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COMMUNICATION CHALLENGES IN NEWBORN SCREENING: WHAT THE PUBLIC KNOWS (AND DOESN'T)

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***ABSTRACT* – Objective:** Extended newborn screening (ENBS) is a worthwhile public health program for the early detection of inherited metabolic disorders, facilitating timely diagnosis and treatments that have significantly decreased the morbidity and mortality associated with these conditions. Despite its importance, public awareness of ENBS remains limited. All families deserve access to clear, accurate, and relevant information to help them navigate the newborn screening system. To assess whether the level of knowledge and perceptions of NBS among the general population are appropriate, a public engagement initiative was conducted.

Subjects and Methods: A public engagement initiative was realized in the city of Naples (Italy) to involve passers-by in the completion of a survey titled "Extended neonatal screening – how much do you know?". The questionnaire was distributed as a paper-based or online form, including 15 items focused on demographics and knowledge of ENBS, with the option to share personal experiences and feedback on the dissemination activity.

Results: We interacted with approximately 600 people, and half completed the questionnaire. The study population was heterogeneous in terms of sex, age, and educational background, with around half belonging to the biomedical/healthcare field. Globally, only 33% of respondents were aware of ENBS. Despite this, appreciation of the outreach activity and the materials provided was high regardless of the knowledge attained.

Conclusions: The data collected in this study reveal a concerning lack of awareness, particularly among those who have already had children. These findings indicate that sporadic recognition or outreach efforts, although beneficial, are insufficient on their own. More sustained and structured schemes are required within clinical settings, where healthcare personnel should be deliberately trained and supported to enhance education and communication with parents.

Keywords: Expanded newborn screening, Metabolic disorders, NBS, Survey, Knowledge gap, Communication challenge.

INTRODUCTION

Newborn screening (NBS) is one of the most effective public health strategies for the early identification of inherited genetic and metabolic disorders[1-4]. Empowering timely diagnosis and early treatments can help prevent the progression of such severe diseases, as leaving them untreated often leads to brain damage, multi-organ failure, or even death[5]. In fact, NBS has significantly ameliorated the prognosis and quality of life through an organized approach that coordinates the monitoring and care of patients applying significant

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surveillance activities[6]. The application of NBS was pioneered by Robert Guthrie, who in 1959 developed a bacterial inhibition assay to detect elevated phenylalanine levels using dried blood spots (DBS). This approach marked the beginning of the first NBS program in the United States, providing a successful proof of concept for early diagnosis of phenylketonuria (PKU). This allowed prompt treatment of pre-symptomatic neonates with the possibility to prevent permanent damages[7]. The panel of pathologies examined through NBS has gradually increased over the years. In the late 1990s, the introduction of tandem mass spectrometry (MS/MS) into NBS laboratories revolutionized the clinical paths, allowing comprehensive, fast, sensitive and accurate quantification of metabolite markers in body fluids[8]. MS/MS enabled the screening for aminoacidopathies (including PKU), organic acidemias, fatty acid oxidation defects, and urea cycle defects, leading to the modern concepts of expanded newborn screening (ENBS)[9-11]. The history of neonatal screening in Italy began with Legislative Decree No. 104 of 1992, which introduced the screening for three diseases: congenital hypothyroidism (CH), cystic fibrosis (CF), and PKU. Subsequently, thanks to scientific progress and over 20 years of practice, it was possible to extend neonatal screening activities to many more inherited metabolic diseases. The panel of diseases subjected to ENBS was established in 2016 (Law 167 of 19 August 2016 and subsequent Ministerial Decree of 13 October 2016) and currently includes over 40 metabolic disorders. Then, following the Prime Ministerial Decree of 12 January 2017, ENBS became part of the Essential Assistance Levels (LEA) and was made mandatory across the entire Italian territory. The ENBS program at the regional or interregional level is a system divided into the following main functional structures, namely: neonatal screening laboratory, laboratory for biochemical diagnostic confirmation, laboratory for genetic diagnostic confirmation, regional reference clinical centers, and regional coordination center of the screening system. The structure and functions of the current Italian organization have been described by Ruoppolo et al[12]. Effective implementation of ENBS not only requires a robust technical infrastructure but also relies on informed parental engagement, especially during critical phases such as post-screening communication and follow-up. Despite its proven clinical and societal benefits, awareness and understanding of NBS among the general population remain limited, particularly outside biomedical contexts. For this reason, increasing public knowledge is crucial.

The psychosocial impact of receiving unexpected newborn screening results or unsolicited genetic information represents a major concern in ENBS, especially in the case of positive results. There is an additional consideration that screening tests can generate false-positive results, inducing strong psychosocial effects on parents and families. However, research suggests that parental stress and anxiety can be significantly mitigated through better parental education about ENBS, improvement of the communication schemes adopted, personalized genetic counseling, and proactive developmental guidance[13,14]. Positive tests, confirmed diagnoses, false positives, or variants of uncertain significance (VUS) can cause significant psychosocial distress, anxiety, confusion, and feelings of helplessness[13,15]. In cases of false positives or VUS, this distress is often intensified by uncertainty, as parents struggle to interpret ambiguous health information and worry about potential future risks[15]. Studies[14,16] have shown that such responses can lead to increased parental stress, altered perceptions of child vulnerability and, in some cases, long-term effects on the parent-child relationship. Notably, even when follow-up testing confirms the absence of disease, lingering concerns and altered parenting behaviors may persist[17,18]. To reduce the psychosocial burden, tailored interventions have been proposed, including early and empathetic communication from trained healthcare professionals, ideally following structured counseling protocols that anticipate emotional reactions[19]. Providing clear, balanced, and jargon-free explanations of screening results, supported by visual or written materials, can enhance understanding and reduce fear[20]. Incorporating psychosocial support by genetic counselors or psychologists and ensuring continuity of care through follow-up consultations may alleviate parental anxiety[21]. Moreover, it is essential to determine the appropriate timing for delivering information, as educating parents on this subject can enhance information retention and contribute to better care for both children and parents[22]. Improved parental education about newborn screening, effective communication strategies, individualized genetic counseling, and anticipatory guidance can mitigate unexpected communications associated with diseases with a non-acute neonatal onset, rendering diagnosis acceptance for families less difficult[13,23].

This work aimed to assess public awareness and attitudes toward ENBS, exploring informational needs and opinions of people from Naples, Italy. By collecting survey responses, we sought to identify knowledge gaps and opportunities to improve communication strategies related to neonatal screening practices. At present, in fact, the implementation of effective educational initiatives is hindered by fragmented communication practices, restricted counseling time during perinatal care, limited resources, and a siloed newborn screening system, all of which create barriers to effective educational experience. Engaging families, especially those from historically underserved communities, is essential to increase knowledge, build confidence, and ultimately improve outcomes for children and their families[24].

SUBJECTS AND METHODS

To evaluate the degree of public knowledge about ENBS and awareness of its relevance within the national healthcare system, a survey was delivered among passers-by in the most frequented areas of the city of Naples (Campania, Italy) during the Christmas days (22-24 December 2025). The interviews were conducted through questionnaire distribution, after which informational material was provided to participants to offer detailed screening-related information. Individuals were invited to participate in the survey regardless of their level of expertise, ensuring the inclusion of both informed and non-specialist respondents. Participants who consented to take part in this initiative were provided with a questionnaire that could be completed either on paper or online, according to their preference. The survey instrument was developed by the authors and comprised 15 questions aimed at collecting demographic information as well as assessing respondents' knowledge and awareness of ENBS. The full list of questions proposed during the survey is reported below, while their relative multiple-choice answers are reported in the Supplementary Material.

Questionnaire on the "Extended neonatal screening – how much do you know?"

1. What is your sex?
2. How old are you?
3. What is your level of education?
4. Do you work in the biomedical/healthcare field?
5. Did you know about neonatal screening?
6. Have you ever heard of any of the diseases included in neonatal screening?
7. Do you have children?
8. At the time of childbirth, have you received information about neonatal screening?
9. Do you have a family member/acquaintance with a hereditary metabolic disease?
10. If yes, was it identified by neonatal screening or subsequently?
11. How do you rate this outreach activity?
12. Indicate your appreciation of the dissemination material.
13. What did you learn during this outreach activity?
14. Do you have any questions about extended neonatal screening?
15. If you like, tell us about your experience with extended newborn screening.

Questionnaires were collected directly by the authors, and all data were handled in accordance with ethical standards, ensuring anonymity and confidentiality. All subjects provided written informed consent for inclusion in the study and anonymous treatment of the data. Indeed, no personally identifiable information was recorded, stored, or processed at any stage of the study. The contribution to this survey was voluntary, and participants were adequately informed about the initiative prior to questionnaire administration. After data collection, all responses were aggregated into a single matrix dataset. Questionnaires were screened for completeness and internal consistency. Incomplete questionnaires, defined as those with more than 20% missing responses, were excluded from further analysis. For questionnaires with a limited number of missing items, available data were retained without imputation, given the descriptive nature of the study, and labeled as NS (Not Specified).

Statistical Analysis

The finalized dataset was subsequently analyzed using descriptive statistical methods[25], including frequency distributions, percentages, and summary measures, to characterize respondents' demographic features and levels of awareness related to ENBS. Statistical analyses and graphical representations were performed using GraphPad Prism software version 10 (San Diego, CA, USA)[26].

RESULTS

A public engagement initiative was carried out in the city of Naples with the aim of assessing the level of knowledge of ENBS within the general population and identifying potential communication challenges affecting its understanding and perception. Through the administration of a structured questionnaire to passers-by, the initiative allowed the collection of quantitative and qualitative data on participants' demographic characteristics, awareness and parental knowledge of neonatal screening practices, and perceptions of the outreach activity itself.

Demographic Characteristics of the Survey Participants

The questionnaires were compiled by 298 people, representing the 51% of the passers-by (total of 583) (Figure 1A), and all related data on their demographic characteristics are detailed in Table 1. People were invited to complete the survey through a paper modality or *via* an online form, with the majority of them choosing the paper modality (62%) (Figure 1B). Globally, the mean response rate to the full questionnaire was 99%, with only sporadic missing responses (1%), mainly involving demographic information. The population that responded to the survey showed a prevalence of female subjects (66%) (Figure 1C). However, it was well distributed in terms of age ranges, which were divided into three groups: young people (under 25 years old), middle-aged adults (between 25 and 40 years old), and older adults

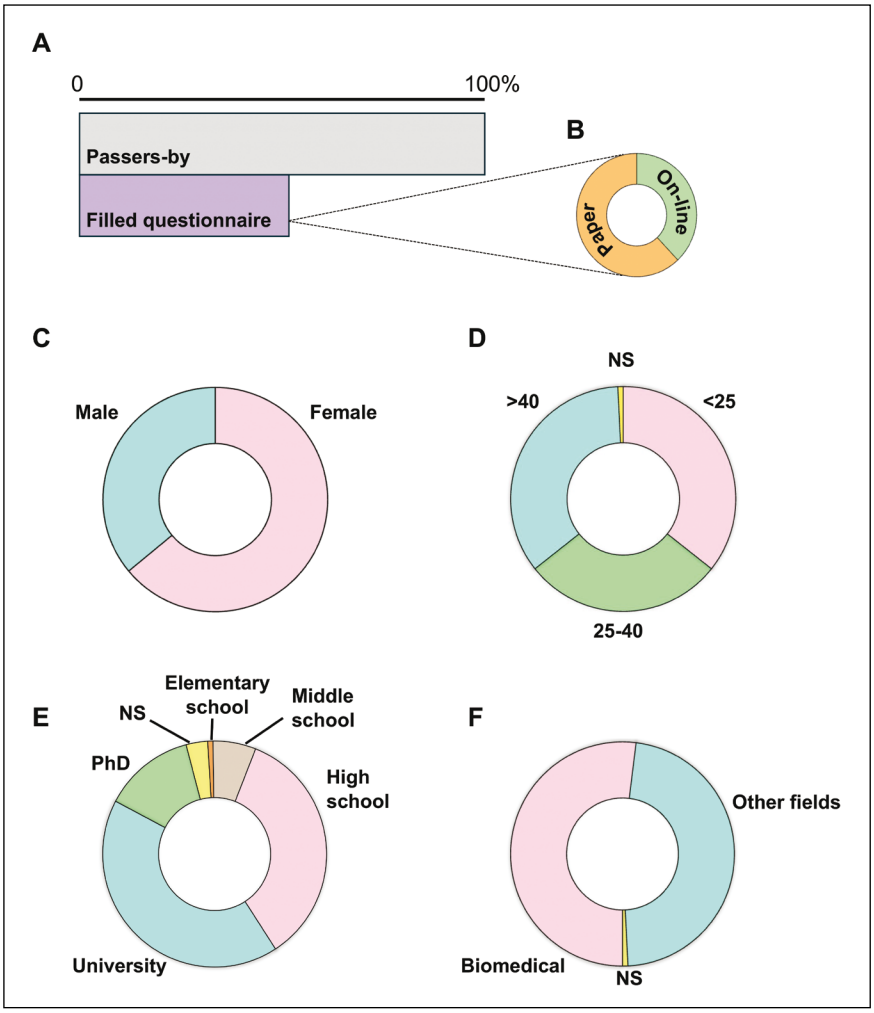

Figure 1. Descriptive analysis of demographic and educational characteristics of the population of respondents. **A**, Distribution of survey respondents over the total of passers-by. **B**, Chosen modality of compilation of the survey. Distribution of the population of respondents according to (**C**) sex, (**D**) age range, (**E**) level of education, and (**F**) professional occupation. NS=Not Specified.

Table 1. Demographic characteristics of the survey participants.

Sex	Males (34%)	Females (66%)				
Age	<25 (36%)	25-40 (28%)	>40 (35%)	NS (1%)		
Education	PhD/ Specialization (12%)	University degree (42%)	High school (35%)	Middle school (6%)	Elementary school (2%)	NS (3%)
Work	Biomedical field (47%)	Others (52%)	NS (1%)			

NS=Not Specified.

(over 40 years old). The 1% did not declare their age (NS, not specified) (Figure 1D). A heterogeneous distribution was observed in terms of educational background: 42% of the participants held a university degree and only 12% had a higher title (PhD or specialization), 35% completed high school, 6% concluded middle school, and 2% of the participants stopped their studies during elementary education. Finally, 3% of respondents did not indicate their level of education (NS) (Figure 1E). The professional occupation of respondents also showed a good level of diversity. Nearly half of them (47%) reported working in the biomedical or healthcare field, while 52% indicated that they did not work in this sector (Figure 1F).

Awareness of Newborn Screening in the General Population and Parental Gaps

The data collected from responses revealed a significant lack of awareness regarding ENBS among the general population. In our survey, in fact, to the question "Did you know about neonatal screening?" 62% of respondents replied "No", demonstrating that they were unaware of ENBS procedures. An additional 5% did not respond to this question (NS), resulting in a minority of individuals (33%) who reported understanding this crucial health initiative (Figure 2A). Considering the heterogeneity of passers-by with relation to their educational and professional background, this lack of knowledge was notably higher among individuals without professional experience in biomedical/healthcare fields (80% of "No" respondents) or among subjects with no high academic qualifications (50% of "No" respondents) (Figure 2B). Finally, people without both a university degree and a biomedical occupation corresponded to 43.3% of "No" respondents (Figure 2B), indicating a positive correlation between educational/professional background and knowledge of ENBS. Among the total interviewees, survey data reveal that 36% of respondents declared to have children (Figure 2C). However, within this group, it emerged that more than half of these parents were unaware that neonatal screening had been performed on their child at birth (Figure 2D).

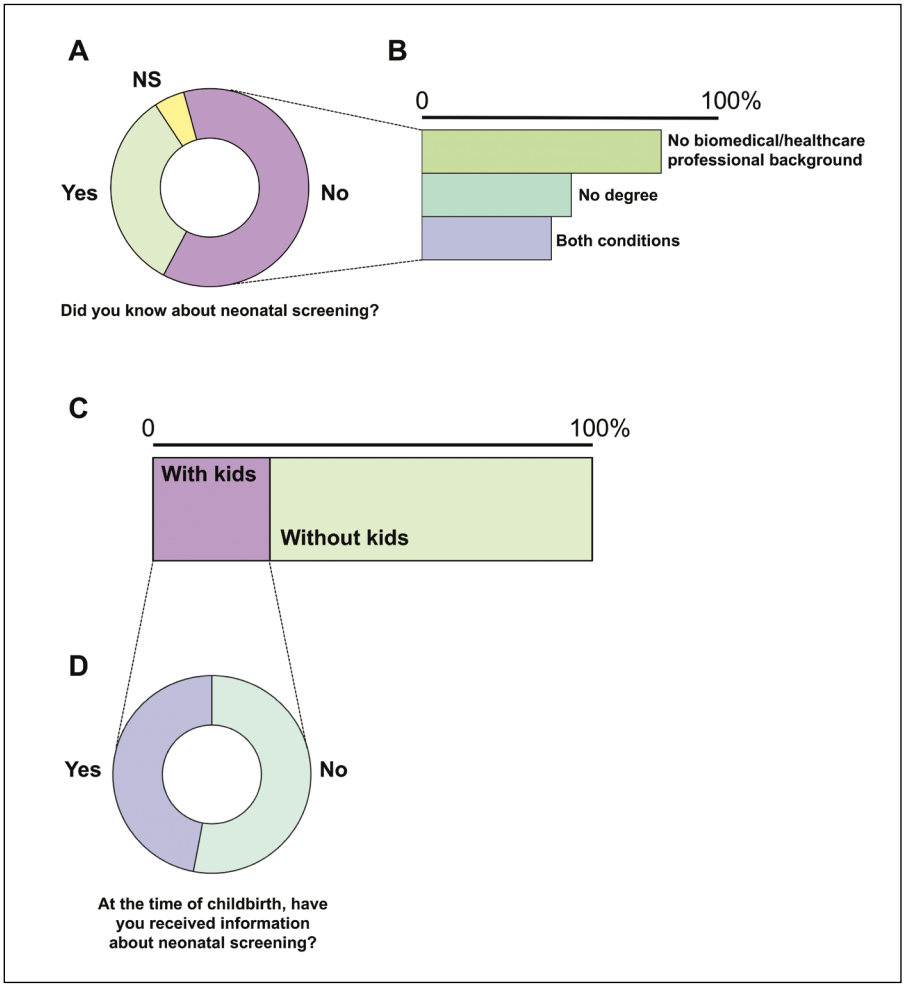

Figure 2. Distribution of respondents to the main questions about ENBS knowledge. **A**, Distribution of respondents to the question "Did you know about neonatal screening?" and (**B**) percentage of "No" respondents according to their professional and educational background. **C**, Respondents reported having or not having children. **D**, Parent awareness about ENBS testing performed at the time of childbirth. NS=Not Specified.

Participant Feedback and Emerging Questions

A specific need to gain more insight into ENBS emerged from questions 11 and 12 regarding appreciation of the outreach activity, and from some emblematic replies collected in the final open-ended questions 13-15. Public feedback gathered during our health outreach activity is summarized in Figure 3. When asked to rate the initiative, most participants gave the highest rating on a 1-to-3 scale, with the vast majority choosing "3" (Figure 3A). Similarly, when asked about the dissemination material provided, most participants selected "5" on a 1-to-5 scale, indicating high satisfaction (Figure 3B). These results suggest that the initiative was well-received and that the materials used were effective at engaging the public. Open-ended responses provided valuable insight into what participants gained from the outreach activity and what information they still want to get. Some subjects discovered the possibility of diagnosis at birth and prevention in early childhood. A few of them showed interest in the coverage of disorders that can be screened, emphasizing how this information had changed their perception of newborn care. Other participants asked for clarification on which tests are mandatory and automatically performed at birth, *vs.* those that are optional or require parental consent.

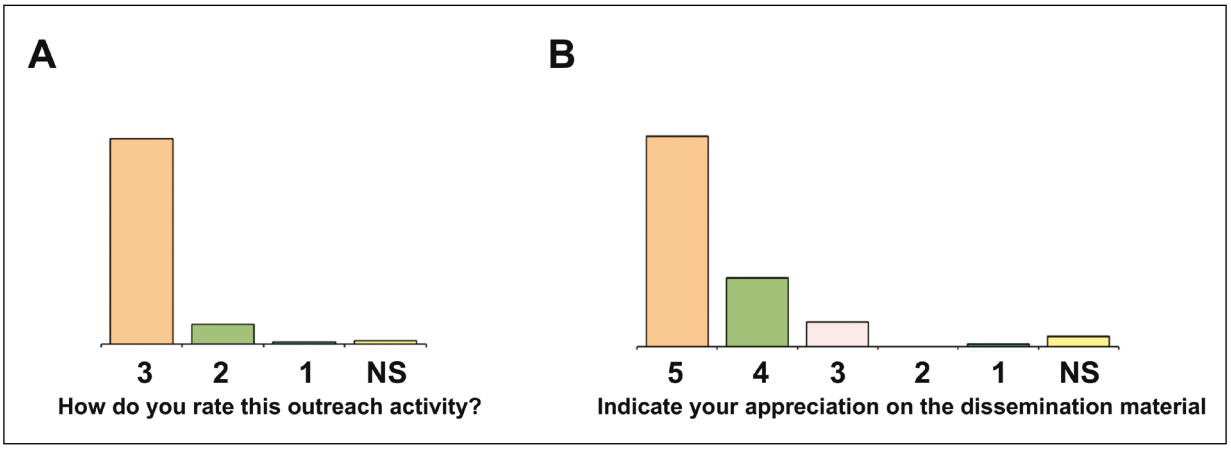

Figure 3. Appreciation rating of participants to the survey about (**A**) the proposed health outreach activity (1-to-3 scale) and (**B**) the dissemination material distributed (1-to-5 scale). NS=Not Specified.

DISCUSSION

The results presented below provide an overview of the surveyed population, highlighting key gaps in awareness, as well as emerging needs for improved information and communication strategies related to ENBS.

This study provides real-world evidence of a substantial gap in public awareness. Despite the high participation rate, around two-thirds of respondents reported no prior knowledge of neonatal screening. While the demographic profile of the surveyed population was relatively heterogeneous, our findings require attention as a considerable proportion was represented by individuals with higher education and professional experience in the biomedical or healthcare field. This aspect, likely related to the voluntary nature of participation and the interest in health-related topics, may have introduced a selection bias and should therefore be considered a limitation of the study. Nevertheless, a substantial lack of awareness regarding ENBS was still observed, suggesting that knowledge within the general population may be even lower than that detected in our cohort.

Literature and historical experience in public health, particularly in preventive screening, clearly show how socio-economic and educational inequalities can influence the understanding and accessibility of health programs, including newborn screening[24,27]. Accordingly, our results corroborate these achievements, highlighting that individuals with lower educational levels or from a different professional occupation are less informed. This underscores the urgent need for more effective and accessible educational strategies to raise awareness about the value of newborn screening and its impact on child health[28,29].

A concerning result that emerged from this survey is the limited awareness among parents of the newborn screening procedures performed on their own children. This gap is alarming as parents' understanding can significantly influence their engagement in follow-up care and interventions necessary for their child's health[30]. Furthermore, adequate information at the time of childbirth or before it may help the communication with the clinical staff in case of recall or notification of a positivity to one of the diseases included in the program. Parents who are insufficiently informed may experience confusion or anxiety in the event of a recall or a positive screening result, potentially compromising trust in healthcare providers and adherence to follow-up pathways. Informed parental engagement is a key component of effective screening programs, as it facilitates timely communications and continuity of care, guaranteeing full engagement in child healthcare[31].

Notwithstanding, an encouraging finding was represented by the high level of appreciation expressed by participants toward the outreach initiative and the dissemination of materials. Participants expressed the need to be better informed, especially before or during hospitalization for childbirth, to be more conscious regarding the health of their newborns or potential risks. The positive feedback indicates that public engagement activities can be effective tools for increasing interest, awareness, and understanding of newborn screening[32]. These reflections highlight the value of knowledge spreading, but also underscore the necessity for clear, accessible, and proactive communication within healthcare settings.

CONCLUSIONS

This study collected data from surveys administered to passers-by in the most frequented areas of the city of Naples with reference to ENBS to quantify the degree of public knowledge. Our initiative included several valuable components: the presence of experts available to answer questions, brochures on ENBS (including lysosomal storage disorders), and informational flyers. These elements offered a comprehensive overview and allowed for personal engagement with the public. The responses to the questionnaire "Extended neonatal screening – how much do you know?" made it clear that more structured communication strategies are needed. Informing the public should not rely solely on events or printed materials. Training on ENBS also needs to be strengthened within healthcare facilities. Nurses and clinical staff involved in the neonatal screening process need to be adequately trained to provide parents with clear and early guidance[33]. Given the crucial importance of these tests for a child's health, early communication must be both informative and empathetic, addressing parents' questions and emotional needs from the very first interaction. This approach is instrumental in preparing families for potential follow-up steps, such as recall notifications or additional testing, to minimize distress and reduce possible harm to children and their families[34]. Indeed, parental reactions are closely linked to the quality of communication they receive throughout the screening process. While the outreach activity was successful in terms of public satisfaction, the persistent knowledge gap highlights the need for ongoing, more integrated communication strategies to guide parents and families through subsequent steps with confidence and clarity.

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Study conception and design: M. Costanzo, S. Bianco, M. Ruoppolo; collection and interpretation of data: all; statistical analysis: M. Costanzo, S. Bianco; manuscript drafting: all; manuscript editing: all; approval to submit: all.

Availability of Data and Material:
All data generated or analyzed in this study are included in the main article and its Supplementary Material.

Conflict of Interest:
The authors have no conflicts of interest to disclose.

Ethics Approval and Informed Consent:

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