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LONG-TERM PSYCHOLOGICAL SUPPORT FOR FAMILIES RECEIVING COMMUNICATION OF POSITIVITY FOR METABOLIC DISEASES AT NEWBORN SCREENING

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***ABSTRACT* – Objective:** Receiving communication of positivity for metabolic disorders at newborn screening can be an extremely stressful experience for families, impacting the well-being and the quality of interaction with the newborn over short and long periods. Family functioning can be affected by various sources of distress, and psychological support is a pivotal intervention to mitigate its consequences.

Materials and methods: This report focuses on the long-term availability of psychological support for families as part of a wider national survey on psychological resources offered in the 23 Italian metabolic centers.

Results: Results showed that only 26% of the centers provide long-term psychological support, and in nearly 70% of cases, families are referred to psychological services available outside metabolic centers.

Conclusions: Given the specificity of metabolic diseases, psychological support should be guaranteed throughout all communication processes and within metabolic centers.

Keywords: Psychological support, Metabolic disorders, Family, Distress.

INTRODUCTION

Expanded newborn screening (ENBS) is a pivotal public health intervention to identify children with rare diseases. It provides diagnosis, immediate care, and treatment (if available) to prevent severe disabilities and save lives[1].

However, the psychological implications of communication of positivity can cause high levels of anxiety and distress and also affect the parental role. Studies have shown that the communication of positivity causes a sudden increase in parental distress that lasts for many months in both true-positive and false-positive cases[2].

According to previous studies, three main areas of distress for families can be identified: ambiguity of the disease, impact on family dynamics, and interaction with the healthcare system[3]. For families, it can be challenging to understand what a metabolic disease is because of the wide variability of conditions and the possible impact on the child's health. High uncertainty leads to strategies to resolve or

reduce distress, such as searching for information online or further consultation with the family doctor or pediatrician. However, these strategies may paradoxically increase the level of distress because the quality of online content cannot be controlled, and parents may read information about severe metabolic conditions. Furthermore, even among healthcare professionals not directly involved in metabolic centers, knowledge of metabolic diseases is limited, prompting parents to seek more information online in a vicious cycle (Figure 1).

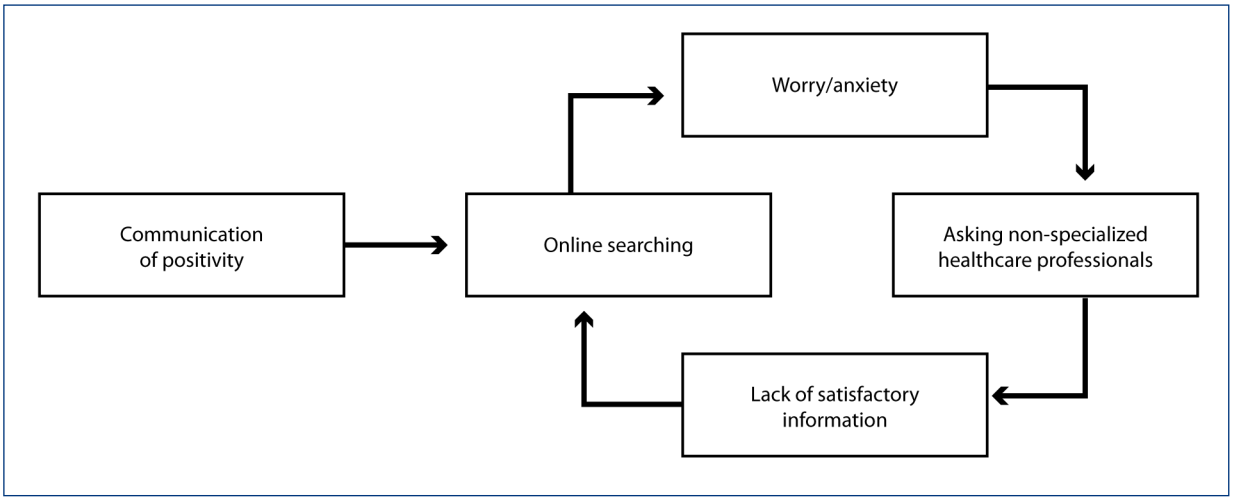

Figure 1. The vicious cycle of searching for health information online.

A health threat can typically cause a reaction of distress that suddenly diminishes after the threat ends. Conversely, in the case of metabolic diseases, distress perceived by families who have received confirmation of positivity persists over time. However, it can be modulated by the initiation of available treatments, psychosocial support, and the progressive adjustment process, including a better understanding of the clinical condition and its impact on the child's health and family well-being. Nevertheless, some studies have shown that for families receiving a false-positive communication, the distress remains higher even after receiving confirmatory test results[4]. The level of distress may also be high for families receiving communication of healthy carrier status, and the awareness of not needing clinical interventions may paradoxically be perceived as a danger to the child's health ("my child is a carrier, so what if…"). A few studies have suggested that false-positive communication may increase parents' perception of their children's vulnerability and, in some cases, increase the use of healthcare services[4]. However, other studies have confirmed the negative psychosocial effects of false-positive results but found no impact on early healthcare service utilization[5].

The Italian Coordinating Center on Newborn Screening, aware of the importance of the quality of parental caring behavior and the impact of communication of positivity[6], has recently highlighted the need to pay attention to the psychological effects of the communication process on children and their families, advocating the involvement of professionals to manage the families' psychological needs[7].

Despite this, a recent survey of Italian metabolic centers[8] reported that the availability of psychological resources is limited, and only 60% of them have a psychologist on their team; furthermore, in nearly 50% of cases, the psychologist is never involved in the first consultations.

This means that most families receiving a positive newborn screening result have to handle the distress of the communication and its consequences on the parent–child relationship on their own.

In recent years, the psychosocial needs of families and patients with metabolic diseases have become more important[9]. Little is still known about the mid-long-term psychological support available for families who have received communication of positivity; however, a European survey[10] showed that 66% of patients and 70% of parents expressed a need for psychological support. However, psychological assistance is limited to 36% for patients and 29% for parents, respectively.

This brief report aims to estimate the availability of mid-long-term psychological support in the Italian ENBS system and define the competencies needed to provide this service in the context of ENBS.

MATERIALS AND METHODS

As part of a national survey developed in 2022 by the Working Group on Clinical Psychology of the Italian Society of Metabolic Diseases and Newborn Screening (PSY-SIMMENS), ENBS centers were asked to report the availability of mid-long-term psychological support for families. A detailed description of the survey is reported elsewhere[8].

RESULTS

All 23 Italian ENBS centers completed the survey. Among the participating centers, only 26% reported the possibility of having mid-long-term psychological support 'always' available, while it was 'often' available in 22% and 'sometimes' in 39% of centers; in 13% of centers, it was never possible to have mid-long-term psychological support.

The reasons for seeking mid-long-term psychological support vary widely, including challenges in coping with the diagnosis, socio-economic difficulties, and treatment compliance.

Notably, 70% of centers refer families to non-specialized public psychological services available in their area, while in 13% of the cases, families have to apply directly to psychological services.

DISCUSSION

In the Italian context, mid-long-term psychological support for families appears to be guaranteed within a minority of metabolic centers. In contrast, in most cases (nearly 70%), families are redirected to local psychological services outside the metabolic centers.

Considering the well-known difficulties that families with metabolic disorders face with non-specialized healthcare services (with limited knowledge and sensitivity towards metabolic conditions)[11,12], it is necessary to provide psychological support within metabolic centers and ensure the possibility of long-term support.

It is important to emphasize that communication of positivity is a punctual event whose effects can last for months or even years through several mechanisms; a direct effect relates to the short-term impact of communication that causes anxiety, distress, and excessive worry about the baby's health that can disrupt parental coping resources, shifting attention from immediate needs (feeding, care) to real or hypothetical (for false-positive cases) future health problems ("what if…"). An indirect effect is related to the impact of distress on parental caring behavior. Caring includes parental responsiveness to the child's physiological and emotional needs: this is an essential parental task in the early years of children's lives, particularly in the newborn's first weeks, to meet their needs.

The quality of caring behavior in the early months of children's lives can have a profound impact on short and long-term health outcomes, not only on children with pathological conditions but also on healthy ones. Previous studies on healthy children have shown that the quality of parental care in the newborn's first months has an impact on child development; for example, in a US longitudinal study[13], children of mothers with increased levels of anxious thoughts about parenting at the first month postpartum showed lower socio-emotional skills at 18–24 months.

Another longitudinal study on 187 mother-child couples from The Netherlands[14] showed that maternal sensitivity may affect children's physical health development even in a low-stress context. A similar pattern was reported in a sample of 603 children from poor communities in Mexico[15]; the authors showed that parental warmth and responsiveness during infancy were significant predictors of child development at ages 3–5 years. Furthermore, in a sample of 488 parents, the authors found a negative correlation between post-traumatic stress disorder symptoms after childbirth and perceived parent-infant bonding at 3 months postpartum for mothers[16].

Observing families receiving communication of positivity and those with children with metabolic disorders, it is easier to speculate that the disruptive impact of positivity may be an additive factor affecting the quality of caregiving and requiring adequate management in the short and medium term. There has been growing interest in the impact of metabolic diseases on parents' burden and well-being; some studies on families of children with metabolic diseases have shown an impairment of the quality of life of patients and families[17,18].

Others reported that, while most parents are satisfied with the care received, some criticism arises about the interaction with healthcare services outside of specialized services, lacking knowledge and sensitivity towards the specificity of metabolic conditions[11,12].

CONCLUSIONS

To ensure adequate psychological support for families, every metabolic center should include a psychologist on the team with specific expertise in metabolic disorders.

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