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Parents' experience of the communication process of positivity at newborn screening for metabolic diseases: A qualitative study

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Abstract

Background: The process of receiving a communication of positivity for metabolic diseases at expanded newborn screening (ENBS) is extremely articulated, involves a variety of actors (parents, maternal and child departments, clinical centres and laboratories) and is open to a variety of outcomes from false positive to true positive cases. Receiving communication of positivity can be highly stressful for parents and requires an adequate communication process to give clear and reliable information without causing excessive worry. This qualitative study describes the parents' experience of receiving a communication of positivity to metabolic diseases at ENBS, and their assessment of the quality of the communication process and steps, with the main aim to identify the process' strengths and weaknesses and to advance tailored recommendations to improve the communication process.

Method: Fourteen in-depth, semi-structured phone interviews were conducted with parents whose children resulted positive to the ENBS. As part of the ENBS communication process, parents received a first phone call communication of positivity and a second in-person communication at metabolic clinical centres (MCC). The framework analysis method was used to organize the data and identify emerging themes.

Results: Parents were largely dissatisfied with the quality and depth of the information received and with the way the healthcare staff delivered the first communication phone call, which failed to create a caring, empathic and safe setting. Many parents tried to reduce the uncertainty by searching online information or consulting with other providers. Nevertheless, the majority of parents described the in-person visit at MCC as clear, welcoming and reassuring.

Conclusion: More efforts are needed to improve the quality of the communication process of the ENBS. Guidelines, recommendations and standard scripts to

communicate positivity are needed along with programmes and educational resources to train tailored communication skills.

KEYWORDS

communication of positivity, communication process, expanded newborn screening, information needs, parents' experiences

1 | INTRODUCTION

Expanded newborn screening (ENBS) for inherited metabolic diseases is one of the most advanced tools in precision medicine and an important public health programme. The programme is aimed to screen all newborns to provide early diagnosis of genetic diseases so that an effective treatment can be promptly started to prevent the long-term consequences of inherited metabolic disorders (Ruoppolo et al., 2022).

In 2020, 404 892 children were born in Italy and nearly 17 000 parents were informed of their child's positive newborn bloodspot ENBS results with nearly 1000 receiving a positive result to the second confirmatory test.

The collection of the dried bloodspot to perform ENBS is made between 48 and 72 h of life, and the timing of the communication can vary between 4 and 15 days after birth, depending on the tested conditions and according to the national guidelines.

The ENBS in Italy includes 47 conditions, but a periodic extension is considered every 3 years following the scientific evidence available and a discussion involving all the stakeholders.

According to Chudleigh and colleagues (Chudleigh et al., 2021), the communication of positive results should be described as a process more than an event, as the variability of diagnostic paths can be extremely high, ranging from false positive results to a clear diagnostic confirmation. Between these two extreme points, there are many intermediate situations that can require a genetic testing for the family, a temporary monitoring of the child's clinical situation or a conclusion of the ENBS process. Although new strategies to reduce the number of false positive cases are sought, there is a demanding need to consider the communication aspects (Malvagia et al., 2020).

Previous studies have shown that communication of ENBS positivity can be stressful for parents (Hayeems et al., 2016; O'Connor et al., 2018; Schmidt et al., 2012), but they have mainly focused on false positives and the psychosocial impact of the content of the communication, not on the communication process itself.

Recently, Conway and colleagues (Conway et al., 2022) made a summary of the recommendations available in the literature to improve the quality and effectiveness of communication in the ENBS process (Buchbinder & Timmermans, 2012; Chudleigh et al., 2016; Davis et al., 2006). Nevertheless, these recommendations cannot be generalized to all countries and healthcare settings but needed to be adapted to specific cultures and organizational aspects of the ENBS process. Other studies focused on positive

Key messages

- The positivity to metabolic disease screening is a stressful event, and parents' reactions are related to the quality of the communication received.
- Parents described the first contact via a phone call as the most critical node of the communication process.
- Parents reported a good quality of communication received at in-person visits at metabolic clinical centres.
- Recommendations on standards script to communicate positivity and training for healthcare providers are advanced.

ENBS results or on specific diseases such as sickle cell hemoglobinopathy or cystic fibrosis (Beucher et al., 2010; Farrell et al., 2020; Salm et al., 2012; Tluczek et al., 2009) and advanced practical communication tips for healthcare providers including to be calm, avoid jargon, listen carefully, encourage questions, recognize parental distress, offer realistic reassurance, assess parents' understanding and refer to specialists in case of need. However, data on positive results for metabolic diseases are scant (Davis et al., 2006), and little is known about how providers communicate and should communicate to parents the results of a provisional positivity that can later be confirmed (true positive), partially modified (healthy carrier) or disconfirmed (false positive). This becomes particularly relevant when multiple healthcare providers are involved in the communication process. Considering this huge variability and the deep impact that communication of an ENBS positive result can have on families, it is extremely important that such communication is timely, clear and tailored to the parents' needs.

In Italy, ENBS is offered free of charge, and to guarantee the maximum homogeneity of application of the legislation (Legge 19 agosto, 2016; Ministero della Salute, Decreto 13 ottobre, 2016) throughout the national territory, a co-ordination centre for ENBS was established at the National Institute of Health.

Despite the national and local efforts to standardize the ENBS process, a deep variability remains in the communication process with parents. The communication process is highly articulated as it involves families, maternal and child departments (MCD), metabolic clinical centres (MCC) and the clinical laboratory and requires trained professionals to communicate to parents the child's positive screen and the need to start a clinical path at MCC.

To date, in Italy, there are 19 laboratories for the screening (about 33 000 average test/year/laboratory with a coverage of 100%) and 15 MCC; in the six regions without a laboratory equipped to process the ENBS samples, analyses are regulated by inter-regional agreements. Despite the national and local efforts to standardize the ENBS process, a deep variability remains in the communication process with parents. In some regions, the ENBS test results are communicated directly by the MCC that will later meet the families to confirm positivity, whereas in other regions, the MCD call the families to programme a second confirmatory test when needed or to schedule a visit at MCC. In this national context, Lombardy is the most populated Italian region (nearly 10 million inhabitants), with 59 MCC where 68 000 children are born each year representing nearly 20% of the total national births. The ENBS process in the region includes one regional laboratory – which perform both screening and diagnostic testing for all newborns – and three clinical centres.

The result communication process has been defined according to a regional protocol (Legge 19 agosto, 2016) and differentiates between three categories of ENBS positivity:

- Negative: No other action is required; the families do not receive communication.
- Low risk: The MCD, in collaboration with the MCC, communicate by phone the test positivity to the family and schedule a clinical appointment at MCC.
- Intermediate and high risk: Activation of the urgent clinical procedure can also involve neonatal intensive care units; the MCC call by phone the families.

Considering the complexity of the communication process and focusing on the Lombardy region process (Figure 1), this qualitative study aims to describe parents' experience with the communication of the ENBS positive results and their perception of the quality of the

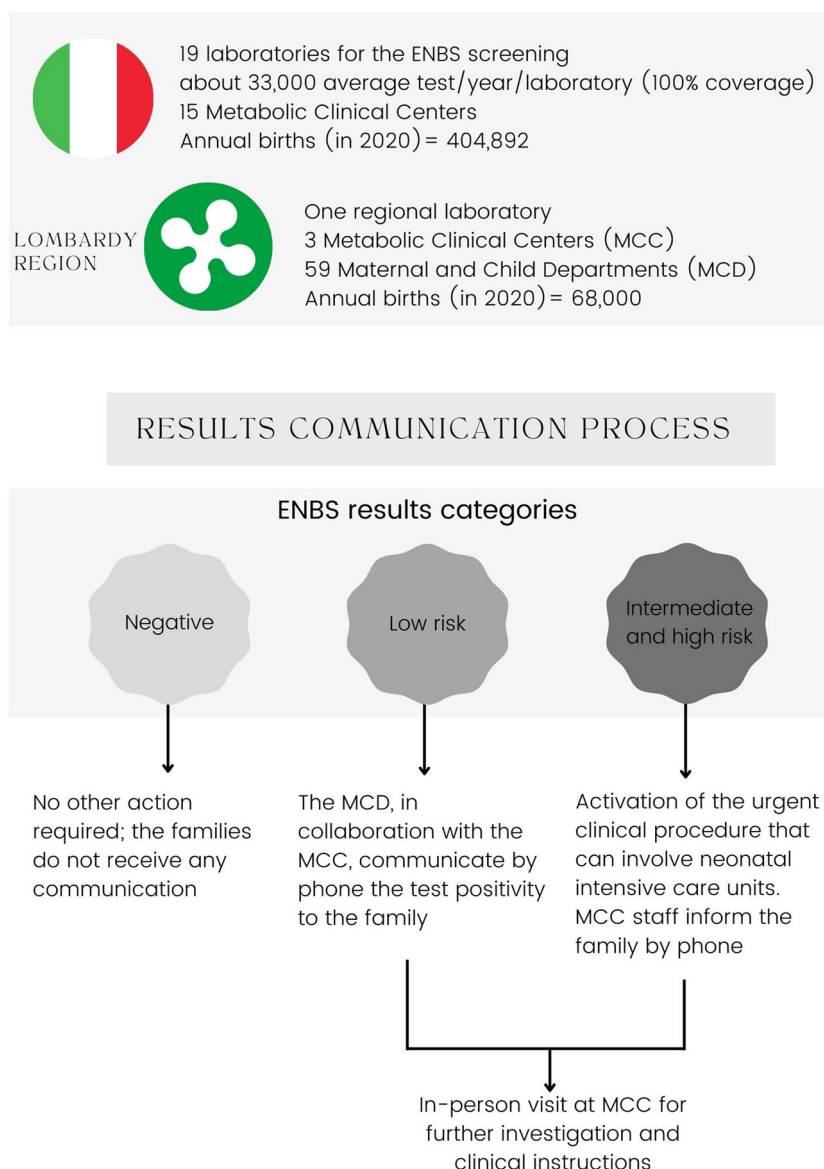


FIGURE 1 Expanded newborn screening (ENBS) network and communication process in Lombardy region.

communication process. These data will serve to identify strengths and weaknesses of the communication process, unveil unmet needs and advance recommendations to guide a functional communication and maximize the efficacy of the communication process.

In particular, we wanted to describe parents' emotional and behavioural reactions to the ENBS positive screen in the two-step communication process currently in place in the healthcare system of Lombardy. Parents' information needs and sources of information were also investigated. Drawing from the study, data recommendations to improve the communication process of ENBS positive results were advanced.

2 | METHODS AND STUDY SAMPLE

2.1 | Study design, participants and procedure

As part of a wider longitudinal in-progress study, 80 parents who received communication of positivity for metabolic diseases at ENBS were contacted for taking part in a qualitative study on the impact of ENBS test positivity on parental distress. Three to 6 months after the first phone test results communication, parents received an email asking for their availability to be contacted by phone for an interview focusing on the experience of the test result communication received. The temporal window between the first test result communication and the interview was selected in such a way to allow parents to detach emotionally from the experience to describe it but close enough to recall the details of the communication.

The interviews – conducted between August 2020 and October 2021 – took about 15 min, were conducted by the same interviewer (not involved in the clinical process) and were audio-recorded. Written informed consent was obtained from all parents who agreed to participate in the study, and also, oral informed consent was recorded before starting interviews. This study was approved by the Ethical Committees of the three MCC involved (no. 2955/2019).

2.2 | Data collection and analysis

Semi-structured interviews included questions focused on the description of the first communication (phone call) received by MCD (about the need to perform a second confirmatory test) and the communication received at the first in-site visit by MCC (confirmation of positivity or false positive result) both in terms of contents provided and communicational approach, as well as the perceived need of searching information after the communications. The semi-structured interview content was based on a topic guide (Table 1) adapted from a previous study on parents' experience with results disclosure (Collins et al., 2013).

De-identified audio-recordings were transcribed verbatim, and framework analysis (Dixon-Woods, 2011; Gale et al., 2013) was applied to organize the data and identify emerging themes. The

TABLE 1 Interview topic guide.

Topic guide
Demographics information
Quality of the first result communication from the MCD (content, professional involved, relational approach)
Reactions to the first result communication (emotional reaction, coping strategies, information seeking)
Quality of the second result communication from the MCC (content, professional involved, relational approach)
Reactions to the second result communication (emotional reaction, coping strategies, information seeking)
Preferences and suggestion to improve the communication process

framework for the analysis was developed by two researchers (ER and MB) and further reviewed by a third researcher (SR). The coding framework was then systematically applied to the data and concurrently modified and refined. One investigator (ER) summarized themes and supporting quotes from each transcript with discussion of key areas with the research team. The framework was examined within and across themes to identify overarching themes and relationships. Data were organized and summarized with an excel matrix, and the framework, findings and interpretation of the analysis were discussed and approved by all co-authors.

2.3 | Participants' characteristics

Seventeen parents replied, giving their availability, and received a copy of the written informed consent (to be returned once signed), the topics covered during the interview and possible date for the interview. Three parents did not reply after the initial contact, whereas 14 provided their consent and completed the interviews and were included in the analysis.

Twelve mothers, one father and one mother–father dyad completed the interviews; the mean age of interviewees was 35.57 ± 4.15 years. All were Italian and married, 12 had a bachelor's or master's degree, one completed high school and one finalized Grade 8. For eight participants, the child who resulted positive to the ENBS was the firstborn in the family, and four participants already had children (Table 2).

3 | RESULTS

Parents' responses were clustered around three main themes, namely, 'communication process, content and information understanding', 'emotional reaction and information seeking' and 'unmet needs and suggestions'. As the content of the themes is strongly different and greatly polarized for the two communication steps, they are presented separately in Sections 3.1 and 3.2.

Recommendations emerging from the data on how to improve the communication process are also reported.

TABLE 2 Participants' characteristics and clinical information.

No.	Clinical sites	Length interview (min)	Parental role	Age	Metabolic disease suspected	Confirmation testing
1	Site 2	19.30	Mother	35	Low-risk CIT I/II	False positive
2	Site 2	11.47	Mother	31	Low-risk GA2	False positive
3	Site 2	12.42	Mother	37	Low-risk OAs	Ongoing follow-up for OAs
4	Site 2	16.42	Mother	44	BTD	False positive
5	Site 1	13.15	Mother	39	GALT	False positive
6	Site 2	30.43	Mother	34	SCAD	False positive
7	Site 2	18.52	Mother	42	Low-risk BTD	False positive but carrier of a pathogenic mutation
8	Site 2	15.01	Mother	34	HPI, B12 deficiency B12, OAs	Ongoing follow-up for HPI
9	Site 1	15.16	Mother	34	Low risk of (VLCADD)	VLCADD healthy carrier
10	Site 1	15.42	Mother	34	BTD	Partial BTD
11	Site 3	17.41	Mother	28	PKU	PKU
12	Site 1	32.08	Mother	33	MCD	Healthy carrier 3-MCCD
13	Site 3	41.45	Mother and father	36/36	PKU	PKU
14	Site 1	16.46	Father	37	MCAD	MCAD

Abbreviations: BTD, biotinidase deficiency; CIT I-II, citrullinemia; GALT, galactosemia; HPI, hyperprolinemia type I; MCAD, medium-chain acyl-CoA dehydrogenase; MCCD, 3-methylcrotonyl-CoA carboxylase deficiency; OAs, organic acidemias; PKU, phenylketonuria; SCAD, short chain acyl-CoA dehydrogenase; VLCADD, very long-chain acyl-CoA dehydrogenase deficiency.

3.1 | The experience and reactions to the phone call

3.1.1 | Communication process, content and information understanding

Participants reported that the phone call was made by different professionals (in eight cases by a paediatrician/neonatologist, in two cases by a nurse and in two cases by a midwife), but in two cases, parents did not recall the qualification of the professional. For the majority of parents (11 out of 14), it was a healthcare professional they never met before, and for the three of them, it was a member of the MCC staff.

Large variability in the content of the phone call was reported by participants. Nevertheless, half of the participants claimed that the phone call received by the MCC was 'quite generic' and did not include either the name of the suspected disease or the test parameter:

I was told my daughter tested positive for a rare disease in the newborn screening but she couldn't tell anything else. (P12_Mother, 33 years old)

Other participants reported that they were informed about a low-grade abnormal result, but they did not get to know either the test values or the name of the condition under scrutiny:

We were told the newborn screening showed were weak positive results, I remember she said 'weak' and

that there was no need to worry. (P2_Mother, 31 years old)

Some participants reported not to be informed about the value but only informed about the name of the suspected disease:

I was told my son tested positive for galactosemia, a rare disease that could cause serious damage to the kid and so more in-depth analysis needed to be made. (P5_Mother, 39 years old)

Only one participant reported to be clearly informed about both the test value and the name of the suspected disease during the first call:

She gave me details about the specific value and the type of disease. (P1_Mother, 35 years old)

3.1.2 | Emotional reaction and information seeking

All of the participants (14) spontaneously described their emotional reaction to the phone call. The most reported emotional responses were a feeling of 'puzzlement', anxiety, panic, alarm, disorientation and uncertainty.

Although some participants (3) reported experiencing a milder concern ('I wasn't nervous, I was concerned because of the abnormal value but I was rather calm' (P3_Mother, 37 years old), the majority of

participants reported experiencing a great concern in response to the phone calls:

We panicked, we totally panicked. (P6_Mother, 34 years old)

We took it really hard, I think I have been cringed for one hour and a half and had palpitations. (P12_Mother, 33 years old)

Searching the web for information was the most common action following the phone call. Some participants revealed that Internet-retrieved information was a source of emotional relief:

We searched for latest research and we found helpful papers; we searched for experts' opinions by looking for modern medical luminaries in the USA and we found some scientific articles that helped us to calm down. (P7_Mother, 42 years old)

On the contrary, for others, Internet-retrieved information was either an additional source of worry or not helpful as a calming strategy.

We were left to ourselves and so we went on Google, which I know is wrong, and on the Internet I obviously found unpleasant things, therefore I had a moment of discomfort. (P10_Mother, 34 years old)

Another common reaction to the phone call communication (reported by four participants) was looking for medical advice from other health professionals (including family doctors, paediatricians and relatives who work in the healthcare system), and very few subjects called back to the MCC to ask for more information.

3.1.3 | Unmet needs and suggestions

Most participants reported the need for a more detailed phone call communication that focuses on what the abnormal test values mean, with some of the participants wishing they would have communicated more details such as the name of the condition under scrutiny. For example, one participant reported that they wished they knew the precise test value and the name of the suspected disease and reported that the person who called

was not able to tell me neither the type of disease nor the test value. (P10_Mother, 34 years old)

Some participants suggested to complement the call with written information sent by email:

It would be nice if they sent a pdf guide, so that families may have written information to rely on. (P13_Mother&Father, 36 years old)

Nearly half of the participants wished that the person who called was more 'trained', namely, more informed about the coming steps in the diagnostic and therapeutic process that follow the visit at the MCC:

She had no idea of what was going to expect us (at the clinical center). (P9_Mother, 34 years old)

Some participants would have preferred to be called directly by the clinic centre where experts on metabolic diseases work, whereas very few revealed the preference for receiving the phone call communication from a doctor, rather than by a midwife.

I wish I was called directly by the clinical center, so that I could ask any questions I wanted. (P4_Mother, 44 years old)

Some participants complained about the excessive alarmism generated by the phone call, when results were not confirmed.

3.2 | The experience and reaction to the intake visit at the MCC

3.2.1 | Communication process, content and information understanding

Almost all participants positively described the experience of the in-person visit at the MCC, reporting that the information received was enlightening and clear. Participants also described the approach of the healthcare staff as welcoming and kind:

the clinical center gave us a sense of humanity, kindness, and courtesy that is extraordinary. It was such a pleasure to meet the doctors, they were very kind and they explained us the matter in a very gentle way. (P5_Mother, 39 years old)

Every doctor was willing to solve our doubts with simple and clear words. (P9_Mother, 34 years old)

Only one participant negatively described the communication process of the visit at the clinic as

I wasn't heard, my needs were not met, they were focused on future steps I had to make. (P12_Mother, 33 years old)

3.2.2 | Emotional reaction and information seeking

For many participants, the visit at the MCC was reassuring as it added relevant information for parents helping them to have a better understanding of the future trajectory of the clinical situation (for both true and false positivity results):

They kind of reassured me telling me what the future of my child could be. (P10_Mother, 34 years old)

The communication was good, they conveyed serenity and did not make you anxious. (P3_Mother, 37 years old)

The doctor at the clinical center told me exactly what I wanted to hear since the beginning of the process: she said 'these are the screening results, this is the abnormal value, that's the first and that's the second step' and she explained what the consequences could be. (P8_Mother, 34 years old)

When describing their reaction to the in-person visit, three participants revealed that they perceived a reduction of uncertainty, 'If on the phone I had the same information I had during the visit I wouldn't have lived in doubt and uncertainty for weeks', and a sense of serenity, 'after leaving the clinical center I felt more serene' (P1_Mother, 35 years old).

On the other hand, some participants perceived that the intake visit was not reassuring because they kept feeling uncertain and worried after the visit.

After the first visit at the MCC, many participants searched for online information. A small portion of participants sought medical advice from other healthcare professionals.

3.2.3 | Unmet needs and suggestions

In order to improve the in-person visit experience at the MCC, many participants focused on organizational aspects that could be improved, whereas some participants suggested changing communication aspects.

Organizational aspects to improve included the desire for a shorter time between the first communication of the ENBS results (phone call) and the scheduled appointment at the MCC. Furthermore, they expressed the need to know in advance details related to the visit, such as how long it takes and what it implies.

I didn't know what to expect from the visit, I was told I had to go to the clinical center to do some tests but the visit turned out to be a day hospital. (P4_Mother, 44 years old)

We wish we knew what was going to happen at the clinical center. (P9_Mother, 34 years old)

Another organizational aspect mentioned as possible improvement was the collaboration and communication between the MCD and the MCC. Participants advanced that they should work in synergy in order to make the communication process clearer and more effective:

The maternal and child department and the clinical center should communicate and share information in order to improve organization. (P9_Mother, 34 years old)

Nearly half of the participants expressed the desire to liaise with only one healthcare stakeholder, and others would have preferred to be called directly by the MCC rather than being contacted first from the MCD and later attend a visit at the MCC. Others would have preferred to liaise with the MCC exclusively and attend the in-person visit there. The need for a reference point had also risen: One participant even suggested building a network of families that share the same experience.

Some parents focused on aspects of communication that could be improved. Those parents were the ones reporting anxiety linked to the intake visit. Some of them reported that anxiety was related to a lack of understanding. One participant advocated for doctors to keep a plain and simple language during the intake visit, and another participant suggested complementing the visit with paper-based information.

They use a technical language that was difficult to follow if you have not studied medicine. I wish they used a simpler and concrete language. (P3_Mother, 37 years old)

The communication was fully verbal. As a parent, you feel anxious, and because of that, I missed some pieces of what was being said, I also wish I could record them. If I had that information written on paper, I could return to it afterwards with a clearer mind. (P7_Mother, 42 years old)

For one of the participants who experienced anxiety during the intake session, the aspect to be improved was not the content of the communication but the relational and emotional dimensions of the communication as the intake visit was not perceived as reassuring:

It's ok to be direct rather than sweetening the pot, but they could be more gentle. (P12_Mother, 33 years old)

TABLE 3 Recommendations derived from the study data.

1. Stronger co-ordination should be established between MCD and MCC, and the connection of the two centres should be communicated to parents;
2. The time window between the first phone call communication and the in-person visit at MCC should be as short as possible;
3. In case of delay in receiving the laboratory results or in scheduling the in-person visit, parents should be regularly contacted and updated;
4. Healthcare staff of the MCD who deliver the first phone call communication of positive results should preferably be known by parents;
5. If a known staff member is not available to deliver the first phone call, the communication should be done by another MCD staff member adequately instructed to follow a standardized script including details about the type of positivity and the possible following steps for families (including different scenarios);
6. Professionals of MCD and MCC who communicate positive results should use clear and appropriate language with parents and avoid medical jargon;
7. Parents should be warned about anxiety-provoking information on ENBS and metabolic disorders freely available online and they should be provided with scientifically sound resources designed for the general public;
8. Leaflets and online resources should be available (at least at a national level) and easily accessible for parents who receive communication of positive results at ENBS;
9. Online resources should offer scientifically sound information tailored to different audiences and therefore include sections using easy-to-understand language designed for a not technical public;
10. After the conclusion of the ENBS communication process, parents of children who received false-positive results or do not need future monitoring should be offered the opportunity to contact MCC if doubts or other needs arise.

Abbreviations: ENBS, expanded newborn screening; MCC, metabolic clinical centres; MCD, maternal and child departments.

3.3 | How to improve the communication process: The recommendations

A set of 10 recommendations based on the study data and emerging themes are advanced and reported in Table 3. Three recommendations (1, 2 and 3) refer to more operational and management aspects of the ENBS communication process, namely, developing stronger co-ordination between MCD and MCC, lessening the waiting time between the first test result communication and the in-person visit and how to manage communication in case of delay in laboratory testing. Three recommendations (4, 5 and 6) are strongly linked to the necessity to standardize and control the information delivery process with the two-fold aim to maximize parents' understanding and minimize their emotional distress. Recommendations 7, 8 and 9 pertain to parents' information searching behaviours and the possible resultant emotional distress and respond to parents' information-related unmet needs emerged from the study. Recommendation 10 is relevant to establishing a communication channel for families who received false positive results to use in case of future needs. Furthermore, recommendations 4 and 10 leverage the relational aspect between parents

and healthcare staff to engage parents and help them in navigating the system.

4 | DISCUSSION

This study provides important information on parents' experience of the communication of positive ENBS results for metabolic diseases. The results highlighted that parents' experience of in-person visits at MCC was extremely positive in terms of creating a perception of a caring, empathic and safe setting and clarity of information received. However, the majority of parents identify the phone call received by the MCD as the most critical node of the communication process characterized by a lack of clarity and which often generates uncertainty and negative emotional reactions.

The need for more information arose regardless of the quality and quantity of information received during the phone call and during the intake visit. Not only those who declared to have received little and generic information by phone desired to know more, but also the only participant who was clearly informed about the actual abnormal value results and about the name of the condition claimed the need for communication to be more detailed. Regarding the in-person visit, even when participants judged it informative and the information received was clear, they reported to have searched for more information online afterwards.

In our sample, parents complained about the quantity and the quality of the information received during the phone call that was described as too vague, increasing the level of parental anxiety without the possibility to have more information until the in-person visit at MCC. Another concern reported by parents was linked to the familiarity with or the professional role of the healthcare staff member who called them; the possibility of being called by the neonatologist or by a known member of the MCD could have reduced the impact of the phone call communication. Previous studies have shown that satisfaction with the communication increases when the professional is specialized (Brockow & Nennstiel, 2019). Nevertheless, it has to be considered that the availability of neonatologists is limited, and because of organizational factors, the call may be done by team members, who may not be trained to communicate ENBS positivity or without all the needed information to deliver a clear communication. The temporal window between the phone call from the MCD and the in-person visit at the MCC was highly variable (ranging from 1 day to 2 weeks), leaving parents in a state of uncertainty that often generated anxiety and led them to search for more information or seek the assistance of other healthcare providers.

In line with previous studies (Dillard et al., 2010), parents sought information mainly online, and the most contacted healthcare professionals were the family paediatricians. No parents reported being directed to institutional websites where scientifically sound and reliable information on ENBS for the general public is available, confirming the results of previous studies (DeLuca et al., 2012) and highlighting the need to develop scientifically sound and trustworthy online resources. Furthermore, parents reported that they would have

welcomed the possibility to contact a healthcare professional trained and specialized in ENBS or professionals or body institutions that could have provided further information on ENBS. No such contacts are currently provided by the organizations involved in the ENBS.

Overall, the high satisfaction reported by parents after the in-person visit at MCC leans towards a good quality of the communication provided by clinicians. Nevertheless, a few parents' information needs remained unmet after the in-person visit such as a clearer communication about the need for future visits (in case of false positive cases or confirmed diagnosis of mild conditions that did not require therapy and follow-up) and written information materials. This aspect can be particularly relevant when communicating false positive results (DeLuca et al., 2011; Morrison & Clayton, 2011). Furthermore, few parents expressed the preference for having a copy of the health records even in case of false positive cases as 'they are difficult information to remember'.

The study findings are coherent with those reported in previous studies on UK and US parents receiving a communication of true or false positive ENBS (Beucher et al., 2010; Brockow & Nennstiel, 2019; Buchbinder & Timmermans, 2012; Chudleigh et al., 2016; Collins et al., 2013; Conway et al., 2022; Davis et al., 2006; Deliberazione Giunta Regionale Lombardia, 110 del 14 maggio, 2018; DeLuca et al., 2011, 2012; Dillard et al., 2010; Dixon-Woods, 2011; Farrell et al., 2020; Gale et al., 2013; Legge 19 agosto, 2016; Ministero della Salute, Decreto 13 ottobre, 2016; Morrison & Clayton, 2011; Salm et al., 2012; Tluczek et al., 2009; Ulph et al., 2017), suggesting that there are some commonalities across countries in the recommendations that can be developed from qualitative studies on ENBS. In particular, information needs, emotional distress linked to the communication of results and the importance of helping parents to navigate the healthcare system and the ENBS programme seem to be central and common topics in the USA as well as in Italy. Furthermore, recommendations on improving and standardizing the communication process of ENBS results are also a common point. An aspect that was relevant to the US context but did not emerge as relevant in our study is linked to information about financial aid and assistance.

This study strongly highlights the need for a clearer and better-structured communication of ENBS positivity which provides parents with a reliable and straightforward picture of the possible clinical trajectories (false positive, true positive). This objective should be achieved at the first test result by phone call communication. The content of the first phone call could be linked to the information leaflet on ENBS that parents received at the time of the test at the MCD, as the leaflet could be lost and its content is difficult to recall because of the overload of information and informational materials that parents usually receive during the pregnancy and at birth in the hospital.

Common scripts for the communication of ENBS positivity results and training and educational programmes for the healthcare staff of the MCD involved in the communication process could improve the quality of the communication, reduce parental distress and improve the parents' perception of the whole communication experience. A stronger co-ordination between MCD and MCC is also recommended.

In the UK, a recent training developed to improve the communication of positive results (Chudleigh et al., 2021) not only showed promising results but also highlighted the specificity of national contexts and the role of organizational factors that must be considered to reduce the variability of communication practices.

A recent position paper of EURORDIS-Rare Diseases Europe (EURORDIS, 2021) defines 11 key principles that should be followed in every European country to implement the ENBS programme. In particular, it has been highlighted the need for uniformity and quality throughout the ENBS process about timing, sample collection methods, follow-up and information shared with parents.

Furthermore, the availability of websites with reliable information on ENBS screened conditions can meet the parents' information need of having an online resource to consult while waiting for the in-person visit at MCC and in case parents need to review the information received during consultations.

Because of the regional variability in the communication processes and the differences in the availability of psychological support, an online resource could also be an important tool to provide contact information to access psychological support.

4.1 | Limitations

Some limitations must be considered. First, although adequate for qualitative study and analysis, the sample size is small and limited to one region of the country. Because of the variability of the communication procedures across the different Italian regions, our results cannot be generalized to the whole country. Furthermore, participants were mainly mothers with the fathers' perspective being strongly under-represented.

Another weakness of the study lies in the lack of the healthcare professionals' perspective. We cannot compare parents' and the healthcare professionals' perceptions and views of the ENBS result communication process. Furthermore, we selected participants from a sample of parents whose children had to undergo a second testing and subsequently attend an in-person visit at MCC. As we did not include parents who received the first test result by phone call communication but did not need to attend the in-person visit at the MCC as the children tested negative at the second test, we do not know whether the experience of those parents receiving only the first phone call communication is similar to that of parents who lived the two steps of the communication process.

Furthermore, the condition for which the positivity needs to be communicated can play a role in the communication process, and because of the limited sample size, we could not perform a stratified analysis.

Finally, we did not collect detailed information on the online sources parents gathered information from, and therefore, we could not verify whether they used scientifically reliable sources and whether and how the content of the information retrieved impacted their understanding and on their whole experience of the ENBS result communication process.

5 | CONCLUSIONS

This is the first study that describes the experience of parents receiving a communication of positivity at ENBS in Italy and advances for a more effective and functional communication process and information tools to provide clear and timely information and to minimize parental distress. Communication training for professionals involved in the communication process is also highly recommended, and stronger integration between MCD and MCC is needed.

HUMAN STUDIES AND INFORMED CONSENT

The study was approved by the local medical ethical committees of all participating centres. All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2000. All caregivers gave written informed consent for this study.

ANIMAL STUDIES

No non-human animal studies were carried out by the authors for this article.

AUTHOR CONTRIBUTIONS

Marco Bani provided complete contribution to the collection of data, as well as substantial contributions to the conception of the study, data analysis and drafting of the manuscript. Erika Raggi, Selena Russo, Serena Gasperini, Serena Motta, Francesca Menni, Francesca Furlan, Graziella Cefalo, Sabrina Paci, Giuseppe Banderali, Paola Marchisio, Andrea Biondi and Maria Grazia Strepparava provided substantial contributions to the analysis and interpretation of data and drafting of the manuscript. All authors gave final approval of this version to be published and agreed to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

CONFLICT OF INTERESTS STATEMENT

Marco Bani, Selena Russo, Erika Raggi, Maria Grazia Strepparava, Serena Gasperini, Serena Motta, Andrea Biondi, Graziella Cefalo, Sabrina Paci, Giuseppe Banderali, Francesca Menni, Francesca Furlan and Paola Marchisio declare that they have no conflict of interest.

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DATA AVAILABILITY STATEMENT

The data that support the findings of this study are available from the corresponding author upon reasonable request.

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