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Patient organizations: advocating for timely newborn screening & improved quality of life

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Abstract

Timely diagnosis is a crucial first step for individuals with rare diseases, significantly affecting their access to treatment, care, and overall well-being. Patient organizations have actively engaged in advocating for Newborn Screening (NBS) to raise awareness about rare diseases. This article examines the efforts of six patient organizations worldwide, each functioning as a national, regional or disease-specific alliance with distinct characteristics. These organizations strongly support NBS due to its proven potential to enable early diagnosis and facilitate timely referral to appropriate treatment and care. They employ several approaches to advocate for the expansion of screening panels or implementation of NBS programs, including education, evidence gathering, and

fostering multidisciplinary collaboration. There are opportunities to engage the public and emphasize the value of NBS as a public health service that benefits everyone.

Keywords: Newborn screening, prevention, patient advocacy, rare diseases, newborn screening policy, newborn screening ethics, bloodspot

INTRODUCTION

Newborn screening (NBS) Programs are public health initiatives designed to identify potential health concerns in babies shortly after birth. Specifically, bloodspot NBS involves the systematic screening of newborns for a range of inherited, genetic or congenital disorders that may not be apparent at birth but could lead to serious and irreversible health complications, including premature death, if left undetected. Timely screening and subsequent diagnosis of diseases enable healthcare professionals (HCP) to provide immediate care and avert irreversible outcomes before symptoms manifest. However, unexpected diagnoses from screening can significantly impact families, necessitating arrangements to support their well-being, including psychological assistance[1]. A diagnosis offers people with rare diseases and their families the opportunity to connect with support groups, enroll in clinical trials, engage in proactive family planning, and make financial and other life plans[2].

There are more than 6,000 known rare diseases to date, of which 70% result from genetic causes and 72% have childhood onset[3]. Rare diseases are a heterogeneous group of diseases with complex clinical presentations, posing various challenges for patients, families, and society[4]. The availability of scientific knowledge and diagnostic technology for bloodspot NBS varies across countries[5]. Currently, there is no global consensus on specific diseases to be included in a national NBS program[6-8]. Each country decides which conditions are screened, considering international scientific recommendations and local contexts.

Patient organizations (POs) have been instrumental in enabling the implementation and introduction of NBS since its inception by Dr. Robert Guthrie in the 1960s for Phenylketonuria (PKU). In this article, we explore the role of six POs in promoting NBS and highlighting their advocacy efforts toward making NBS a reality in their contexts.

PATIENT ORGANIZATIONS IN ACTION

This Perspective is primarily a collaboration between the international rare diseases research consortium (IRDiRC) and the patient advocacy constituent committee (PACC). Organizational characteristics or other characteristics are described including, such as their disease focus, national alliances, regional alliances, and locations across different continents, are described.

Colombian federation of rare diseases

Colombia has made significant progress in the legislation for rare diseases. Newborn Screening Law was approved in 2019, which aims to regulate diseases that impact quality of life, to prevent disease progression, sequelae, and disability. Implementation has been limited since its approval. Currently, active NBS is undertaken in Columbia for congenital hypothyroidism (CH), together with broader NBS for hearing, vision, and congenital heart defects (CHD). Implementation of screening for additional diseases for NBS, such as congenital adrenal hyperplasia (CAH), PKU, Galactosemia, cystic fibrosis (CF), and Biotinidase Deficiency/Hemoglobin defects, is pending a resolution from the Ministry of Health in order to generate guidelines for national implementation and earmarking of resources and reimbursement.

The Colombian Federation of Rare Diseases (FECOER) is a non-profit organization (NPO) launched in 2011 to promote the inclusion, integration, and care of people living with rare diseases (PLWRD), including supporting and monitoring the implementation of the NBS program. FECOER includes 48 organizations/support groups for PLWRD, working together to develop and implement advocacy strategies in public policy, education, and awareness. The primary FECOER approach for NBS is via the National Board for Rare-Orphan Diseases, which works to design and disseminate actions for public policy to improve the lives of people living with rare diseases through stakeholder participation. The National Board was established through Resolution 1,871 of 2021[9], which includes 15 health sector representatives, the Ministry of Health, health insurers/funders, service providers, scientific societies, and POs. FECOER also collaborates with scientific societies and academia to educate and sensitize HCP and the wider community about the challenges faced by patients/families accessing NBS.

Cyprus alliance for rare disorders

In Cyprus, the national bloodspot NBS program is limited to screening for PKU and CH, while debate continues around potential expansion to include eight more manageable and epidemiologically relevant metabolic disorders via a pilot. NBS advocacy efforts have been led by the Association for Patients with Inherited Metabolic Disorders and Friends "Aspida Zois", Cyprus Muscular Dystrophy Association, and Cyprus Association for Primary Immunodeficiencies and Friends, both individually and collectively under the umbrella of the Cyprus Alliance for Rare Disorders[10]. In 2022, the POs organized a successful petition that led the Council of Ministers to reconsider and approve the expansion of NBS to include eight additional inherited metabolic diseases[11]. In addition, the Cyprus Alliance for Rare Diseases regularly issues position paper and proposals aimed at policy makers to improve early diagnosis and care services. Such advocacy efforts involving POs remain largely unseen and unreported in the scientific literature.

Cyprus has a long history of prevention of rare inherited conditions through prenatal screening for thalassemia. With 12% of the population estimated as being asymptomatic carriers and 650+ thalassemia patients, couples are required by the Church to be screened and counselled premaritally for thalassemia[12,13]. Newborns whose parents have been identified as thalassemia carriers are considered to be at increased risk and their NBS includes thalassemia screening. This program has been in place since 1980, and Church involvement has contributed to the successful implementation. It is an example of successful collaboration between patients, families, HCP, and decision-makers who agree on the significant burden of thalassemia, describing it as their "national disease". In addition, prenatal screening is available for Duchenne and Becker Muscular Dystrophy, Spinal Muscular Dystrophy, CF, Huntington's disease, and other single-gene disorders when there is a family history. For these families, the State covers the cost of a defined number of cycles of *in-vitro* fertilization at the specific Center of Reference.

Participation in relevant European groups and international conferences, such as EUROPLAN National Conferences and International Society for Neonatal Screening, helps to expand knowledge on NBS and the potential benefits when implemented[14,15]. Local POs often seek collaboration with specialists and national alliances to gather scientific facts and support their advocacy efforts. Publicizing issues through media channels has been a key tactic in achieving awareness and leveraging State involvement for these conditions.

Rare diseases South Africa

NBS has been offered sporadically in South Africa since the early 1990s, mainly for CH. However, competing health priorities, most notably HIV/AIDS and Tuberculosis (TB), have resulted in the neglect of community genetic services, including the diagnosis and treatment of congenital disorders and rare diseases. The 2021 National Clinical Guidelines for Genetic Services specify clinical examination of a newborn, auditory screening, pulse oximetry for CHD, and biochemical screening for inborn errors of

metabolism (IEM). However, implementation of these guidelines remains fragmented. NBS is currently only available to a few South Africans (< 0.5% of million annual births) across both the state healthcare sector, which serves 86% of the South African population, and the private healthcare sector, which serves the remaining 14%)[16]. This includes either out-of-pocket NBS for a panel of conditions privately or CH NBS at a limited number of state facilities in the Western Cape[17]. To implement these provisions countrywide for the population served by the state served portion of the population requires, state funding is required.

Founded in 2013, Rare Diseases South Africa (RDSA) is an NPO dedicated to increasing recognition and support for people living with PLWRD and congenital disorders. Its mission is to enhance health services and overall quality of life for these individuals. RDSA has successfully launched initiatives that have positively impacted the lives of over 6,000 patients, engaging with governmental departments, organs of state, industry and strategic stakeholders to raise awareness and to improve policy implementation. In February 2023, RDSA held a two-day NBS event involving key decision-makers (national/provincial Departments of Health (DOH) and other global and local stakeholders involved with NBS or keen to expand these services. A key outcome of this event is a planned pilot project to demonstrate the benefits, costs, outcomes, technical and capacity requirements of expanding CH NBS countrywide[17]. The National DOH supports this approach and the demonstration project proposal for two provinces is under development for funding. Discussions are underway on how to best use existing infrastructure, including HIV programs, particularly those involving blood samples taken at birth, to expand NBS in South Africa[17]. This initial project will be completed by the end of 2025 and will provide key information to decision-makers to consider expanding CH NBS services in the country[17].

Asia Pacific alliance of rare disease organizations

The populations affected by rare diseases in the Asia-Pacific region face exacerbated challenges due to a lack of awareness and limited capacity for screening, diagnosis, and care[18]. The Asia Pacific Alliance of Rare Disease Organizations serves as the regional advocacy body, comprising national and regional POs as members[19]. Bloodspot NBS practices vary across the region both in terms of the number of conditions screened and the level of coverage. For example, the expanded NBS program in Thailand covers 40 conditions, while the program in Japan includes 20 conditions[5,7,20,21]. In India, less than 4% of > 25 million newborns annually are screened for a limited number of conditions[22], while in the Philippines, 94.6% of newborns are screened for 29 conditions[23]. Relatively lower levels of education and health literacy among parents add to the challenges of successful NBS programs[24]. Nevertheless, while mature and impactful rare disease POs are relatively limited in the region, often compounded by the prevalent culture of patriarchal health systems, a number of organizations, along with dedicated healthcare professionals, have made significant strides by collaborating with national health agencies to advocate for NBS[25-27].

While Taiwan's NBS covers 21 conditions, the Taiwan Foundation of Rare Disorders has helped expand and subsidize screening for the region's indigenous populations[28]. Rare Voices Australia has similarly supported both the call for and advised on a mechanism for the funding of a national approach to review screened conditions as part of their national strategic action plan for rare diseases[26,29]. However, screening panels are often limited to conditions for which treatment is available. Despite their success, initiatives often fail to scale up due to a lack of follow-up intervention and care plans.

In the absence of health coverage for screening and follow-up care, NBS programs in the region remain dependent on a country's political will and commitment to universal health coverage (UHC). Funding mechanisms and follow-up care are largely dependent on individual government buy-in. Countries that

collaborate with patient organizations appear to have relatively improved programs. These examples demonstrate that PO partnerships are critical for continued NBS expansion and sustainability of intervention and care post diagnosis. The number and maturity of regional POs are growing, and there is growing interest in learning from the successes and leveraging the opportunity to develop advocacy. Together, they will augment the work of healthcare providers in supporting affected families with knowledge and support services.

EURORDIS - rare diseases Europe

NBS is a priority topic for EURORDIS, an umbrella PO for rare diseases in Europe with 1042 member POs across 74 countries. By exchanging best practices and engaging in structured European Union (EU) collaboration, EURORDIS aims to achieve equity in NBS in Europe. EURORDIS adopts multiple approaches to advance NBS, such as the NBS Working Group, a multistakeholder group of 35 members from 15 countries[30].

EURORDIS recognizes the potential of new NBS technologies and genomic testing and plays a critical role in the Innovative Medicines Initiative (IMI)-funded project Screen4Care, which aims to accelerate rare disease diagnosis through NBS and digital technologies. Within the project, EURORDIS has contributed by developing and launching a Rare Barometer survey on NBS, which was responded by over 6,000 individuals living with a rare disease and their families, providing an empirical basis for identifying robust criteria for actionable diseases benefiting most from NBS and for developing a list of actionable diseases for genetic NBS[31]. Moreover, EURORDIS enhances and encourages patient involvement in the project through its stakeholder NBS working group, which provides input on all activities and challenges related to the development of genetic NBS and its implications for patients, as well as through the Patient Advisory Board. EURORDIS' efforts have gathered momentum and recognition from European institutions, with 21 EU countries supporting the EU Council Presidency Call to Action from the Expert Conference on Rare Diseases "Towards a new European policy framework on rare diseases", to the European Commission and Member States in October 2022.

SMA Europe

Spinal muscular atrophy (SMA) is a rare, genetic neurodegenerative disease that affects mainly infants, young children, and adults. Three new treatments have been approved by the European Medicines Agency (EMA) between 2018-2022, improving the patient life expectancy and prognosis. However, children receiving treatment still develop motor disabilities, with severity varying based on the age of diagnosis and the extent of motor impairment at the start of treatment. Clinical trial evidence demonstrates that all three new treatments are more efficacious when initiated earlier.

The SMA NBS Alliance was created in February 2020 to accelerate the adoption and implementation of NBS for SMA in Europe. In 2020, only three European countries had SMA NBS for parts of the countries or as a pilot[32]. The Alliance is led by a steering committee of patient advocates and HCP. It is supported by pharmaceutical companies working in SMA, manufacturers developing screening assays, and universities contributing medical, ethical, and economic knowledge. Alliance activities began by developing campaign materials to educate, engage, and raise awareness, and in late 2020, published a White Paper detailing the scientific, economic, ethical, and social case for SMA NBS to help national authorities respond to calls for inclusion of SMA on national NBS panels[33]. By late 2022, > 50% babies born in Europe were being screened for SMA within the first week of life. Today, the Alliance provides targeted support to national SMA organizations in countries where SMA screening is unavailable, through organizing conferences, drafting arguments, launching communication campaigns, and sharing best practices and learnings between countries. The Alliance also supports/funds meetings of European experts to recommend the best screening techniques and harmonize practices.

ADVOCACY FOR EARLY DIAGNOSIS AND DISEASE MANAGEMENT

Diagnosis is a crucial step for PLWRD, impacting their treatment access, care, and overall well-being. Globally, POs are engaging in NBS advocacy and discussion at various levels and examples in this article offer indications of their contributions. Despite their different locations, contexts, and disease focuses, the points of commonality between these POs are their advocacy efforts, facilitation of collaboration, supporting awareness and education, and policy influence. These efforts are rarely documented in the scientific media but often help leverage the implementation and expansion of NBS.

Each organization emphasized the importance of raising awareness and educating both HCPs and the wider community and is actively involved in advocacy efforts about the benefits of NBS. The objective of POs to raise awareness, communicate the lived experiences/expertise of PLWRD, families and caregivers who have been impacted by conditions that could have been prevented or mitigated adds value and purpose to the development of NBS programs[34,35]. In addition, they can offer perspectives on screening conditions for which there are no treatments, conditions which are manageable with dietary changes, and conditions for which there is effective treatment if administered before the onset of symptoms[36]. The input of patient representatives is critical for NBS discussion and decision making.

POs play a central role in facilitating collaborations and political commitment. They can influence policy at the national or regional level, participate in policy discussions, and advocate for resource allocation toward rare diseases and NBS[17,23,26,27,30,35]. Collaboration is a key strategy, whether with other PO, healthcare professionals, societies, academia, or governmental bodies. To this end, they use varied channels such as media, events, publications, and educational initiatives to disseminate information and mobilize support for their advocacy efforts. They may direct research agendas and strategies, form partnerships, collect learnings from other diseases, and engage with scientific literature to gather or compare data[37-39]. They can provide resources for developing therapeutic protocols, creating effective health policies, and providing continuous medical education. Their impact is determined by their knowledge, ability to present evidence, and networking capacity with stakeholders[35,40]. Many developing countries lack NBS programs, and POs are recognized to be essential to ensure equitable access to NBS[5,7,35].

CONCLUSION

While NBS programs are expanding to include more conditions, they only include less than 1% of known rare diseases. Ethical dilemmas and uncertainty surrounding the consequences of a rare disease diagnosis may hinder parental acceptance of NBS, particularly when a child appears healthy. Parents may fear societal stigma, discrimination, and socio-economic burdens. Impacted families understand the unavailability, inaccessibility, or unaffordability of necessary services and treatments. Raising public awareness helps overcome these barriers by fostering an understanding of the importance and potential benefits of NBS programs.

POs play a crucial role in the development and implementation of NBS policy as they work to enhance messages, promote collaboration, and facilitate consensus among diverse stakeholders. NBS offers invaluable benefits that will shape the future of healthcare and a more equitable society. A well-informed society is pivotal in driving forward the acceptance and effectiveness of NBS programs, ensuring every child has the opportunity for a healthier start in life.

DECLARATIONS

Authors' contributions

Made contributions to the conception and design of the study and performed analysis and interpretation: Malherbe HL, Jain R, Antoniadou V, Ouillade MC, Gil Cardozo DF, Gumus G, Kitikiti NS, McKay L, Wang CM

All authors contributed to the review and editing of the manuscript.

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Kitikiti NS is a full-time employee of Takeda Pharmaceuticals International AG. All other authors declared that there are no conflicts of interest.

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Not applicable.

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