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ADOLESCENT PERCEPTIONS OF HEALTHCARE SERVICES FOR INHERITED METABOLIC DISORDERS AND GENETIC DISEASES: INSIGHTS FROM A SURVEY IN A PEDIATRIC HOSPITAL

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***ABSTRACT* – Objective:** To evaluate adolescent perceptions of healthcare services for Inherited Metabolic Disorders (IMDs) and genetic diseases in a tertiary pediatric hospital, identifying key elements for tailoring care to this unique patient group.

Patients and Methods: A survey comprising 29 questions, adapted from a validated tool, was distributed to adolescent patients (aged 12–18 years) with IMDs or genetic disorders at the Pediatric Hospital "Giovanni XXIII" of Bari (Italy). Data collection occurred between January and June 2022. Key domains included communication and information, facility quality, waiting times, visit durations, and illness experience. Descriptive statistics were employed, and statistical significance was set at $p<0.05$.

Results: Among 40 eligible participants, 27 completed the survey (response rate: 67.5%). Satisfaction was highest in communication and information (69.2%) and illness experience (69.8%), whereas waiting times received the lowest ratings (55.3%). Facility quality was rated positively overall (64.7%), with suggestions for improvements in personal spaces and age-appropriate facilities. More than half of respondents advocated for the inclusion of psychologists and patient association representatives in the care team. Notably, 62.3% supported teleconsultations, and 70.4% endorsed social media for improving disease management. Satisfaction levels did not significantly vary by age or hospital proximity.

Conclusions: Adolescents generally perceive hospital services positively but highlight areas for improvement, including reduced waiting times, expanded multidisciplinary care, and the introduction of telemedicine. Addressing these needs could enhance engagement and ensure adolescent-friendly healthcare services in IMD management.

Keywords: Metabolic disorders, Adolescence, Hospital management, Healthcare services.

List of Abbreviations: HEEADSSS: Home, Education and Employment, Eating, peer-related Activities, Drugs, Sexuality, Suicide/depression, and Safety from injury; IMD: Inherited Metabolic Disorder; MetabERN: European Reference Network for Hereditary Metabolic Disorders; SD: Standard Deviation.

INTRODUCTION

Inherited Metabolic Disorders (IMDs) result from enzymatic deficiencies caused by genetic defects in biochemical pathways. Consequently, toxic substrates or metabolites may accumulate, while essential products may become deficient, leading to complications in organs, such as the liver, brain, heart, muscle, and kidneys. These complications may manifest at various stages of life, either early or later in development[1].

Comprehensive diagnosis and treatment include regular and careful monitoring and follow-up throughout the patient's lifetime, necessitating several accesses in the hospital, both in inpatient and outpatient settings.

Advances in newborn screening and therapies have enabled earlier diagnosis and treatment, allowing many patients to survive into adulthood[2].

Adolescence is a particularly complex period marked by cognitive, physical, emotional, and social maturation[3].

In the case of clinical conditions, such as IMDs and genetic diseases, "healthcare assistance" faces an additional challenge for adolescents as they shift from parental care to independent self-management. This period is often accompanied by feelings of anxiety and inadequacy, which may lead to decreased compliance and disengagement from treatment. The healthcare assistance, in this period of life, needs to take into consideration psychosocial development, including the ability to consolidate identity, achieve independence, and establish adult relationships. To address these challenges, individualized support should be provided by specialized centers through multidisciplinary teams comprising clinicians, dietitians, and psychologists[4].

Guidelines emphasize the importance of patient-centered care that addresses adolescents' unique psychosocial needs. The HEEADSSS psychosocial interview framework—covering the domains of Home environment, Education and employment, Eating, peer-related Activities, Drugs, Sexuality, Suicide/depression, and Safety from injury – serves as a valuable tool for assessing adolescents' mental, emotional, and social health[5].

Despite these recommendations, adequate services, including standardized operating procedures and specific training for adult physicians specializing in IMDs, remain underdeveloped in Europe. A survey conducted by Stepien et al[6] among all 77 centers of the European Reference Network for Hereditary Metabolic Disorders (MetabERN) revealed that most respondents were metabolic pediatricians, and only approximately 40% had received formal training in managing adolescent metabolic patients. Moreover, compared to primary care settings, the quality of specialized assistance provided to hospitalized adolescent patients has been poorly documented. This study aims to evaluate perceptions of healthcare services of adolescent patients affected by IMDs and genetic diseases, referring to a tertiary care pediatric hospital. The goal is to identify key elements useful for delivering appropriately tailored care to adolescent patients.

PATIENTS AND METHODS

An online survey with 29 questions (see **Supplementary Material**) was distributed to patients from the Department of Metabolic and Genetic Disorders of the Pediatric Hospital "Giovanni XXIII" in Bari (Italy) between January and June 2022. Patients aged between 12 and 18 years were included.

The survey was based on a previously validated questionnaire[7], which comprises five sections, including general information and four areas investigated: communication and information, facility quality, waiting times and the duration of visits and their illness experience.

Patients were contacted by phone up to two times. Those who accepted to participate in the survey signed informed consent and were asked to complete a survey on the Google Forms platform by receiving a QR code via email. Patients independently completed the questionnaire.

Ethical approval was obtained from the Independent Ethics Committee of the Policlinico University Hospital of Bari, Italy (Prot. No. 0063763).

Statistical analysis

The responses were converted into scores (see **Supplementary Material**) and analyzed using descriptive statistics, with data presented as rating percentages (total score obtained/total score max) and averages (mean ± standard deviation). The chi-square test was used for nominal variables, and the Student's *t*-test was applied for continuous variables. A $p<0.05$ was considered statistically significant. Data analysis was carried out using the software Stata MP17 (StataCorp LLC, College Station, Texas, USA).

RESULTS

General information

Out of 40 patients recruited, 27 completed the survey (response rate 67.5%). Thirteen out of 27 (48.1%) patients were aged 12–13 years, 4/27 (15%) were 14–15 years old and 10/27 (37%) were 16-17 years. Ten were female (37%), 17 were male (63%). Of the 27 patients, 3 (11.1%) had a genetic disorder, while the remaining 24 (88.9%) were affected by an IMD. Among patients with IMDs, 18 (75%) were affected by PKU, one had urea cycle disorder (OTC deficiency), one was affected by hereditary fructose intolerance, two by cystinuria and two by hereditary lipid disorders (sitosterolemia and cholesteryl storage disorder).

Six out of 27 patients (22%) reported living <15 km as a distance between their residence and the hospital, 6/27 (22%) reported a distance between 15 km and 30 km, while 15/27 (55%) of respondents live >30 km distant from the hospital.

In the Information and Communication category, patients provided favorable feedback, with a rating of 69.2%, achieving a total score of 467 out of a maximum of 675 points. This corresponds to a mean score of 17.30 ± 2.76 on a scale ranging from 0 to 25. The Experience of the Illness category showed a rating of 69.8%, a total score of 226 out of 324, and a mean score of 8.37 ± 1.76 within a range of 0 to 12. The Quality of the Structure received a moderately positive evaluation, with a rating of 64.7%, a total score of 297 out of 459, and a mean score of 11.00 ± 1.84 on a scale from 0 to 17.

The waiting times category received a lower rating of 55.3%, with a total score of 269 out of 486 and a mean score of 9.96 ± 2.16 within a range of 0 to 18. For the facility quality category, a rating of 64.7%, with a total score of 269/486 and a mean score of 11.00 ± 1.84 within a range of 0–17, was reported. The category concerning the illness experience received a rating of 69.8% based on a total score of 226/324 and was associated with a mean score of 8.37 ± 1.76 on a scale of 0 to 12.

The overall satisfaction rating was 64.7%, with an aggregate score of 1259 out of 1944 points, translating to a mean total score of 46.63 ± 6.01 on a scale of 0 to 72. The scores assigned to individual questions, response categories, and the overall score were consistent across age groups and the distance from their residence to the hospital (p>0.05) (see **Supplementary Material**). Rating distribution according to areas investigated is summarized in Table 1. A more detailed description of responses for each question in each category is summarized in Table 2.

Information and Communication

Patients expressed a high level of satisfaction with aspects related to communication and the provision of information. The information received about their health status was well-rated, with a mean score of 2.44 ± 0.51, corresponding to 81.5% of the maximum score. Similarly, the collaboration among staff received positive feedback, scoring 2.26 ± 0.59 (75.3%). Patients felt welcomed by the healthcare staff, reflected in a score of 3.04 ± 0.85 (75.9%) and rated positively the answers provided when asking questions about their health or proposed treatment (2.33 ± 0.62, 77.8%). Respect for privacy was also favorably evaluated, with a score of 2.41 ± 0.50 (80.2%). Patients gave lower scores about being left alone with the doctor during visits (score 1.15 ± 0.86, 38.3%) and about their involvement in scheduling medical visits, rated at 1.81 ± 0.83 (60.5%). The impact of hospital visits on free-time activities, such as homework, hobbies, or socializing, also scored moderately at 1.85 ± 0.91 (61.7%).

Table 1. Rating distribution according to areas investigated.

	Percentage of the maximum score	Total Vote/Max	Expected score range	Mean ± SD
Information and Communication	69.2%	467/675	0-25	17.30 ± 2.76
Waiting times	55.3%	269/486	0-18	9.96 ± 2.16
Facility quality	64.7%	297/459	0-17	11.00 ± 1.84
Illness experience	69.8%	226/324	0-12	8.37 ± 1.76
Overall satisfaction	**64.7%**	**1259/1944**	**0-72**	**46.63 ± 6.01**

Table 2. Survey results for each question of areas investigated.

Area	Question	Expected score	Rating (%)	Mean ± SD
Information and Communication	How do you rate the information you received about your health status?	From 0 to 3	81.5%	2.44 ± 0.51
	How did you find the collaboration among the ward staff?	From 0 to 3	75.3%	2.26 ± 0.59
	How welcomed do you feel by the healthcare staff when you come to this hospital?	From 0 to 4	75.9%	3.04 ± 0.85
	How do you rate the answers provided to you when asking questions about your health or the proposed treatments?	From 0 to 3	77.8%	2.33 ± 0.62
	How do you evaluate the respect for your privacy when personal information is provided?	From 0 to 3	80.2%	2.41 ± 0.50
	How much would you like to be left alone with the doctor during the visit?	From 0 to 3	38.3%	1.15 ± 0.86
	How involved do you feel in deciding the timing of medical visits at the hospital?	From 0 to 3	60.5%	1.81 ± 0.83
	To what extent do medical visits interfere with your free time activities (homework, going out with friends, hobbies, sports, etc.)?	From 0 to 3	61.7%	1.85 ± 0.91
Waiting times	How do you evaluate the frequency of your hospital visits?	From 0 to 3	71.6%	2.15 ± 0.46
	How did you find the waiting time between entering the hospital and the medical visit?	From 0 to 4	41.7%	1.67 ± 0.68
	How did you find the duration of medical visits at the hospital?	From 0 to 4	48.1%	1.93 ± 0.38
	How did you find the waiting time between the end of the visit and discharge?	From 0 to 4	52.8%	2.11 ± 0.75
	How did you find the duration of hospitalization/ therapy (day service/day hospital)?	From 0 to 3	70.4%	2.11 ± 0.58
Facility quality	How suitable does the hospital environment seem for your age?	From 0 to 4	57.4%	2.30 ± 0.61
	How do you rate the organization of spaces dedicated to managing metabolic diseases?	From 0 to 3	67.9%	2.04 ± 0.59
	How do you evaluate the presence of children around you in this hospital?	From 0 to 3	79.0%	2.37 ± 0.49
	How did you find the spaces dedicated to you (e.g., phone charging stations, free seating, privacy for phone calls, etc.)?	From 0 to 4	61.1%	2.44 ± 0.85
	How do you feel about the possibility of being treated at an adult hospital?	From 0 to 3	61.7%	1.85 ± 0.66
Illness experience	How involved did you feel in decisions regarding your therapy and follow-up appointments?	From 0 to 3	76.5%	2.30 ± 0.54
	Do you feel capable of managing your illness and its therapy independently?	From 0 to 3	63.0%	1.89 ± 0.70
	How important do you consider the treatment of your condition?	From 0 to 3	92.6%	2.78 ± 0.42
	How limiting do you find your condition in your daily life?	From 0 to 3	46.9%	1.41 ± 1.01

Waiting times

While the frequency of hospital visits was rated positively at 2.15 ± 0.46 (71.6%), the waiting time before medical visits scored 1.67 ± 0.68 (41.7%), and the duration of medical visits, rated at 1.93 ± 0.38 (48.1%). The waiting time between the end of the visit and discharge received a moderate score of 2.11 ± 0.75 (52.8%), while the duration of hospitalization/therapy was rated at 2.11 ± 0.58 (70.4%).

Facility quality

Patients considered the environment suitable for their age, scoring 2.30 ± 0.61 (57.4%), and rated the organization of spaces dedicated to metabolic disease management at 2.04 ± 0.59 (67.9%). The presence of children in the hospital was also positively rated (2.37 ± 0.49, 79%). Personal spaces, such as areas for phone charging and privacy, were evaluated at 2.44 ± 0.85 (61.1%). The idea of being treated in an adult hospital scored comparatively lower at 1.85 ± 0.66 (61.7%).

Illness experience

Patients expressed a strong sense of the importance of their care and treatment, with the perceived importance of treating their condition scoring 2.78 ± 0.42 (92.6%). Involvement in decisions regarding therapy and follow-up appointments was also rated positively, with a score of 2.30 ± 0.54 (76.5%). Autonomy in managing their condition independently was rated moderately at 1.89 ± 0.70 (63.0%), and the perceived limitations of their illness on daily life scored relatively low at 1.41 ± 1.01 (46.9%).

Furthermore, concerning the question "Which professional figure would you add to the current follow-up team?", for 44.4% of patients (12 individuals), the current follow-up team, consisting of a physician, nurse, and dietitian, meets their needs. A total of 55.6% of adolescents (15 individuals) view the addition of "another professional figure" to the team positively. Specifically, 37% of patients (10 individuals) would find it useful to expand the team to include a representative from the Rare Metabolic Diseases Association, while 44.4% of patients (12 individuals) would like the inclusion of a psychologist. Seven patients (25.9%) consider the support of both professionals beneficial for the follow-up team. The results are summarized in Table 3. Responses did not differ by age ($p>0.05$), whereas greater satisfaction with the composition of the follow-up team was reported by patients living farther from the hospital ($p>0.05$) (see **Supplementary Material**).

Finally, the possibility of introducing remote teleconsultations in addition to in-person visits was supported by 62.3% of participants (17/27). Furthermore, 70.4% (19/27) believed that better use of social media could improve disease management both within and outside the hospital. The responses to these questions did not vary by age or distance from the hospital ($p>0.05$) (see **Supplementary Material**).

DISCUSSION

By evaluating perceptions of healthcare services in terms of communication and information, facility quality, waiting times, duration of visits and illness experience, this survey has identified key elements for delivering appropriately tailored care to adolescent patients with IMDs.

Table 3. Distribution of responses to the question: "Which professional figure would you add to the current follow-up team?"

Response	n	Percentage (%)
None	12	44.4%
Would add only a Representative from the Rare Metabolic Diseases Association	3	11.1%
Would add only a psychologist	5	18.5%
Would add both	7	25.9%

This study revealed that while adolescent patients generally appear satisfied with the care received in a pediatric hospital, there is considerable room for improvement to better meet their needs. Patient satisfaction levels align with findings from other studies[7,8]. However, it has been demonstrated that patient satisfaction and their perception of care do not always correspond to high-quality care delivery[9].

Adolescents expressed a particular appreciation for communication and information. This finding highlights that the attention given to these aspects is positively regarded and valued by this patient group. Such attention likely enhances compliance with the provided care, making it easier for adolescents and their parents to return for follow-up visits[10].

However, evaluations of waiting times and visit durations were less favorable. Specifically, less than half of the patients (41.7%) considered the waiting time between entering the hospital and the start of the medical visit to be acceptable, while only 48% deemed the duration of the visit appropriate. These findings highlight an opportunity to improve patients' and parents' perceptions of care quality. It is well-established that shorter waiting times enhance adherence to medical visits, especially among adolescents, who are generally less inclined to wait. Reducing waiting times could encourage greater engagement in medical care[11].

Another important area of focus is the composition of the follow-up team, currently consisting of a physician, nurse, and dietitian. More than half of the adolescent patients expressed dissatisfaction, preferring the inclusion of additional professional figures, such as a representative from a patient association, a psychologist, or both. Adolescence is a particularly sensitive period for psycho-physical development, which becomes even more complex for individuals with chronic illnesses. Generally, adolescents are reluctant to seek psychological support. Therefore, the expressed need for psychological support by patients in this hospital – contrary to the general trend – is a critical point that warrants further exploration to deliver high-quality care[12].

The possibility of introducing remote teleconsultations in addition to in-person visits was supported by 62.3% of patients, while 70.4% considered the use of social networks essential for improving disease management both within and outside the hospital. Telemedicine services are currently the subject of studies confirming improved patient access to care, though it remains unclear whether patients are satisfied with telemedicine[13]. Since this service is not currently provided to pediatric or adolescent patients, the data suggest that adolescents would be willing to engage in teleconsultations. Whether these meet their expectations was not investigated. By implementing this service for pediatric hospital patients, future studies could assess whether and to what extent patients are satisfied with telemedicine services. Additionally, such research could determine whether both adolescent patients and their parents appreciate remote visits and follow-ups.

These results indicate that adolescent patients generally perceive the pediatric hospital positively, though there is considerable room for improvement. As noted, perceived quality does not always equate to actual care quality. While the hospital environment was deemed welcoming and appropriate in most areas investigated, certain needs, particularly regarding waiting times and the composition of the follow-up team, were not fully met.

Although this study focuses solely on the care provided to adolescents in a hospital in Bari (Italy), it is reasonable to hypothesize that similar perceptions might exist in other hospitals across Italy. Given the generally uniform nature of the national healthcare system, there are many similarities among hospitals in service delivery. Furthermore, most healthcare professionals receive similar training and are likely to provide comparable standards of care.

CONCLUSIONS

Despite the generally positive feedback obtained from this survey, analyzing adolescent patients' perceptions of the quality of care provided has offered valuable insights for improving the quality of care. Reducing waiting times, providing psychological support, and implementing telemedicine services appear to be key areas for enabling our hospital to become a truly "adolescent-friendly" facility.

Artificial intelligence-assisted technologies:

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

Authors' contributions:

Conceptualization, original draft preparation, writing and review of the manuscript, A.T.,A.S; resources and data curation, G.M., D.G.D., V.A, A.M.; review and editing, R.D., L.M. All authors have read and agreed to the published version of the manuscript.

Availability of data and material:
All data generated or analyzed during this study are included in this published article or its supplementary material.

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

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