights significant regional differences, with some variation even reported within individual regions, underscoring potential disparities in patient access to FSMPs.

Notably, only 48.3% of dietitians believed that a standardized and uniform regional procedure for FSMP distribution exists, whereas 51.7% perceived differences based on the patient's local health district. These findings raise concerns regarding the uniformity of care at the national level and suggest possible inequalities in access to essential dietary resources.

An important aspect pertains to prescription modalities. A majority of dietitians (65.5%) reported that both protein substitutes and low-protein foods are prescribed within a single prescription plan, while 34.5% indicated that separate documents are used. Furthermore, approximately half of the respondents reported using a computerized prescription system, whereas 30% continue to rely on paper-based forms. The remaining respondents indicated the use of both methods.

By contrast, in the UK, a centralized digital prescription service managed by the National Health Service is in place<sup>12</sup>, representing a potentially valuable model for Italy to adopt in order to improve the efficiency, traceability, and uniformity of FSMP access.

Approval of prescription plans by local health authorities or hospital pharmacies was reported to be nearly universal among participants, whereas approval by the Regional Coordination Centre for Rare Diseases was less commonly cited (31%). This discrepancy suggests potential variability between regional procedures, highlighting the absence of a fully coordinated national system.

The reported delay between FSMPs' prescription and product collection, exceeding 2 weeks in 34.5% of cases, combined with the fact that many dietitians indicated patients are sometimes required to purchase FSMPs at their own expense, raises concerns about the overall efficiency of the distribution system and the timeliness of access to essential dietary products.

According to the Ministry of Health Guidelines on FSMPs<sup>13</sup>, these products are specifically formulated for patients whose dietary needs cannot be met through standard dietary modifications alone. In the Italian regulatory framework, FSMPs are legally classified as food products. As a result, unlike pharmaceuticals, they do not require a mandatory physician prescription.

In this context, we examined the role of dietitians in the selection and prescription of FSMPs, highlighting their central contribution to the dietary management of inherited metabolic disorders. According to the dietitians interviewed, involvement in the selection of low-protein foods occurred in 72% of cases, often without the participation of a physician. Similarly, 48% reported being involved in the selection of protein substitutes, frequently acting independently in this aspect of care.

However, it is noteworthy that dietitians are generally not involved in the formal authorization of the prescription plan. Specifically, the prescription plan is signed exclusively by the physician in 79% of cases for protein substitutes and 72% of cases for low-protein or ketogenic foods. Only a minority of cases – approximately 10% – involved the dietitian as the sole signatory of the prescription plan for low-protein foods.

These findings underscore the importance of close collaboration between dietitians and physicians, which is essential to ensure balanced and effective dietary management. However, they also reveal a critical issue: the contributions of dietitians are often not formally recognized or documented. This concern is further supported by the study of Gugelmo et al<sup>14</sup>, which highlights that many professional activities carried out by dietitians remain untracked and uncompensated, raising questions about the long-term sustainability of these services.

Moreover, a significant portion of dietitians' time is reportedly dedicated to communication with hospital and territorial pharmacies to resolve FSMP dispensing issues. Despite its importance, this administrative workload is neither officially acknowledged nor remunerated, reducing the time available for direct patient care. A similar situation has been reported in other countries, including the UK<sup>12</sup>.

In light of these challenges, it is essential to formally recognize and support the role of dietitians in FSMP prescription. A notable example comes from the UK, where, since 2016, dietitians have been authorized to act as supplementary prescribers for medicines following training accredited by the Health and Care Professions Council<sup>12</sup>. This model illustrates the potential benefits of expanding the dietitian's role in prescribing, with the aim of enhancing the quality of care and optimizing the therapeutic process in collaboration with physicians and the wider multidisciplinary team.

Reimbursement policies for FSMPs vary considerably across Europe and globally<sup>15,16</sup>. Across Europe, protein substitutes are generally reimbursed, whereas low-protein foods are not consistently covered. For example, Italy provides full reimbursement for both categories, while countries such as Spain, Germany, the Netherlands, and Poland do not reimburse low-protein foods or only in low-income cases<sup>17-19</sup>. In the UK, FSMPs are reimbursed<sup>20</sup>; however, challenges remain that may affect long-term adherence to dietary therapy<sup>12</sup>.

Further research is warranted to assess the current reimbursement landscape for FSMPs in Europe, particularly in light of recent policy updates. Existing analyses have largely focused on products intended for phenylketonuria, while reimbursement coverage for other inherited metabolic disorders remains insufficiently explored.

The cost of FSMPs and the difficulties associated with access remain central issues<sup>21</sup>. In our survey, a majority of respondents (86.2%) reported that patients sometimes need to purchase FSMPs independently, primarily due to supply shortages (26.7%) or insufficient regional budget allocations (20.7%). The latter issue is particularly prominent in regions where local regulations impose predefined monthly spending limits. Emilia-Romagna<sup>22</sup> and Veneto<sup>23</sup> are among the regions with monthly spending limits outlined in their policies.

Notably, spending limits for low-protein foods often do not adjust to rising food prices associated with inflation.

In other regions, different cost-containment strategies have been adopted. For example, in Tuscany, a dedicated entity known as ESTAR (Ente di Supporto Tecnico Amministrativo della Sanità Toscana) is responsible for the procurement of goods and services. A negotiated framework agreement with multiple suppliers under predefined conditions has been implemented as part of this system.

The framework agreement is a type of long-term contract used by public entities, such as hospitals, schools, or other institutions, to facilitate the procurement of goods or services in a more efficient and streamlined manner. In the context of FSMPs, this approach allows for the purchase of all products included within the base auction price of the assigned lot (Regione Toscana – ESTAR, Determinazione del Direttore del Dipartimento No. 1040 del 19/07/2022)<sup>24</sup>.

Our study highlights several challenges in the procurement process and the management of healthcare funds, which may lead to supply chain disruptions and create additional burdens for patients and their families. Insufficient access to low-protein foods may prompt patients to discontinue their dietary treatment, thereby placing them at a serious risk of adverse health outcomes.

The findings also emphasize the involvement of pediatricians and GPs in the FSMP prescription process. This aspect warrants further attention, as the additional step of prescription drafting by pediatricians or

GPs introduces a potential risk of transcription errors<sup>12</sup>. This risk is compounded by the fact that, although these professionals are essential to the healthcare system, they often lack specialized training in metabolic disorders and FSMPs. Their limited familiarity with the specific formulations and clinical indications of these products may compromise prescription accuracy, with potential consequences for patient safety.

To our knowledge, no prior study has been conducted in Italy to investigate the challenges that patients with IMDs face in accessing FSMPs. In contrast, a patient survey<sup>25</sup> conducted in the UK identified substantial issues within their FSMP distribution system. Reported difficulties included obtaining prescriptions from GPs, receiving incorrect quantities, delayed or incomplete deliveries, out-of-stock items, prescription refusals or restrictions by GPs, and instances of out-of-date, damaged, or poor-quality products.

In our survey, several challenges also emerged during data collection and analysis. One of the primary issues was the lack of consistent terminology across regions, which complicated the alignment and comparison of responses. Regional healthcare organizations – such as ASL, USL, ASUR, and AST – are referred to differently depending on the territory, leading to confusion in identifying the applicable regulations, administrative responsibilities, and supply procedures.

Additionally, regional decrees are often difficult to find and interpret, particularly when they are not explicitly tailored to the management of IMD. This lack of clarity complicates the standardization and application of regulations. Another significant challenge is the lack of uniformity in practices within single regions. Despite theoretically standardized policies, local implementation varies significantly, leading to disparities in access to and distribution of FSMPs.

The rarity of IMDs further exacerbates these challenges, as local health employees may not be fully informed about the FSMP prescription process. As a result, families may be directed to inappropriate administrative pathways, originally designed for other conditions, which in turn limit their access to FSMPs.

Furthermore, transcription errors could lead to the supply of incorrect FSMPs, which could potentially be harmful to the patient, especially if multiple professionals, who are not experts in dietetics and nutrition for IMDs, are involved in the procurement process.

Finally, the findings reveal that dietitians often possess fragmented and incomplete knowledge of FSMP-related regulations, underscoring the need for more standardized and comprehensive training across regions. Enhancing dietitians' familiarity with the legislative framework could significantly improve the efficiency and consistency of FSMP management throughout Italy.

This study has several limitations. First, it does not achieve full national coverage, as responses were collected from dietitians in only 60% of Italian regions. Second, as the survey was designed to capture the individual perceptions of dietitians, responses from the same center were not aggregated. Therefore, data reflect dietitians' perceptions, which are inherently subjective and may not fully capture institutional or patient-level realities. This aspect should be considered in future investigations for broader coverage and further collection of quantitative data.

Future research should aim to incorporate the perspectives of patients and caregivers, which would provide a more comprehensive and balanced understanding of the barriers to FSMP access and the effectiveness of current distribution systems.

## CONCLUSIONS

Although FSMPs are included in the Essential Levels of Care (LEA) in Italy, disparities in regional policies and administrative procedures continue to hinder equitable access. These inconsistencies may negatively impact dietary adherence and, consequently, clinical outcomes. Indeed, difficulties related to FSMP administration can substantially affect treatment adherence and reduce the quality of life for individuals with inherited metabolic disorders. To address these challenges, national-level harmonization of policies is urgently needed to reduce regional health inequalities<sup>26</sup>.

Another key finding of this study is the significant role of dietitians in the selection of FSMPs and ongoing patient monitoring despite their limited formal authority in prescribing. Additionally, their involvement in administrative activities, such as coordinating with pharmacies, is often unrecognized and uncompensated, further highlighting the need to formally acknowledge and support their contributions within the multidisciplinary care team.

In the future, a comprehensive analysis of FSMP reimbursement and distribution policies will be essential to identify critical issues and provide strategic insights for optimizing FSMP management across Italian regions. To achieve this, regional resolutions should be systematically reviewed, and local procedures for FSMP access should be analyzed to identify best practices that could inform national-level implementation.

The regulatory framework and reimbursement procedures for FSMPs must be designed to ensure fair and timely access to appropriate products for all patients, regardless of geographic location.

To improve FSMP access and management, several key priorities should be addressed: standardization of regional and national policies, formal recognition of the dietitian's role, enhancement of professional training, strengthening of interdisciplinary collaboration, and integration of telemedicine into routine care pathways.

A coordinated national approach is crucial to ensure equitable and timely dietary management for patients with inherited metabolic disorders. Such an approach would support improved metabolic control, enhance quality of life, and promote the economic sustainability of the national healthcare system.

#### **ARTIFICIAL INTELLIGENCE-ASSISTED TECHNOLOGIES:**

No artificial intelligence-assisted technologies were used in the production of this article.

#### **AUTHORS' CONTRIBUTIONS:**

Study conception and design: Micaela Gentilucci, Elena Massimino, Cinzia Pistolesi, Giulia Paterno, Simona Salera, Alessandra Tavian, collection and interpretation of data: Micaela Gentilucci, Simona Salera, Alessandra Tavian; statistical analysis: Alessandra Tavian; Simona Salera; manuscript drafting: Micaela Gentilucci, Simona Salera, Alessandra Tavian; manuscript editing: Micaela Gentilucci, Simona Salera, Alessandra Tavian; Elena Massimino approval to submit: Micaela Gentilucci Alessandra Tavian.

#### **AVAILABILITY OF DATA AND MATERIAL:**

The datasets generated and analyzed during the current study are available from the corresponding author upon reasonable request.

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#### **CONFLICT OF INTEREST:**

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

#### **CONSENT TO PARTICIPATE:**

Informed consent was not required as clinical outcome or patient-specific data were not included.

#### **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 Determina 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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