

INVESTIGATING THE CURRENT STATE OF THE ITALIAN WORKFORCE IN THE CONTEXT OF INHERITED METABOLIC DISEASES: AN IN-DEPTH SURVEY OF DIETITIANS ACROSS THE COUNTRY

G. GUGELMO¹, F. BERCHIELLI², C. BONFANTI³, A. CIPRIANI⁴, I. FASAN⁵, S. M. BERNABEI⁶, S. FERRARO⁷, A. FINIZI⁸, M. GENTILUCCI⁹, S. GIORDA¹⁰, C. LORO⁵, C. PANCALDI¹¹, S. PAROLISI¹², L. PONTILLO¹³, R. PRETESE³, S. QUATTRINI¹⁴, A. RE DIONIGI⁸, M. G. REGAZZI¹¹, A. ROSSI¹¹, S. SALERA¹⁵, M. TOSI^{16,26}, A. FAVARO¹⁷, G. PATERNO¹⁸, R. CARELLA¹⁸, E. MASSIMINO¹⁹, C. PISTOLESI²⁰, N. DURATTI²⁰, A. TAVIAN²¹, A. POZZOLI²², S. LA FATA²³, M. P. IERARDI¹³, A. BOSETTI²⁴, S. DE LEO²⁵, J. ZUVADELLI⁸, G. BRUNI⁴, A. DIANIN¹⁷

• • •

¹Department of Medicine, Division of Metabolic Diseases, University Hospital of Padua, Padua, Italy

²Division of Pediatrics, S. Chiara General Hospital, APSS, Trento, Italy

³Pediatric Department, Metabolic Rare Disease Unit, Fondazione IRCCS San Gerardo dei Tintori, Monza, Italy

⁴Dietetic Unit, Meyer Children University Hospital IRCCS, Florence, Italy

⁵Department of Medicine – DIMED, University Hospital of Padua, Padua, Italy

⁶Department SITRA, Nutritional Rehabilitation Unit, Ospedale Pediatrico Bambino Gesù IRCCS, Rome, Italy

⁷Department of Health Sciences, Pediatric Unit, Magna Graecia University of Catanzaro, Catanzaro, Italy

⁸Clinical Department of Pediatrics, San Paolo Hospital, ASST Santi Paolo e Carlo, Milan, Italy

⁹Department of Pediatrics, "Santo Spirito" Hospital of Pescara, Pescara, Italy

¹⁰Department of Pediatrics, Metabolic Diseases, University of Turin, Turin, Italy

¹¹Clinical Nutrition and Metabolism Unit, IRCCS AOUBO, Bologna, Italy

¹²UOSD Metabolic Diseases, AORN Santobono-Pausilipon, Naples, Italy

¹³Nutritional Center, Giannina Gaslini Pediatric Institute, IRCCS (Scientific Institute for Research, Hospitalization, and Healthcare), Genoa, Italy

¹⁴Department of Pediatrics, Polytechnic University of Marche, Ancona, Italy

¹⁵Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico, Regional Clinical Center for Expanded Newborn Screening, Milan, Italy

¹⁶Department of Health Sciences, University of Milan, Milan, Italy

¹⁷Regional Centre for Newborn Screening, Diagnosis and Treatment of Inherited Metabolic Diseases and Congenital Endocrine Diseases, Pediatric Unit C, University Hospital of Verona, Verona, Italy

¹⁸Department of Metabolic Diseases, Clinical Genetics and Diabetology, Giovanni XXIII Children Hospital, Azienda Ospedaliero-Universitaria Consorziale, Bari, Italy

¹⁹Department of Clinical Medicine and Surgery, Federico II University, Naples, Italy

²⁰Dietetic Unit, Careggi University Hospital, Florence, Italy

²¹SOSD Technical Health Care Professions - Dietitian and Regional Coordinating Centre for Rare Diseases, Azienda Sanitaria Universitaria Friuli Centrale, Udine, Italy

²²Pediatric and Neonatology Unit, Regional Referral Clinical Center for Inborn Errors of Metabolism, Guglielmo Da Saliceto Hospital, Piacenza, Italy

²³CRR Malattie Metaboliche Ereditarie per la Sicilia Occidentale, Pediatric Clinic, ARNAS Civico Di Cristina Benfratelli, Palermo, Italy

²⁴Department of Pediatrics, Centro di Nutrizione Clinica, Diabetologia e MME, Vittore Buzzi Children's Hospital, Milan, Italy

²⁵Department of Human Neuroscience, Sapienza University of Rome, University Hospital Policlinico Umberto I, Rome, Italy

²⁶Department of Pediatrics, Unit of Child Neurology and Psychiatry, Vittore Buzzi Children's Hospital, Milan, Italy

CORRESPONDING AUTHOR

Giorgia Gugelmo, RD; e-mail: giorgia.gugelmo@unipd.it

ABSTRACT – Objective: To investigate the expertise, role, and involvement of Italian dietitians in the dietary management and nutritional follow-up of patients affected from inherited metabolic diseases (IMDs) in referral centers.

Materials and methods: A total of 17 (2014 Survey) and 27 (2024 Survey) multiple-choice or short-answer questions were distributed online to affiliated Italian centers, categorized per general information, types and characteristics of activities, number of healthcare professionals in the facilities and their education/training.

Results: One to three dietitians are present in most metabolic centers, but 60% of participants devote no more than half of their working time to IMDs (2024 Survey). The number of patients per dietitian in a unit is currently one hundred or more for 60% of respondents. The distribution of resources and workforce is very uneven among the affiliated centers and poorly aligned with the allocated full-time equivalents. The training and education of specialized dietitians, as well as the amount of investment in research activities to advance the understanding of nutritional management and the outcomes of these pathologies, appear insufficient. The involvement of the figure of the dietitian in the process of sharing knowledge and experience through databases and conferences and in the elaboration of national guidelines still appears insufficient.

Conclusions: The role of the dietitian has evolved over the last decade, becoming indispensable within a multidisciplinary team dedicated to IMD. Therefore, future initiatives must focus on the specialized education and training of this figure and an adequate recognition of his or her activities within a truly interdisciplinary activity.

KEYWORDS: Inherited metabolic disease, Dietetics, Dietitians, Healthcare system, Italian survey.

LIST OF ABBREVIATIONS: FTE - Full Time Equivalent; HCPs - Healthcare professionals; IMDs - Inherited metabolic diseases; MNT - Medical nutrition therapy; MNTP - Medical Nutrition Therapy for Prevention; SIMMESN - Italian Society for the Study of Hereditary Metabolic Diseases and Newborn Screening.

INTRODUCTION

Inherited metabolic disorders (IMDs) are a diverse group of more than 1685 rare genetic conditions correlated with specific biochemical phenotypes (for instance, enzyme deficiencies). These conditions impact the metabolism of many biomolecules^{1,2} and may lead to complications in the liver, brain, heart, muscle and kidney^{1,3,4}.

To better manage symptoms and to improve the quality of life of affected individuals, many IMDs require specific dietary treatments that may include avoiding or reducing the intake of certain foods, using dietary supplements and special medical purpose foods to prevent nutrient deficiencies, planning an appropriate meal time to prevent fasting-induced metabolic stress, and introducing different dietary protocols during intercurrent diseases^{4,5}. In light of the recent discovery of these disorders and the increased survival of patients through early diagnosis and treatment, the study of patients' clinical and nutritional outcomes needs further, more in-depth and specific investigations. Furthermore, the number of foods for special medical purposes indicated for IMDs has increased over the years, and their therapeutic role and nutritional quality are important matters of research^{6,7}. However, as a yet poorly reported topic in clinical research, the long-term efficacy and nutritional impact of IMD-focused dietary interventions is still unclear⁷⁻¹¹. Attention has also been focused on the national healthcare systems, highlighting the importance of building an equitable and accessible dietetic service for patients with IMDs who require nutritional support.

Dietitians represent a category of healthcare professionals (HCPs) who hold at least a bachelor's degree and support individuals, families, groups, and populations in achieving nutritionally adequate, safe, tasty, and sustainable diets¹². Following medical nutrition therapy (MNT) as an evidence-based application of the Nutrition Care Process to assess nutritional requirements in different settings throughout the lifespan, dietitians provide tailored advice and treatment to maintain, reduce risk to, or restore health, resulting in the prevention, delay, or management of a disease^{13,14}. This also applies to the dietary management of metabolic disorders^{8,15}. Metabolic dietitians are specialized in managing patients with IMDs, developing personalized dietary plans (including urgent emergency revisions), and implementing dedicated education programs^{16,17}. Nowadays, the role of metabolic dietitians has expanded thanks to the advances in understanding the biochemical pathways underlying these disorders and the development of targeted therapies, enabling the evaluation of new therapies in clinical trials or the implementation of guidelines and protocols for best practice management in clinical settings¹⁶. One example is the introduction of telemedicine, which has accelerated the overall care, assistance, and treatment of various diseases, including IMDs, due to the emergence of the COVID-19 pandemic. Despite the lack of adequate (sometimes still crucial) infrastructures and equipment to carry out specific testing or treatments, telemedicine has been reported to be advantageous for patients in terms of saving time and money to travel to the hospital and thus facilitating access to the service^{18,19}.

Given the complexity of IMDs and the need to ensure comprehensive care, the dietitian's role involves close collaboration with a multidisciplinary team, including medical doctors, biochemists, nurses and other HCPs^{20,21}.

In recent years, concerns have arisen about potential clinical inadequacies and the inability to meet the nutritional demands of IMD patients, thus widening the gap between patient needs and dietetic service availability²². This urges further investigations into the composition of referral clinical centers in terms of expertise and specialties, as well as the involvement of dietitians, their training, and their role in the management and follow-up of metabolic patients, highlighting potential variations among treatment centers, to ultimately ensure equal access to effective and affordable services²³.

This work aims to provide an overview of the Italian workforce of dietitians dedicated to IMDs over the last decade, to explore the use of telemedicine in this context, and to identify and assess the unmet needs in the healthcare system and the working conditions of dietitians. The identified needs were further discussed, and potential strategies were finally suggested to improve the current state and conditions of Italian dietitians in the context of IMDs.

MATERIALS AND METHODS

2014 Survey

A survey consisting of 17 multiple-choice and short-answer questions (**Supplementary Material 1**) was distributed online to all Italian dietitians working on IMDs through their respective clinical center contacts in 2014. Although the questionnaire was not formally validated, it included questions previously developed and used in similar surveys to assess the role of dietitians in different countries²². The 2014 Survey consisted of 17 questions, categorized into four areas: (i) general information, such as the reference clinical center, number of dietitians in the IMD team, age, and type of contract (questions 1, 2, 3, 6, 7); (ii) types and characteristics of activities, including the presence of a multidisciplinary team, number of IMD treated patients, involvement in national or local projects, research, publications, current difficulties, and suggestions for improving the role of the dietitian in IMDs (questions 9, 10, 11, 12, 13, 14, 15, 16, 17); (iii) number of HCPs working in the facility (questions 9, 10); and (iv) education/training of HCPs, including professional qualifications, relevant training experiences, and areas of responsibility (questions 4, 5, 8).

Participating professionals were asked to complete a self-reported survey considering data as of 31 July 2014.

2024 Survey

An online survey with 27 multiple choice and short answer questions (**Supplementary Material 2**) was distributed to all Italian members of the Nutrition and Dietetic working group of the Italian Society for the Study of Hereditary Metabolic Diseases and Newborn Screening (SIMMESN) and to the Italian dietitians working in IMDs through their respective clinical center contacts. The questionnaire was not val-

dated; however, it included questions previously present in the 2014 questionnaire survey, with some new sections on new issues related to the current context. The subject of the questions was discussed and agreed upon in the dietary meetings of the SIMMESN Nutrition and Dietetic working group.

The questions of the 2024 Survey were categorized into four areas: (i) general information, such as the region where the service was provided, care settings, and the number of treated patients (questions 1, 2, 7, 18); (ii) types and characteristics of activities, including the presence of a dedicated team, current difficulties and proposals for improving the nutritional care, the involvement in research or audit work (questions 15, 16, 17, 19, 20, 21, 22, 23, 27 items); (iii) the use of telemedicine (3, 9, 10, 11, 12, 13, 24, 25, 26) and the number of HCPs working in or with the facility, expressed as the total number and full-time equivalent (FTE). For instance, if 40 hours are considered as a full-time workweek, an employee working 40 hours per week will have an FTE of 1.0. In contrast, a part-time employee working only 20 hours per week will have an FTE of 0.5.; and (iv) education/training of HCPs (questions 5, 6, 8).

Participating professionals were asked to complete a self-reported survey considering data as of 31 May 2024. Ethical approval was not required as clinical outcome or patient-specific data were not included.

STATISTICAL ANALYSIS

All data from both the 2014 and 2024 Surveys were analyzed using descriptive statistics. When data were available for both surveys, the outcomes for specific issues were qualitatively compared between 2014 and 2024. Data from open questions were extracted and presented per specific and recurrent topic.

RESULTS

General Information

In 2014, 20 HCPs responded to the survey: 8/20 were between 25–35 years old (yo), one participant was in the range of 35–44 yo, 2/20 were 45–54 yo, and 9/20 were above 55 yo. Dietitians from ten affiliated centers were located in northern regions, six in central regions, and four in the south of the country. Regions including Valle d'Aosta, Piemonte, Trentino-Alto Adige, Umbria, Abruzzo, Molise, Puglia, Calabria, Basilicata, Sicilia and Sardegna lacked a reference dietitian in the 2014 Survey.

In the 2024 Survey, 37 HCPs participated, of whom 17/37 were between 25 and 34 yo, 14/37 were between 35 and 44 yo, only one participant was less than 25 yo, 3/37 were 45–54 yo, and 2/37 were above 55 yo. Participants were affiliated with centers located mainly in the northern regions (23/37), seven in the center of Italy, and seven in the southern area or Islands. None of the participants in the 2024 Survey were affiliated with centers located in the region of Valle d'Aosta, Molise Sardegna and Basilicata.

In 2014, except for two cases only qualified with a Economic-Dietitian Diploma, all participants graduated in dietetics (18/20) and obtained a master's degree (5/20) and/or a master's course (4/20). Concerning the specific education and training related to IMDs, most participants (15/20) reported regularly going to congresses, five completed training courses, and only one attended a module from a university course. Except for three Biology graduates with additional specialization in Nutrition, all participants of the 2024 Survey majored in dietetics (34/37), combined with a master's degree (14/34) and/or a master's course (7/34). Two participants also held a PhD, and one of them further graduated in Medicine. In the specific context of IMD training and education, several individuals (32/37) participated in targeted congresses, some attended training courses (12/37). In contrast, only a few cases (6/37) attended a single module dedicated to IMDs from a university master course in the UK.

In 2014, 60% (12/20) of participants were employed with a permanent contract, three had a temporary contract, one had an annual research project, and four had a scholarship. In 2024, around 80% of the participants (29/37) had a permanent contract, one had a research contract, 5/37 had a scholarship, 2/37 were free-lancers; the working experience was up to 10 years for more than 50% of them (27/37), up to 20 years (7/37 cases) or above 20 years (3/37) (Table 1).

Table 1. General information of the participants in the 2014 and 2024 Surveys.

Year	Age				
	<25 years, n (%)	25–34 years, n (%)	35–44 years, n (%)	45–54 years, n (%)	>55 years, n (%)
2014	–	8 (40%)	1 (5%)	2 (10%)	9 (45%)
2024	1 (2.7%)	17 (46%)	14 (37.8%)	3 (8.1%)	2 (5.4%)

Year	Contract				
	Permanent, n (%)	Temporary, n (%)	Scholarship, n (%)	Research project, n (%)	Freelance, n (%)
2014	12 (60)	3 (15%)	4 (20%)	1 (5%)	–
2024	29 (78.4%)	0	5 (13.5%)	1 (2.7%)	2 (5.4%)

Year	Qualification					
	Economic-Dietitian Diploma, n (%)	Dietetics (Bachelor), n (%)	Master's Degree, n (%)	Master (II level), n (%)	PhD, n (%)	Biology Degree, n (%)
2014	2 (10%)	18 (90%)	5 (25%)	4 (20%)	–	–
2024	0	34 (91.9%)	14 (37.8%)	7 (18.9%)	2 (5.4%)	3 (8.1%)

Year	Geographical allocation		
	North, n (%)	Center, n (%)	South, n (%)
2014	10 (50%)	6 (30%)	4 (20%)
2024	23 (62.2%)	7 (18.9%)	7 (18.9%)

The activities of the dietitians

In the 2014 Survey, only one, two, or three dietitians worked in the affiliated center in seven (7/20), nine (9/20), and four (4/20) cases, respectively. Similarly, in the 2024 Survey, only one (14/37) or two (14/37) dietitians were usually present in the affiliated center; otherwise, there were three dietitians per unit (8/37), and only one case reported four dietitians in the center. All dietitians referred to at least one physician to work within the team. Of their total weekly hours, participants of the 2024 Survey were asked about the number of hours specifically dedicated to the care of IMDs rather than other activities. Fifteen participants reported spending more than 80% of their working hours on IMDs, whereas more than half of participants (21/37) dedicated to IMDs only less than 50% of their working time (FTE <0.5), in fourteen of these cases (14/21) even less than 30%. Within the dedicated hours, a high number of dietitians (26/37) had in charge patients of both adult and pediatric age, while only four and seven participants worked with a focus on children and adult patients, respectively. In 2014, most dietitians (17/20) also reported working with both adults and children affected from IMDs; only one focused on adult age, and the other two only worked with children. Based on the reported range of IMD patients and the number of dietitians per unit, for 35% of cases (7/20) in 2014 and a higher number of cases (60%, 22/37) in 2024, there was one dietitian for more than a hundred patients in the unit. The number of patients per dietitian was 26–100 in 40% and 21% of cases in 2014 and 2024, respectively; conversely, in 25% of responses from the 2014 Survey and 19% from the 2024 Survey, there was one dietitian for less than 25 patients (Figure 1).

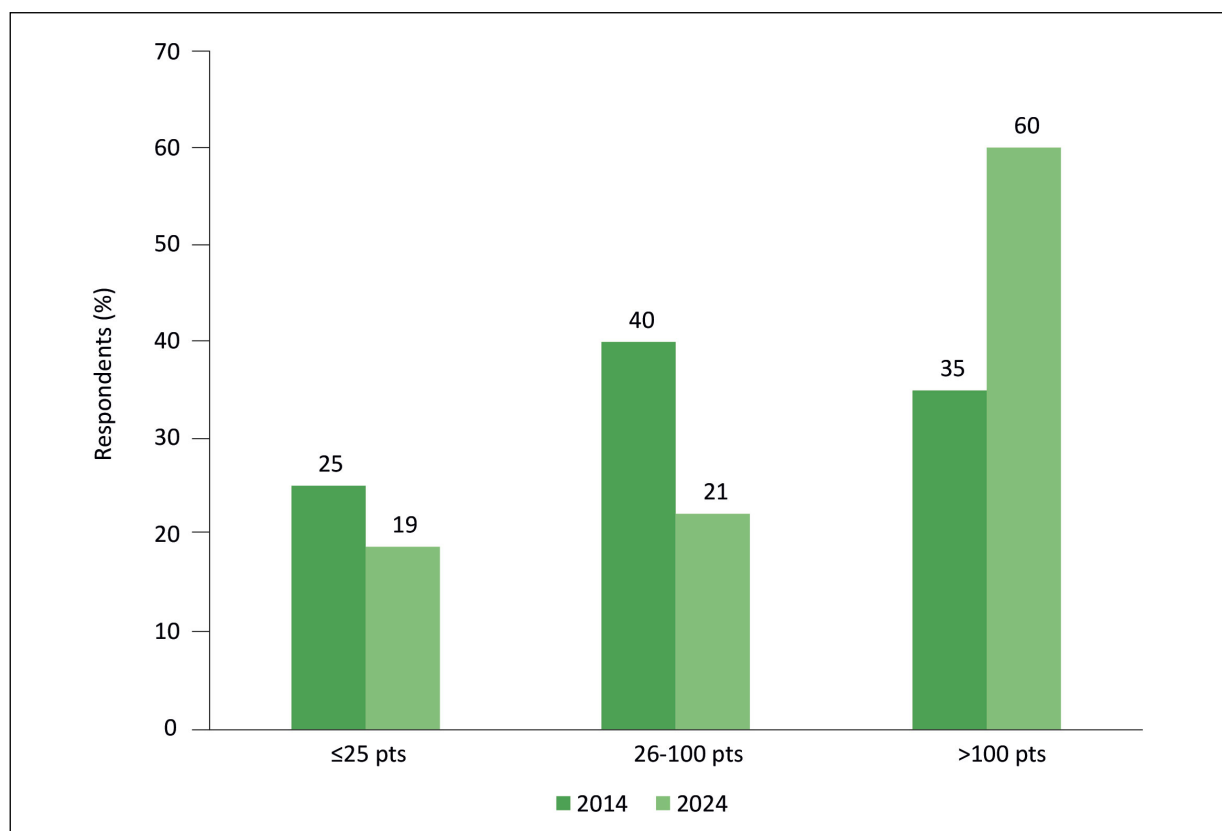


Figure 1. Cases (%) of respondents in charge of >100 patients, 26–100 patients or ≤25 patients in the unit, in 2014 (dark green) and 2024 (light green). *Abbreviation:* pts = patients.

Concerning the 2024 Survey, in half of these cases (11/22), dietitians also reported a FTE lower than 0.5 dedicated specifically to IMD activities (Table 2 and Figure 2). In addition, some respondents declared to work with other dietitians (no. 3) in their IMD center, who unfortunately did not participate in this survey.

Among four main settings proposed (hospital ward, service facilities/outpatient clinics, research and remote assistance), participants of the 2024 Survey were further asked to specify a percentage range of time/week working with IMDs in each type of activity (detailed results are summarized in Table 3). Working in service facilities seems to be a prevalent activity, in which a high number of participants (20/37) reported spending more than 50% of their IMD-focused working time. On the other hand, very little time seems to be generally dedicated to IMDs in the context of hospital departments and research, with a significant number of dietitians spending only 0–25% of their IMD-dedicated time (29/37 in hospital and 32/37 for research activity). Moreover, 26/37 participants reported assisting their IMD patients remotely for at least 25% of their working hours. As for the other potential activities, only 6/36 dietitians appear to invest the IMD-dedicated time in training or meeting with colleagues and other team members (7/36).

In the context of telehealth, seventeen participants clearly affirmed that they offer remote assistance to their patients for a range of 1–5 hours per week within their working hours. Most of them (14/17) further reported performing additional service over time. By analyzing the absolute number of tracked and overtime hours for each participant, in 9/14 cases, it emerged that the overtime hours were superior to the tracked ones.

As of the year 2014, all dietitians reported working as a part of a multidisciplinary team to assist IMD cases, which included pediatricians (20/20), nurses (16/20), psychologists (12/20) and child neuropsychiatrists (7/20). Similarly, almost the totality of participants in 2024 (35/37) were also part of a multidisciplinary team taking care of IMD cases. Besides the dietitians, other figures mainly present in the team were pediatricians (33/35), nurses (22/35), psychologists (21/35), child neuropsychiatrists (13/35),

Table 2. Workload of dietitians (2024 Survey) expressed by the total FTE per center, based on responding cases and related to a range of patients referred to the center.

Centers	No. Respondents	Total FTE*	Range of hospital patients/center
1	2	1.1	101–150
2	1	0.3	16–25
3	1	0.5	251–300
4	1	1	601–750
5	1	0.9	151–200
6	1	0.2	51–75
7	1	1	151–200
8	1	1	76–100
9	2	0.6	26–50
10	2	1.1	601–750
11	3	1.5	301–400
12	1	0.01	0–15
13	2	2	151–200
14	1	0.2	51–75
15	2	1.6	601–750
16	2	0.8	251–300
17	3	2.9	501–600
18	1	0.3	51–75
19	1	1	601–750
20	2	0.3	26–50
21	1	0.3	151–200
22	2	1.2	301–400
23	1	NA	16–25
24	1	0.2	101–150
25	1	0.3	251–300

*Total FTE is calculated based on the number of respondents. More than 100 patients per single dietitian are outlined in bold.

and neurologists (9/35). In three separate questions of the 2024 Survey, participants were also asked to provide the absolute number within the unit of dietitians and physicians specialized in IMDs and a range of patients under their assistance.

Final considerations and identification of unmet needs

All participants were asked to rate up from 1 to 5 the level of difficulty in specific aspects, such as the collaboration with other colleagues, the workload, the complexity of pathologies, the digitalization of services, logistics and availability in case of emergencies. A high number of participants (28/37) expressed very low (difficulty 2) or no difficulty (difficulty 1) concerning the collaboration with other team members. By contrast, several participants manifested a difficulty level of 3 to 5 regarding the workload (32/37) and emergency case management (30/37) (Figure 3).

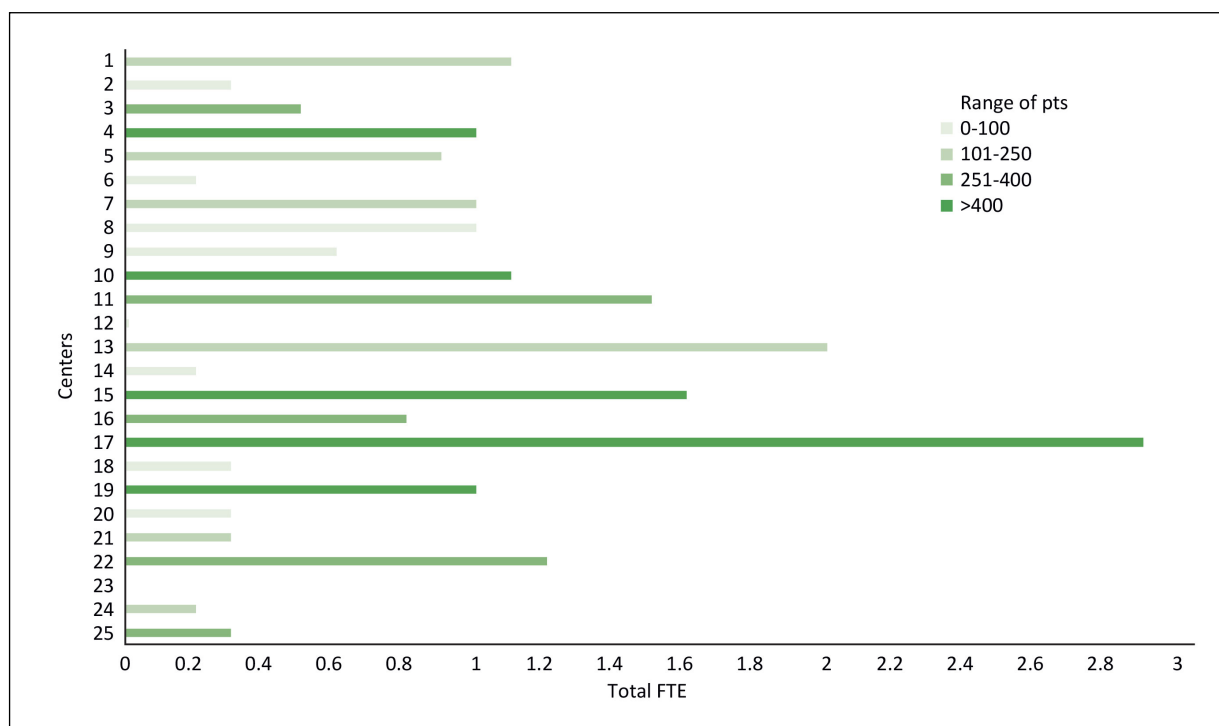


Figure 2. Dietitian workforce in full-time equivalents across anonymized Italian IMD centers. *Abbreviation:* pts = patients.

Table 3. Time (%) of IMD-dedicated hours in specific operative settings by dietitians (2024 Survey).

		Time (%) of IMD-dedicated hours				
		0–10%	10–25%	25–50%	50–75%	75–100%
Number of dietitians	Hospital	25	4	6	1	1
	Service facility or outpatient clinic	1	3	13	11	9
	Research	21	11	3	1	0
	Remote Assistance	2	9	15	4	7

In both 2014 and 2024, the dietitians surveyed were asked, in a more open form, to provide additional comments on their difficulties in performing their tasks and to indicate specific issues that could improve the activity and the general conditions of their role (see [Supplementary Material 1](#) and [Supplementary Material 2](#) for the detailed answers). In line with the results from the 2024 Survey, in question 20 and others, dietitians expressed complaints about the disproportional workload and the low-paid and often unrecognized activities carried out (with a specific reference to the telehealth services and emergency interventions). Furthermore, according to the answers provided, the dietitian's role in the care and assistance of IMD patients is often not acknowledged as appropriate. Given the difficult management of such complex pathologies, dietitians surveyed also expressed the need for a more specific and exclusive role of the dietitian for IMDs in terms of expertise and working hours, as well as to increase educational opportunities by creating specialized university master's programs. Furthermore, to benefit from shared knowledge and experience in a more accessible way, participants often suggested the creation of a network among IMD dietitians and clinical databases, major involvement in research

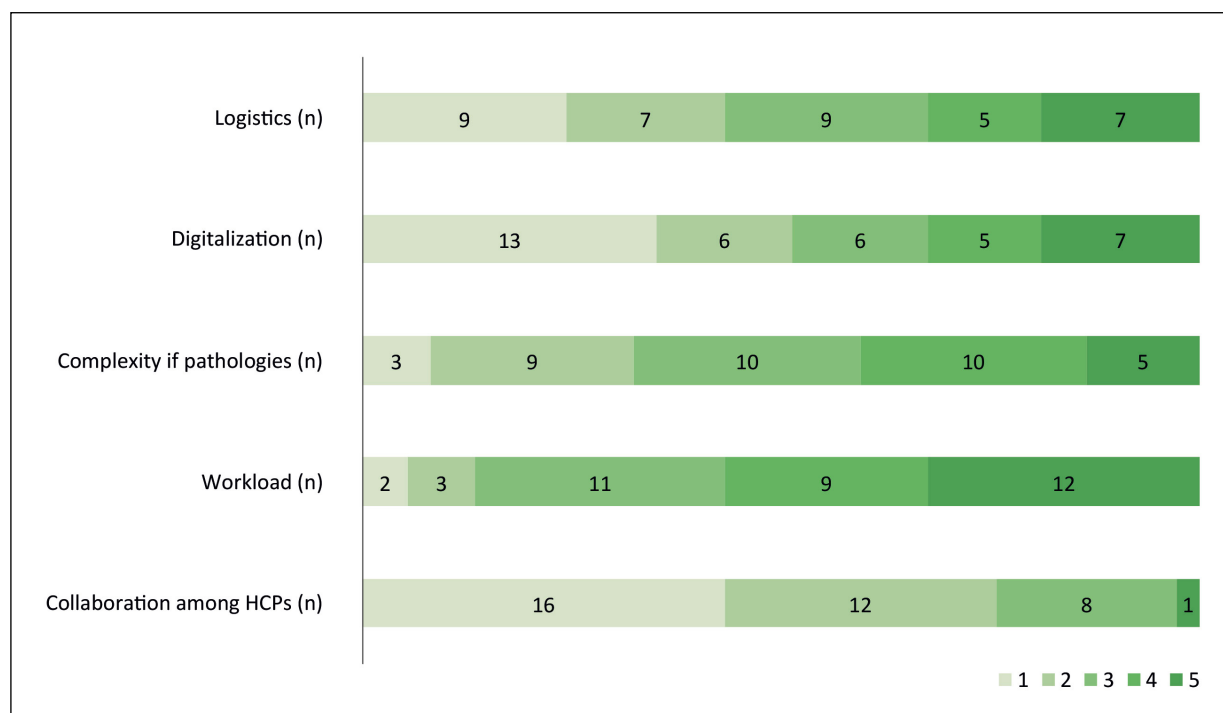


Figure 3. Difficulty of specific working-related issues as perceived by participants of the 2024 Survey. *Abbreviation:* n = number of respondents. Difficulty was ranged from 1 (no difficulty – light green) to 5 (very difficult – dark green).

projects, and national and international conferences. Compared with the results from the 2014 Survey (9/20 cases), the number of dietitians involved in research projects and scientific publications has increased to 80% (30/37). However, the time dedicated to this IMD-focused activity is still very low, as described above (32/37 cases spent less than 25% of their IMD-focused activity on research).

Furthermore, another frequently expressed need has been to facilitate the clinical management of such complex diseases with the national harmonization of guidelines in dietetics. Fourteen out of 37 (37.8%) participants have never been involved to date; however, they have expressed an interest in taking part in initiatives in this regard and think such experience would be useful.

DISCUSSION

Concerns about potential clinical inadequacies and incapacity to meet the nutritional demands of IMD patients suggest further investigations into the composition of referral clinical centers in terms of expertise and specialties^{22,23}. This work aimed to understand the current state and evolution of dietitian practices in Italy, focusing on managing IMDs over a decade.

During this period, job stability increased among dietitians, with permanent contracts rising from 60% in 2014 to 80% in 2024. With a significantly high prevalence in northern regions, the disproportionate geographical distribution of affiliated centers in 2024 compared with 2014 reflects a growing difference in the organization among the IMD regional or interregional dietetic services.

The clinical management by dietitians and, therefore, the quality of service provided to IMD patients also appears to be significantly variable with the center and, in some cases, very challenging, depending on the number of dietitians available and especially the FTE assigned (one to three dietitians are usually present in the center, and more than half of dietitians surveyed in 2024 (21/37) work with IMDs for an FTE less than 0.5). Such a scenario has also been reported elsewhere; for instance, nearly 20 years ago in the UK, the Joint Committee on Medical Genetics reviewed the existing services for IMDs and reported that 92% of British IMD care centers had a specialized IMD dietitian on their team but only 33% had more than one whole-time-equivalent²². In a more recent Canadian investigation²³, all participating IMD

centers reported at least one physician and one registered dietitian, assigned with FTE ranging from 0.2 to 2.8. To prevent work overload for dietitians, which mainly affects the quality of care provided to the patient, the distribution of resources and personnel within a team should be geographically better distributed and proportional to the patients in charge, also considering the actual FTE assigned.

The IMD clinical management often includes specialized life-saving interventions and ongoing health-care support with frequent adjustments of nutritional therapy²⁴. In this context, telemedicine has recently become crucial in-patient care for individuals with IMDs^{25,26}. However, most of this activity is not tracked and remunerated. According to the results of this work, most dietitians who responded to offer remote assistance reported providing this service over time. Time dedicated to inpatient admissions is lower than that of the service facility and remote assistance. Further investigations should address this aspect and its origins, whether patients can effectively manage emergencies at home or whether local hospitals that do not know the dietary needs of patients work in collaboration with referring metabolic dietitians. Therefore, a better monitoring and accurate quantification of this time, as well as direct salary costs for non-reimbursed care coordination activities by multidisciplinary programs, is needed²⁷.

Respondent dietitians reported spending limited time on research activities for IMDs, among other operative settings, rather than remote assistance. Nevertheless, in order to advance the understanding of IMDs and the long-term consequences of restrictive diets and to develop effective treatments, greater engagement in research activities should be encouraged by providing dietitians with adequate resources and time.

The complexity and novelty of IMDs have been strongly highlighted by dietitians in both surveys as a critical element in clinical management, which requires specific expertise of the dietitians. As diet remains the main treatment for many IMDs, dietitians still play an indispensable role within the multidisciplinary team^{21,28-35}, which should exclusively focus on IMDs with defined and specialized responsibilities in order to enhance the efficiency and effectiveness of patient management through clinical pathways or integrated care pathway of such complex disorders. Besides targeted congresses and training courses, the implementation of formal education on metabolic dietetics with master's programs at universities appears crucial for continuous professional development and the quality of the service provided.

SIMMESN includes a dietetic working group that could effectively empower dietitians to navigate future workload complexities and maintain high standards of patient care by prioritizing initiatives aimed at promoting research investigations of nutrition interventions and enhancing professional support and development, such as education tailored for emerging dietary practices³⁶. Moreover, several authors of this publication are members of the European Reference Network for Rare Hereditary Metabolic Disorders (MetabERN) (project ID No. 739543). Thanks to these projects, collaborative multi-center studies on practices and expert consensus should be performed employing the most used methodologies, such as the Delphi and GRADE approaches³⁷.

As the surveys also revealed, participation in conferences and congresses and establishing databases and networks to share knowledge and experience could also facilitate the clinical management of IMDs, thereby improving patient care. The development and implementation by multidisciplinary teams of standardized guidelines across the country for the management of IMDs, including dietary treatment, could further ensure harmonized consistency in care and improved outcomes.

In line with the evolving role of the dietitian¹⁶, our work outlines the importance of major involvement in activities other than patient management. For instance, the development of guidelines and protocols or the definition of expenditure limits for reimbursement and other policymaking for the healthcare system by providing important information regarding patient requirements of foods for special medical purposes. In the USA, this was already the case concerning developing a bioinformatics tool system by the Medical Nutrition Therapy for Prevention (MNT4P) program and the Public Health Informatics Institute, aimed at creating a digital health footprint for patients with IMDs identified through newborn screening³⁸.

LIMITATIONS

The study described in this work presents some limitations. Despite the increasing number of responding centers from 2014 to 2024, some centers may not be registered or accessible, which could impact the representativeness of the survey results. In addition, determining the exact time and services devoted to IMDs remains challenging. Variations in workload and unrecorded telemedicine activities contribute to the difficulty in accurately estimating the time and commitment of services provided by dietitians in managing IMDs.

CONCLUSIONS

The results of the two subsequent surveys provided an overview of the dietary management of IMD patients in Italy over the decade, offering insights into challenges and priorities for dietitians. The dietitian's role remains crucial for the nutritional care and support of many IMDs but requires specialized education and exclusiveness of the role within the multidisciplinary team. Given the continuing evolution of the role, metabolic dietitians should be acknowledged for all their activities, including patient care, telehealth services, research, and possible involvement in developing harmonized guidelines and protocols, reflecting adequate job titles, compensation, and opportunities for professional growth.

ARTIFICIAL INTELLIGENCE-ASSISTED TECHNOLOGIES:

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

AUTHORS' CONTRIBUTIONS:

Study conception and design: GG, AD; collection and interpretation of data: GG, AD; statistical analysis: GG, AD; manuscript drafting: GG, AD; manuscript editing GG, AD; approval to submit: GG, AD, JZ and GB. The other authors participated in the survey with their data contribution and provided comments and reviewed the original paper.

AVAILABILITY OF DATA AND MATERIAL:

All data generated or analyzed during this study are included in this published article and supplementary materials.

CONFLICTS OF INTEREST:

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

ETHICS APPROVAL:

Ethical approval was not required as clinical outcome or patient-specific data were not included.

FUNDING:

No funding was received for this study.

INFORMED CONSENT:

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

ORCID ID:

Giulia Bruni: 0009-0004-3546-4160
 Alessandra Cipriani: 0009-0007-8084-982X
 Alice Dianin: 0009-0000-9539-2649
 Giorgia Gugelmo: 0000-0002-3557-8078
 Sara Quattrini: 0000-0003-4192-2055
 Martina Tosi: 0000-0003-4859-5288
 Juri Zuvadelli: 0009-0009-9750-3360

REFERENCES

1. Ferreira CR, Rahman S, Keller M, Zschocke J; ICIMD Advisory Group. An international classification of inherited metabolic disorders (ICIMD). *J Inherit Metab Dis* 2021; 44: 164-177.
2. <http://www.icimd.org/>
3. Saudubray JM, Mochel F, Lamari F, Garcia-Cazorla A. Proposal for a simplified classification of IMD based on a pathophysiological approach: A practical guide for clinicians. *J Inherit Metab Dis* 2019; 42: 706-727.
4. Bernstein LE. 2015. *The Nutrition Management Of Inherited Metabolic Diseases: Lessons from Metabolic University*. July 2015 DOI: 10.1007/978-3-319-14621-8 Edition: First Publisher: Springer Editor: Bernstein LE, Rohr F, Helm JR. ISBN: 978-3-319-14620-1
5. Norris MK, Kanungo S. Role of Metabolic Nutrition in Newborn Screening and Inherited Metabolic Disorders. *OBM Genetics* 2023; 7: 196.
6. Salera S, Coacci S, Cipriani A, Gentilucci M, Pierattini V. Special low protein foods in the management of Inborn Errors of Metabolism: overview on availability, composition and comparison with regular foods in Italy. *JIM* 2024; 1: e532.

7. Garcia-Arenas D, Barrau-Martinez B, Gonzalez-Rodriguez A, Llorach R, Campistol-Plana J, García-Cazorla A, Ormazabal A, Urpi-Sarda M. Effect of Special Low-Protein Foods Consumption in the Dietary Pattern and Biochemical Profile of Patients with Inborn Errors of Protein Metabolism: Application of a Database of Special Low-Protein Foods. *Nutrients* 2023; 15: 3475.
8. Rocha JC, Ahring KK, Bausell H, Bilder DA, Harding CO, Inwood A, Longo N, Muntau AC, Pessoa ALS, Rohr F, Sivri S, Hermida Á. Expert Consensus on the Long-Term Effectiveness of Medical Nutrition Therapy and Its Impact on the Outcomes of Adults with Phenylketonuria. *Nutrients* 2023; 15: 3940.
9. Mochel F. What can pediatricians learn from adult inherited metabolic diseases? *J Inherit Metab Dis* 2024. doi: 10.1002/jimd.12729. Epub ahead of print.
10. Gugelmo G, Lenzini L, Francini-Pesenti F, Fasan I, Spinella P, Valentini R, Miraval A, Avogaro A, Vitturi N. Anthropometrics, Dietary Intake and Body Composition in Urea Cycle Disorders and Branched Chain Organic Acidemias: A Case Study of 18 Adults on Low-Protein Diets. *Nutrients* 2022; 14: 467.
11. Luengo-Pérez LM, Ambrojo A, Fernández-Bueso M, Guijarro M, Ferreira A, Luzes G, Pereira M, Calhau C, Rocha JC. Muscle Quality and Risk of Metabolic Syndrome in Adult Patients with Inherited Metabolic Diseases. *Endocr Metab Immune Disord Drug Targets*. 2023 Dec 11. doi: 10.2174/0118715303279040231129055755. Epub ahead of print.
12. <https://www.efad.org/definition-of-a-dietitian/>
13. Swan WI, Vivanti A, Hakel-Smith NA, Hotson B, Orreval Y, Trostler N, Beck Howarter K, Papoutsakis C. Nutrition Care Process and Model Update: Toward Realizing People-Centered Care and Outcomes Management. *J Acad Nutr Diet* 2017; 117: 2003-2014.
14. Definition of Terms. Academy of Nutrition and Dietetics. Published 2021. Accessed June 02, 2024. <https://www.eatrightpro.org/practice/quality-management/definition-of-terms>
15. Frazier DM, Allgeier C, Homer C, Marriage BJ, Ogata B, Rohr F, Splett PL, Stembridge A, Singh RH. Nutrition management guideline for maple syrup urine disease: an evidence- and consensus-based approach. *Mol Genet Metab* 2014; 112: 210-217.
16. Singh RH, Kaczmarczyk MM. Standards of professional practice for genetic metabolic dietitians. *Genet Med* 2008; 10: 290-293.
17. Acosta PB, Ryan AS. Functions of dietitians providing nutrition support to patients with inherited metabolic disorders. *J Am Diet Assoc* 1997; 97: 783-786.
18. Koç Yekedüz M, Doğulu N, Sürücü Kara İ, Öncül Ü, Bakırarar B, Kullu P, Ar Y, Köse E, Eminoğlu FT. Pros and Cons of Telemedicine for Inherited Metabolic Disorders in a Developing Country During the COVID-19 Pandemic. *Telemed J E Health* 2022. doi: 10.1089/tmj.2021.0610. Epub ahead of print.
19. Brunetti-Pierri N, Fecarotta S, Staiano A, Strisciuglio P, Parenti G. Ensuring continuity of care for children with inherited metabolic diseases at the time of COVID-19: the experience of a metabolic unit in Italy. *Genet Med* 2020; 22: 1178-1180.
20. Stepien KM, Kieć-Wilk B, Lampe C, Tangeraas T, Cefalo G, Belmatoug N, Francisco R, Del Toro M, Wagner L, Lauridsen AG, Sestini S, Weinhold N, Hahn A, Montanari C, Rovelli V, Bellettato CM, Paneghetti L, van Lingen C, Scarpa M. Challenges in Transition From Childhood to Adulthood Care in Rare Metabolic Diseases: Results From the First Multi-Center European Survey. *Front Med (Lausanne)* 2021; 8: 652358.
21. Burlina A, Leuzzi V, Spada M, Carbone MT, Paci S, Tummolo A. The management of phenylketonuria in adult patients in Italy: a survey of six specialist metabolic centers. *Curr Med Res Opin* 2021; 37: 411-421.
22. Burton H, Sanderson S, Dixon M, Hallam P, White F. Review of specialist dietitian services in patients with inherited metabolic disease in the United Kingdom. *J Hum Nutr Diet* 2007; 20: 84-92.
23. Lamoureux MF, Tingley K, Kronick JB, Potter BK, Chan AK, Coyle D, Dodds L, Dyack S, Feigenbaum A, Geraghty M, Gillis J, Rockman-Greenberg C, Khan A, Little J, MacKenzie J, Maranda B, Mhanni A, Mitchell JJ, Mitchell G, Laberge AM, Potter M, Prasad C, Siriwardena K, Speechley KN, Stockler S, Trakadis Y, Turner L, Van Karnebeek C, Wilson K, Chakraborty P; Canadian Inherited Metabolic Diseases Research Network. *Metabolic Clinic Atlas: Organization of Care for Children with Inherited Metabolic Disease in Canada*. *JIMD Rep* 2015; 21: 15-22.
24. Rogers J, Reed MP, Blaine K, Manning H. Children with medical complexity: A concept analysis. *Nurs Forum* 2021; 56: 676-683.
25. Hoogveen IJ, Peeks F, de Boer F, Lubout CMA, de Koning TJ, Te Boekhorst S, Zandvoort RJ, Burghard R, van Spronsen FJ, Derks TGJ. A preliminary study of telemedicine for patients with hepatic glycogen storage disease and their healthcare providers: from bedside to home site monitoring. *J Inherit Metab Dis* 2018; 41: 929-936.
26. Lampe C, Dionisi-Vici C, Bellettato CM, Paneghetti L, van Lingen C, Bond S, Brown C, Finglas A, Francisco R, Sestini S, Heard JM, Scarpa M; MetabERN collaboration group. The impact of COVID-19 on rare metabolic patients and healthcare providers: results from two MetabERN surveys. *Orphanet J Rare Dis* 2020; 15: 341.
27. Ronis SD, Grossberg R, Allen R, Hertz A, Kleinman LC. Estimated Nonreimbursed Costs for Care Coordination for Children With Medical Complexity. *Pediatrics* 2019; 143: e20173562.
28. Boy N, Mühlhausen C, Maier EM, Ballhausen D, Baumgartner MR, Beblo S, Burgard P, Chapman KA, Dobbelaere D, Heringer-Seifert J, Fleissner S, Grohmann-Held K, Hahn G, Harting I, Hoffmann GF, Jochum F, Karall D, Konstantopoulous V, Krawinkel MB, Lindner M, Märtner EMC, Nuoffer JM, Okun JG, Plecko B, Posset R, Sahn K, Scholl-Bürgi S, Thimm E, Walter M, Williams M, Vom Dahl S, Ziağaki A, Zschocke J, Kölker S. Recommendations for diagnosing and managing individuals with glutaric aciduria type 1: Third revision. *J Inherit Metab Dis* 2023; 46: 482-519.
29. Welling L, Bernstein LE, Berry GT, Burlina AB, Eyskens F, Gautschi M, Grünewald S, Gubbels CS, Knerr I, Labrune P, van der Lee JH, MacDonald A, Murphy E, Portnoi PA, Öunap K, Potter NL, Rubio-Gozalbo ME, Spencer JB, Timmers I, Treacy EP, Van Calcar SC, Waisbren SE, Bosch AM; Galactosemia Network (GalNet). International clinical guideline for the management of classical galactosemia: diagnosis, treatment, and follow-up. *J Inherit Metab Dis* 2017; 40: 171-176.
30. van Wegberg AMJ, MacDonald A, Ahring K, Bélanger-Quintana A, Blau N, Bosch AM, Burlina A, Campistol J, Feillet F, Gizewska M, Huijbregts SC, Kearney S, Leuzzi V, Maillot F, Muntau AC, van Rijn M, Trefz F, Walter JH, van Spronsen FJ. The complete European guidelines on phenylketonuria: diagnosis and treatment. *Orphanet J Rare Dis* 2017; 12: 162.
31. Forny P, Hörster F, Ballhausen D, Chakrapani A, Chapman KA, Dionisi-Vici C, Dixon M, Grünert SC, Grünwald S, Haliloglu G, Hochuli M, Honzik T, Karall D, Martinelli D, Molema F, Sass JO, Scholl-Bürgi S, Tal G, Williams M, Huemer M, Baumgartner MR. Guidelines for the diagnosis and management of methylmalonic acidemia and propionic acidemia: First revision. *J Inherit Metab Dis* 2021; 44: 566-592.

32. Van Calcar SC, Sowa M, Rohr F, Beazer J, Setlock T, Weihe TU, Pendyal S, Wallace LS, Hansen JG, Stembridge A, Splett P, Singh RH. Nutrition management guideline for very-long chain acyl-CoA dehydrogenase deficiency (VLCAD): An evidence- and consensus-based approach. *Mol Genet Metab* 2020; 131: 23-37.
33. Albokhari D, Ng BG, Guberinic A, Daniel EJP, Engelhardt NM, Barone R, Fiumara A, Garavelli L, Trimarchi G, Wolfe L, Raymond KM, Morava E, He M, Freeze HH, Lam C, Edmondson AC. ALG8-CDG: Molecular and phenotypic expansion suggests clinical management guidelines. *J Inherit Metab Dis* 2022; 45: 969-980.
34. Altassan R, Radenkovic S, Edmondson AC, Barone R, Brasil S, Cechova A, Coman D, Donoghue S, Falkenstein K, Ferreira V, Ferreira C, Fiumara A, Francisco R, Freeze H, Grunewald S, Honzik T, Jaeken J, Krasnewich D, Lam C, Lee J, Lefeber D, Marques-da-Silva D, Pascoal C, Quelhas D, Raymond KM, Rymen D, Seroczynska M, Serrano M, Sykut-Cegielska J, Thiel C, Tort F, Vals MA, Videira P, Voermans N, Witters P, Morava E. International consensus guidelines for phosphoglucomutase 1 deficiency (PGM1-CDG): Diagnosis, follow-up, and management. *J Inherit Metab Dis* 2021; 44: 148-163.
35. Čechová A, Altassan R, Borgel D, Bruneel A, Correia J, Girard M, Harroche A, Kiec-Wilk B, Mohnike K, Pascreau T, Pawliński Ł, Radenkovic S, Vuillaumier-Barrot S, Aldamiz-Echevarria L, Couce ML, Martins EG, Quelhas D, Morava E, de Lonlay P, Witters P, Honzik T. Consensus guideline for the diagnosis and management of mannose phosphate isomerase-congenital disorder of glycosylation. *J Inherit Metab Dis*. 2020; 43: 671-693.
36. <https://www.simmesn.it/it/societa/gruppi-di-lavoro/371-gruppo-intrasocietario-di-dietetica-e-nutrizione.html>
37. Stockler-Ipsiroglu S, Potter BK, Yuskiv N, Tingley K, Patterson M, van Karnebeek C. Developments in evidence creation for treatments of inborn errors of metabolism. *J Inherit Metab Dis* 2021; 44: 88-98.
38. Singh RH, Brown SJ, Hale PM, Narlow K, Gurung S, Salvatore ML, Tchamako JK. Using Informatics to Build a Digital Health Footprint of Patients Living With Inherited Metabolic Disorders Identified by Newborn Screening. *J Public Health Manag Pract* 2022; 28: E340-E344.