



# Supporting Patient Advocacy for Newborn Screening



**SMA Europe**

**CRA** Charles River  
Associates

*This publication has been financed by Novartis Gene Therapies. The conclusions, interpretations and opinions expressed herein are those of the authors alone. Novartis Gene Therapies declines any responsibility for the content of this publication.*

Dear all,

This toolkit, for internal use, is intended to assist you in identifying and preparing for potential challenges you may face, in your country, when advocating for high quality and equitable screening and including suggestions on how patient advocacy groups can better engage in process to support good decision making.

It has been developed through a collaboration of SMA Europe and Novartis Gene Therapies based on findings of a survey of SMA Europe members and a series of follow-up interviews with a number of national SMA organisations across Europe. While the experiences reported are based on those of SMA groups, learning and guidance provided in the toolkit may be applied to other rare diseases facing similar challenges.

The toolkit should be considered a 'living document' with updates expected in future covering the experiences of other countries, and as the NBS landscape continues to evolve.

We would like to thank all those who have contributed to the development of this toolkit.

### What this is



- A toolkit identifying a range of likely challenges patient advocates for NBS may face nationally in European countries (and may be applicable to other countries globally)
- Provides potential solutions to the challenges with examples of case studies from SMA patient organisations

### What this is not



- A comprehensive list of all challenges you may face when advocating for NBS expansion
- Providing mandatory advice on how to conduct your own advocacy activities

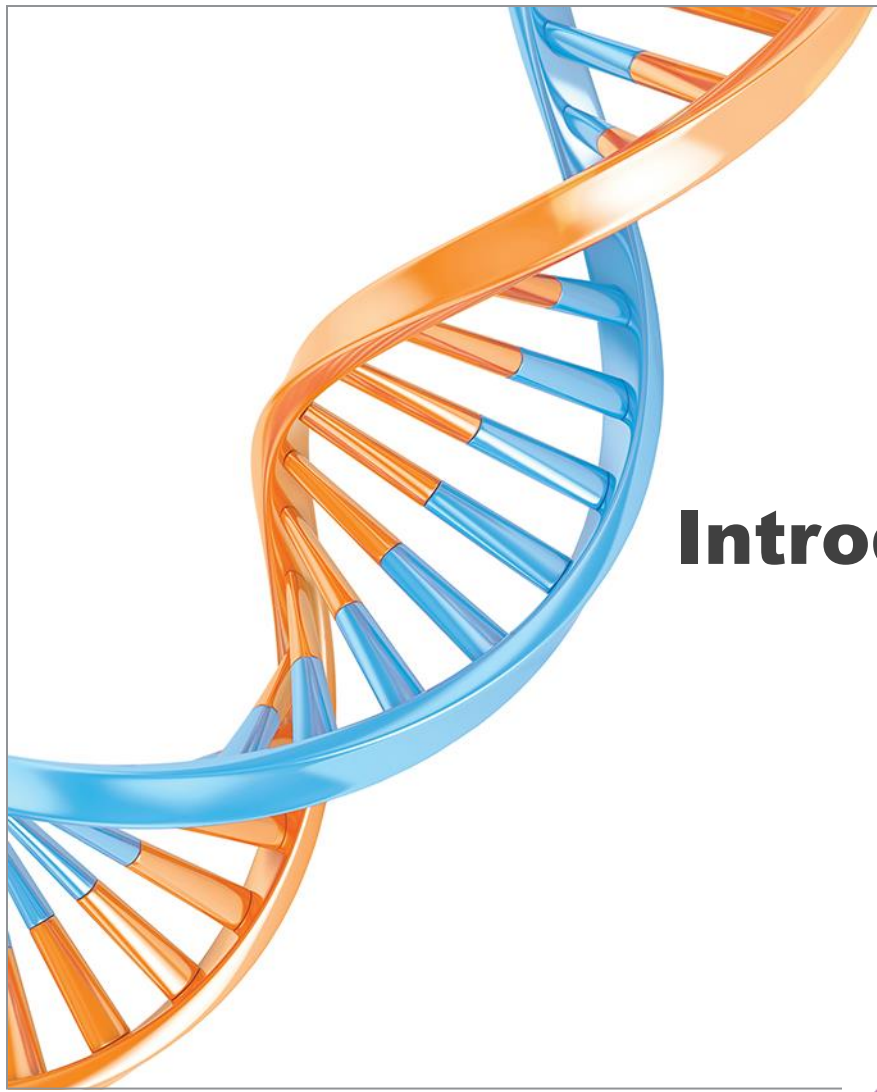
*Many challenges and solutions can be country-specific and you should adapt any activities to your country's current situation*

***Content in this toolkit is derived from surveys and interviews with patient groups and should be considered their opinion only***

# Glossary of terms

<b>HTA</b>	<b>Health Technology Assessment</b> <i>A process to evaluate new health technologies (e.g. treatments or diagnostic processes (such as NBS)) to determine if they should be provided by a healthcare system and the level of funding to be provided. This can include a clinical and economic assessment to determine if the health technology is a good use of the healthcare systems resources</i>
<b>MoH</b>	Ministry of Health
<b>NBS</b>	Newborn Screening
<b>PAG</b>	Patient Advocacy Group
<b>SMA</b>	Spinal Muscular Atrophy









## **Introduction and supporting resources**



**SMA Europe**

**CRA** Charles River  
Associates

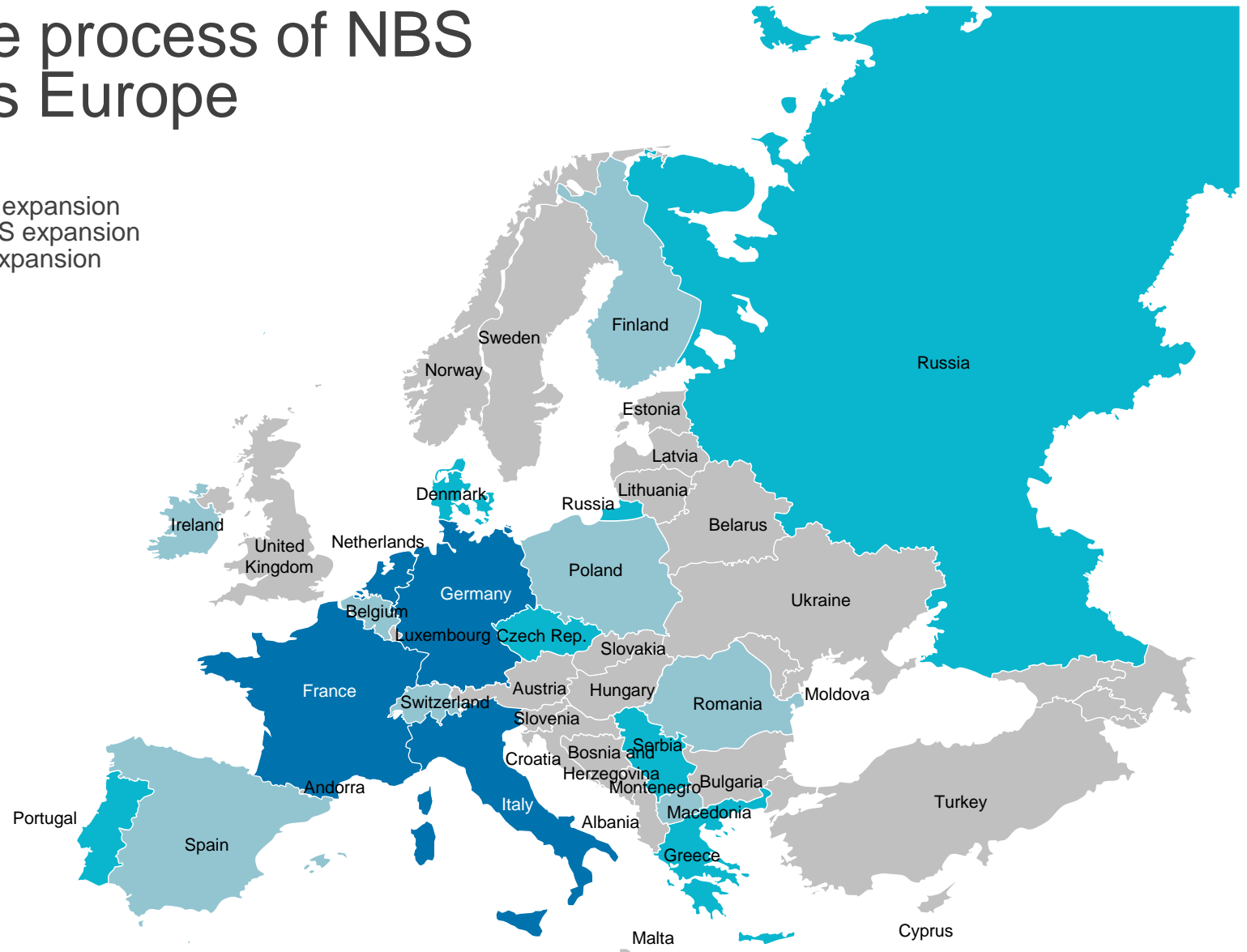
# PAG\* involvement in the process of NBS expansion varies across Europe

-  PAGs consider themselves formally involved in NBS expansion
-  PAGs consider themselves informally involved in NBS expansion
-  PAGs do not consider themselves involved in NBS expansion
-  No survey response

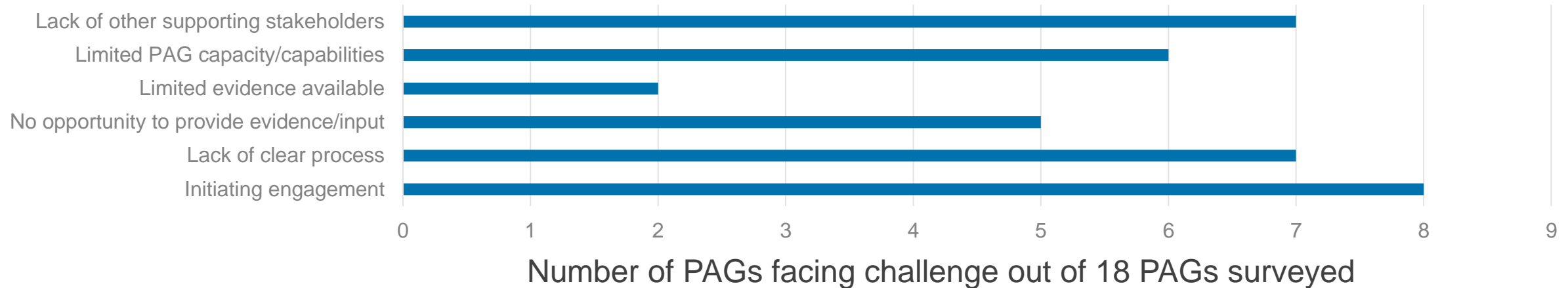
## Key insights

- Very few countries surveyed have a clear, pre-defined role for PAGs in the process of NBS expansion (4/18 countries have formal engagement processes for PAGs)
- However, even PAGs who stated that they did not consider themselves involved in the process (either formally or informally) were often able to conduct activities to engage with and influence key stakeholders

\*PAGs surveyed were members of SMA Europe – some work across multiple neuromuscular diseases as well as SMA



# PAGs in most countries surveyed are facing multiple interacting challenges



## Key insights

- The majority of PAGs (16/18 countries surveyed) feel that evidence supporting the benefits of SMA NBS is available, however many (7/18 countries) find that but that the process of getting the test included on NBS panels is unclear, which makes it difficult to find opportunities to communicate the evidence to decision-makers effectively
- Most PAGs highlighted several interacting challenges, for example having limited resources and capabilities makes it more difficult to initiate engagement



# PAGs surveyed reported many organisations and stakeholders who are involved in decision-making for NBS expansion

**Patient advocacy Groups (PAGs):** Other patient organisations may have similar goals and could benefit from collaboration with patient groups

**Physicians:** Specialists and medical societies/associations can be key allies in supporting the case for NBS, and help in developing testing protocols and providing technical analysis to support the case for NBS

**Ethics Committees:** Potentially as part of the MoH/ screening committee a clear ethical need for NBS has to be made

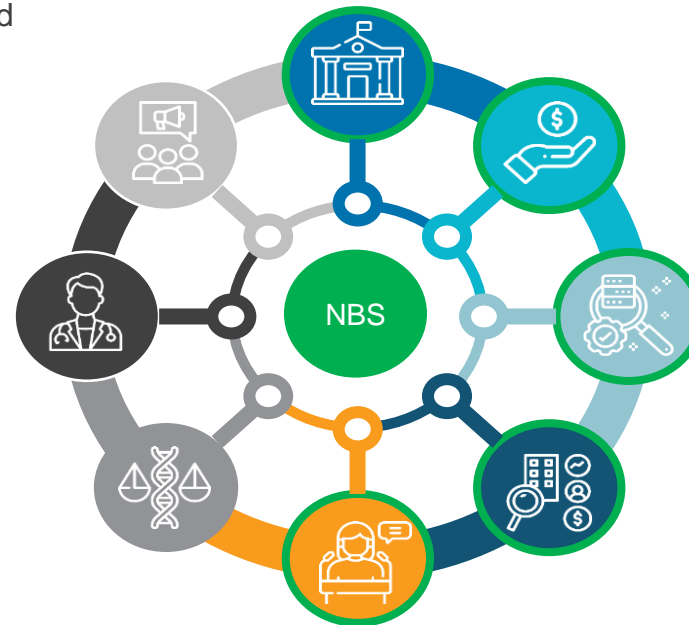
**Politicians:** Members of national/ regional legislatures (and Members of the European Parliament) can raise the issue of NBS with the government or relevant ministers

**Ministry of Health (MoH):** The Minister of Health is typically the ultimate decision maker, taking into account advice from multiple sources including civil servants within the Ministry, to make a final decision on NBS

**Payers:** National/regional insurance agencies or healthcare systems will be responsible for funding any expansion in NBS

**Health technology Assessment (HTA) Bodies:** Specialist agencies, typically part of the MoH/Payer, who may assess any new health technology, including NBS testing, to determine if it is an appropriate use of resources

**Screening Committees:** A specialist public health committee focusing on screening programs, typically part of the MoH. May also be involved in clinical or economic assessment of NBS expansion proposals



Note: PAGs should also consider other targets for communication and advocacy such as the public, media organisations or civil rights groups



# From the survey of SMA Europe members several key interconnected barriers to patient advocacy to expand NBS were identified

## Lack of process for inclusion of PAG voice

[See more](#)

Even if PAGs have a clear strategy for advocacy and have supporting information they wish to share, it can be difficult to find opportunities to effectively engage in NBS decision making

## Limited capacity in PAG

[See more](#)

PAGs are often small organisations and lack financial and human resources for large scale advocacy work

## Difficulties engaging and communicating arguments

[See more](#)

PAGs can experience push-back against their arguments and evidence and may find it difficult to engage and advocate effectively to decision makers

## Logistical, infrastructural and implementation issues

[See more](#)

Practical issues around the implementation of large-scale NBS can be used as an argument against approval and can raise barriers against the roll out of screening or limit the benefits of the programme

## Legal and procedural delays and inefficiencies

[See more](#)

Incorporation of new tests in some countries is slow and inefficient as a result of the lengthy legal or political processes involved

Key stakeholders  
library

Additional  
resources library

Case studies  
library





# Summary of best practices



For each meeting or interaction, **think about the audience's situation, goals, motivations and capabilities**, in order to work out which arguments or information will appeal to them and be most impactful



Consider any local sensitivities and adjust communication accordingly, for example referring to the 'heel prick test' rather than 'newborn screening' in countries where this term has negative connotations



Where possible **use local data and examples** to support arguments – if this is not possible, think about using examples from countries with similar healthcare systems



Remember the **value of personal connections** and focus on finding a balance between applying necessary pressure and potentially damaging relationships



Where possible, **collaborate with other organisations or individuals** who have similar goals and can provide expertise or resources to help overcome challenges



**Encourage holistic thinking** wherever possible e.g. highlighting the wider care and management costs associated with disease-related disability as a potential benefit of improving access to better treatment outcomes, or the potential for investments in infrastructure to be beneficial and generate savings across multiple diseases as new treatments become available



# From the survey and follow-up discussions key actions and best practices were identified by patient groups across Europe (1/3)

**Advocates can influence the development of new processes for NBS expansion and should anticipate the potential key challenges throughout the NBS process and plan early to address any concerns**

*Challenge*

NBS is often a lengthy and complex process (often with no clear process on how to proceed with expanding an existing programme) and the hurdles faced by advocates can vary significantly by country and where they are in the NBS process

*Key example of PAG activities*

- In Poland Fundacja SMA facilitated discussions between key clinicians, testing lab scientists and patient advocates to develop a detailed plan for how to design and implement NBS in Poland, which was a key part in making the case for NBS to the national decision makers. However once approved NBS was still subject to regional implementation which led to further delays in diagnosis
- In Germany Initiative SMA worked with testing labs and using examples of previous NBS expansion to make the case for rapid rollout of NBS once a national programme was approved

*Conclusions*

To make the case for NBS patient advocates may need to create new processes in collaboration with experts  
It is important to remember that succeeding in establishing a pilot programme or full NBS programme can still lead to further challenges in implementation which may not have been anticipated



# From the survey and follow-up discussions key actions and best practices were identified by patient groups across Europe (2/3)

**Where possible, collaborate with other organisations or individuals who have similar goals and can provide expertise or resources to help overcome challenges**

*Challenge*

Many patient groups have limited resources and may lack the financial and human resources for large scale advocacy work or political/clinical expertise and could therefore benefit by pooling their efforts where possible to advocate for NBS for multiple conditions

*Key example of PAG activities*

In Romania, Asociatia SMACARE worked closely with the national rare disease organisation to highlight the need for NBS, not just for SMA but to highlight the future cost savings expanding the current NBS infrastructure would bring, through earlier diagnosis and improved treatment outcomes

*Conclusions*

NBS is a key issue across many rare diseases and is set to become an even greater issue as more treatments Currently diseases such as SMA are under consideration for NBS due to treatments and validated tests being available, however the calls for NBS will continue to grow.

Governments should consider spending on NBS infrastructure as an investment to be beneficial and generate savings across multiple diseases as new treatments become available



# From the survey and follow-up discussions key actions and best practices were identified by patient groups across Europe (3/3)

**For each meeting or interaction, think about the audience's situation, goals, motivations and capabilities, in order to tailor your arguments or information to be most impactful**

*Challenge*

Approving and implementing NBS can be a highly complex process with many decision makers and advisors in the process and it can be difficult to communicate the arguments for NBS in an impactful way

*Key example of PAG activities*

- In Portugal the APN developed a formal white paper for government and health authorities with input from scientists, doctors, pharmaceutical companies and providing detailed comparisons with other European countries, Canada and Australia, supporting approval of a pilot study
- In Greece, MDA Hellas presented patient and parent testimonial videos to government ministers (including videos of SMA patients who were diagnosed early and subsequently received early treatment) to demonstrate the importance of early diagnosis and therefore the need for NBS to support better treatment options

*Conclusions*

Although the overall arguments may be similar, multiple advocacy approaches should be considered across all relevant decision makers

Interactions with key decision makers should be followed by reflection to allow you to continue tailor and develop appropriate and impactful arguments to make the case for NBS



# To successfully expand NBS to include a new test, PAGs will need clear messages to support the value of NBS

Theme	Supporting messages
<p><b>Overview of NBS and the need for NBS implementation</b></p>	<p>Universal NBS is a public health priority that ensures equitable access to early disease detection and treatment (when available) for all children regardless of geography or socioeconomic status. Despite the availability of disease-modifying therapies, key diseases, such as SMA, are not universally included in NBS panels<sup>1</sup></p>
	<p>NBS is not universally available, and even if available, often does not include key treatable diseases on the panel due to differing approaches to amending NBS panels and differences in the supporting infrastructure / pathways to implement changes<sup>1</sup></p>
	<p>The emergence of disease modifying therapies, such as gene therapies has accelerated the urgency to identify and treat rare diseases earlier in the disease course, which underscore the urgent need for continued adoption of universal NBS<sup>2-5</sup></p>
	<p>The real-world impact of early diagnosis followed by prompt treatment supports the urgency for broader investment in and implementation of NBS for rare diseases with available treatments. NBS is an accurate and effective method for early diagnosis of many rare diseases including SMA, highlighting the need for universal adoption to improve patient lives when treatment is available<sup>6,7</sup></p>

(1) CRA (2021) *A Landscape Assessment of Newborn Screening in Europe*. Available at <https://media.crai.com/wp-content/uploads/2021/11/19130322/CRA-LS-Insights-NBS-Policy.pdf> (Accessed 14 November 2022)  
 (2) Servais L, et al. *MDA* 2022; (3) Glascock J, et al. *J Neuromuscul Dis* 2018;5(2):145-158.; (4) Dangouloff T, Servais L. *Ther Clin Risk Manag*. 2019;15:1153-1161.;  
 (5) European Alliance for Newborn Screening in SMA (2021) *Spinal muscular atrophy: Screen at birth, save lives*. Available at: [https://www.sma-screening-alliance.org/wp-content/uploads/2021/11/Spinal\\_muscular\\_atrophy\\_Screen\\_at\\_birth\\_save\\_lives\\_Whitepaper\\_SMA\\_NBS\\_Alliance\\_v2\\_25NOV2021.pdf](https://www.sma-screening-alliance.org/wp-content/uploads/2021/11/Spinal_muscular_atrophy_Screen_at_birth_save_lives_Whitepaper_SMA_NBS_Alliance_v2_25NOV2021.pdf) (Accessed 14 November 2022); (6) Kariyawasam DST et al. *Genet Med Genet Med*. 2020;22(3):557-565.; (7) Vill K, et al. *J Neuromuscul Dis* 2019;6(4):503-515.



# To successfully expand NBS to include a new test, PAGs will need clear messages to support the value of NBS

Theme	Supporting messages
<b>Importance of NBS in early diagnosis &amp; treatment of rare diseases</b>	<p>Early detection of diseases such as SMA, and intervention with disease modifying therapies may lead to improved disease management and improvements in patient survival and quality of life. Efforts to support pre-symptomatic treatment of conditions such as SMA via NBS and other early detection methods have transformed identification and early treatment<sup>1-3</sup></p>
	<p>The implementation of NBS can enable rapid detection and early treatment, and may support improved outcomes for patients with rare diseases, with the implementation of NBS and connection to an appropriate treatment centre being shown to significantly reduce the age of treatment intervention<sup>3-5</sup></p>
	<p>In diseases such as SMA, clinical guidelines recommend to initiate treatment as early as possible due to the strong evidence demonstrating significantly improved health outcomes versus later treatment<sup>2,3</sup>. NBS can be valuable in supporting early diagnosis and treatment</p>
	<p>Where implemented, NBS has revolutionized disease prognosis by enabling early detection and treatment, and may lead to improved patient outcomes, which can reduce the burden on both caregivers and the healthcare system<sup>6,7</sup></p>
	<p>In the absence of NBS, rare diseases such as SMA are typically diagnosed by clinical presentation of signs and symptoms with subsequent genetic testing, but lack of awareness of symptoms and inequities in healthcare systems often lead to delays in diagnosis, requiring vigilant monitoring of symptoms to detect diseases early and to ensure early treatment<sup>8</sup></p>

(1) Govoni A, et al. *Mol Neurobiol* 2018;55(8):6307-6318.; (2) Glascock J, et al. *J Neuromuscul Dis* 2018;5(2):145-158.; (3) Glascock J, et al. *J Neuromuscul Dis* 2020; (4) Dangouloff T, Servais L. *Ther Clin Risk Manag.* 2019;15:1153-1161.; (5) Servais L, et al. *MDA* 2022 (6) Vill K, et al. *J Neuromuscul Dis* 2019;6(4):503-515.; (7) Kariyawasam DST et al. *Genet Med.* 2020;22(3):557-565.; (8) Lin CW, et al. *Pediatr Neurol.* 2015;53(4):293-300.



# To successfully expand NBS to include a new test, PAGs will need to engage with a number of external stakeholders

Please select a country to see the key stakeholders and decision makers that have been identified for NBS

[Denmark](#)

[Italy](#)

[Russia](#)

[Finland](#)

[Netherlands](#)

[Spain](#)

[France](#)

[Norway](#)

[Sweden](#)

[Germany](#)

[Poland](#)

[Switzerland](#)

[Iceland](#)

[Portugal](#)

[Ukraine](#)

[Ireland](#)

[Romania](#)

[United Kingdom](#)



# In addition to this toolkit there are many previously developed materials to support your advocacy work

Resource	Description
<a href="#">SMA Screening Alliance Advocacy tools</a>	A repository of materials developed by the SMA NBS alliance and other organisations (EURORDIS, Screen4Rare and others) to support the case for NBS and launching a call for action across Europe.
<a href="#">Newborn Screening by Genomic Sequencing: Opportunities and Challenges</a>	A journal article covering potential opportunities and challenges genomic sequencing could have for NBS programmes and notes key questions to consider for genomic screening such as routine screening considerations, the ethical, communication, data management, legal, and social implications of genomic screening programmes.
<a href="#">White paper for the inclusion of spinal muscular atrophy in newborn screening</a>	A white paper submitted to key decision makers in Portugal to support the case for NBS for SMA, and has been subsequently used by other countries across Europe.
<a href="#">Rare 2030 Knowledge base: diagnostics</a>	A European Union funded project aiming to gather key insights from experts across Europe on key topics relating to rare diseases, including diagnosis and NBS and calling for European collaboration.
<a href="#">A landscape assessment of newborn screening (NBS) in Europe</a>	Previous research conducted by Charles River Associates examining NBS across Europe and making several policy recommendations to address the key challenges identified in NBS and to support future evidence-based expansion of NBS.
<a href="#">Screen4Care</a>	An ongoing research project aiming to rapidly decrease time to diagnosis for rare diseases by driving the development and uptake of genomic sequencing and genetic testing and aiming to identify patients early through their medical records.
<a href="#">Screen4Rare</a>	A multi-stakeholder initiative launched by IPOPI, ISNS and ESID aimed at exchanging knowledge and best practices on NBS for rare diseases
<a href="#">International Neonatal Screening Day</a>	An initiative to raise awareness of NBS worldwide on the 28 <sup>th</sup> June each year.





# To support the case for NBS a Cost-Effectiveness Model in several European countries has been developed

## What is a cost-effectiveness model and how was it made?



The analysis is a comparison of the costs of 2 scenarios 1) a scenario with NBS and 2) a scenario without NBS, and is specific for SMA<sup>1</sup>



The analysis includes total SMA treatment costs as well as patients' health benefits<sup>1</sup>



The analysis assumes high usage of onasemnogene abeparvovec for presymptomatic patients but also assumes that some patients are treated with other available SMA treatments<sup>1</sup>



With the same research question «*Is NBS cost-effective?*» other institutions can develop a similar analysis

[For further explanation of what a cost effectiveness model is please see a short video by Prof. Laurent Servais](#)

(1) Velikanova, R., et al. *Value Health*. 2022;25(10):1696-1704.



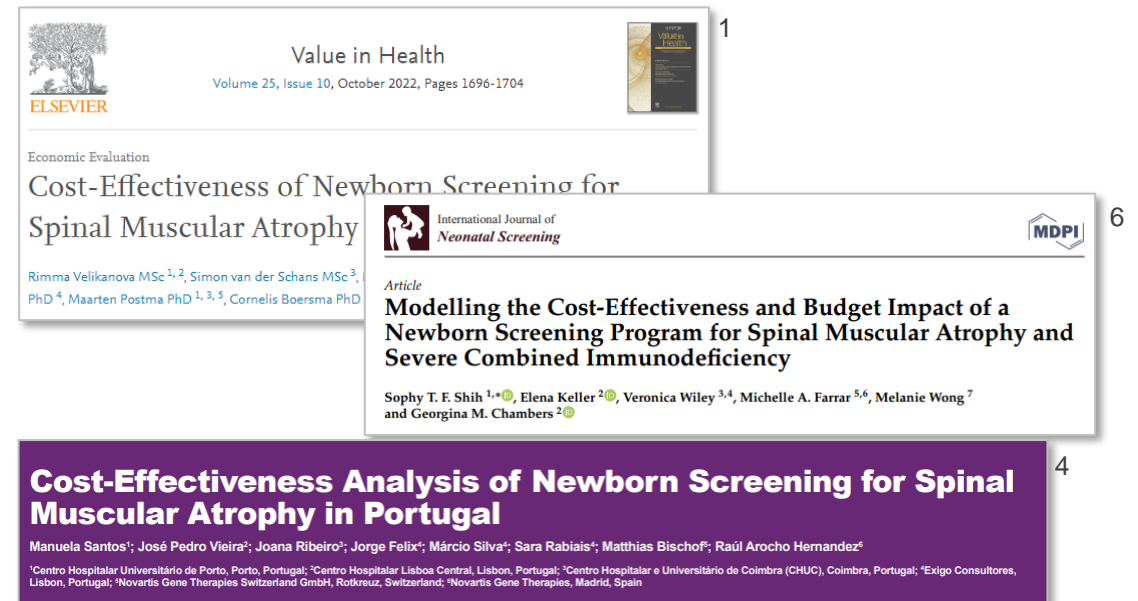
# The model shows that NBS for SMA is a cost-effective use of resources across markets and can save healthcare systems money

## What are the results?

With the availability of NBS for SMA and early treatment, total healthcare costs including hospital admissions, breathing equipment, wheelchair assistance, physical therapy and additional costly healthcare services will be significantly reduced and results in cost savings to the healthcare system.<sup>1</sup>

In all European markets and Japan for which Cost Effectiveness Analysis (CEA) of NBS for SMA was assessed versus a scenario without NBS (including Netherlands<sup>1</sup>, Belgium<sup>2</sup>, Ireland<sup>3</sup>, Portugal<sup>4</sup> and Japan<sup>5</sup>), NBS for SMA is less costly and more effective than a strategy without NBS.

These results have been published in scientific journals and are available at the websites below

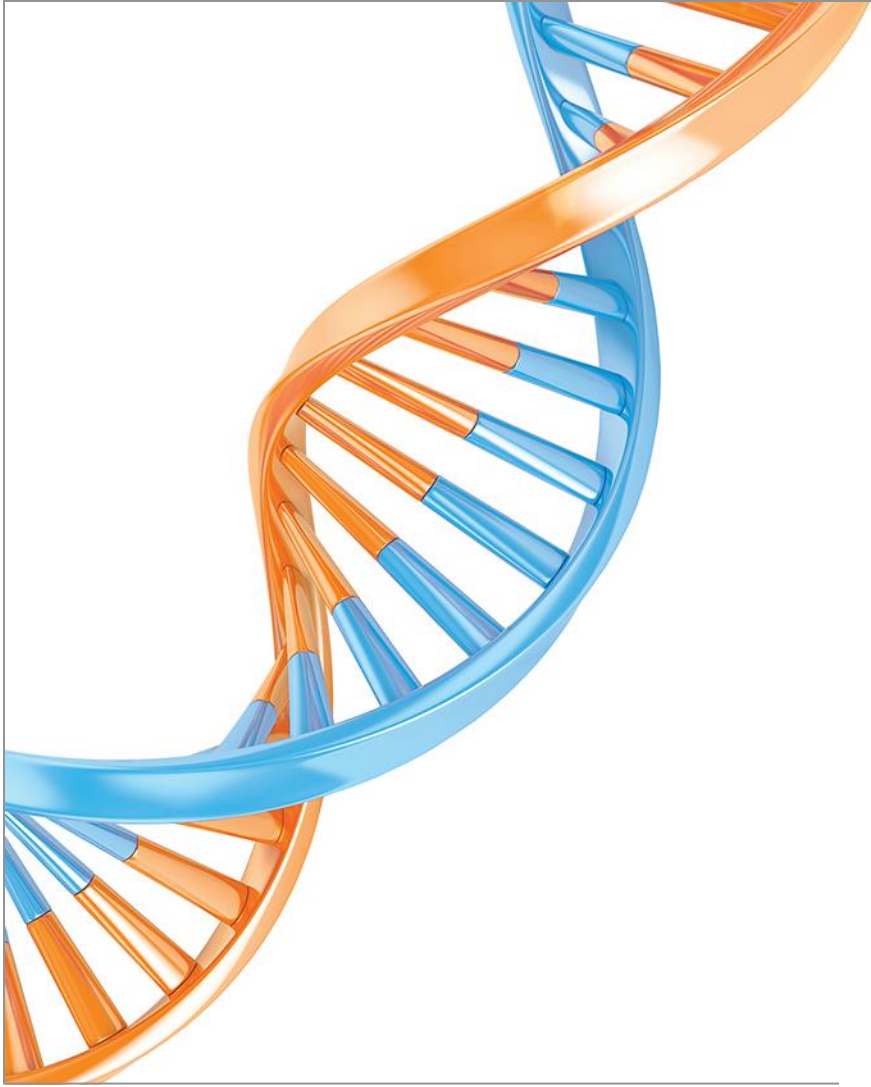


These arguments can be used to demonstrate the benefits of NBS across all markets, including those that do not formally include cost-effectiveness analyses as part of their standard decision-making processes

(1) Velikanova, R., et al. *Value Health*. 2022;25(10):1696-1704.; (2) Dangouloff, T., et al. *Developmental Medicine & Child Neurology*. 2022; Advanced online publication.; (3) Weidlich, D., et al. (2022). *Cost-Effectiveness Analysis of Heel Prick Screening Test for Spinal Muscular Atrophy in Ireland* Available at: [https://novartis.medicalcongressposters.com/FileUpload/QRPDF/SMA%20Europe\\_CEA%20HPST%20Ireland\\_FINAL.pdf](https://novartis.medicalcongressposters.com/FileUpload/QRPDF/SMA%20Europe_CEA%20HPST%20Ireland_FINAL.pdf) (Accessed 14 November 2022);



(4) Santos, M., et al. (2022) *Cost-Effectiveness Analysis of Newborn Screening for Spinal Muscular Atrophy in Portugal*. Available at: [https://novartis.medicalcongressposters.com/FileUpload/QRPDF/SMA%20Europe\\_CEA%20NBS%20Portugal\\_FINAL.pdf](https://novartis.medicalcongressposters.com/FileUpload/QRPDF/SMA%20Europe_CEA%20NBS%20Portugal_FINAL.pdf) (Accessed 14 November 2022); (5) Uda, A., et al. (2022) *Cost-Effectiveness Analysis of Newborn Screening for Spinal Muscular Atrophy in Japan*. Available at: [https://novartis.medicalcongressposters.com/FileUpload/QRPDF/SMA%20Europe\\_CEA%20NBS%20Japan\\_FINAL.pdf](https://novartis.medicalcongressposters.com/FileUpload/QRPDF/SMA%20Europe_CEA%20NBS%20Japan_FINAL.pdf) (Accessed 14 November 2022); (6) Shih, STF., et al. *International Journal of Neonatal Screening*. 20;8(3):45.



## **Key challenges and solutions**

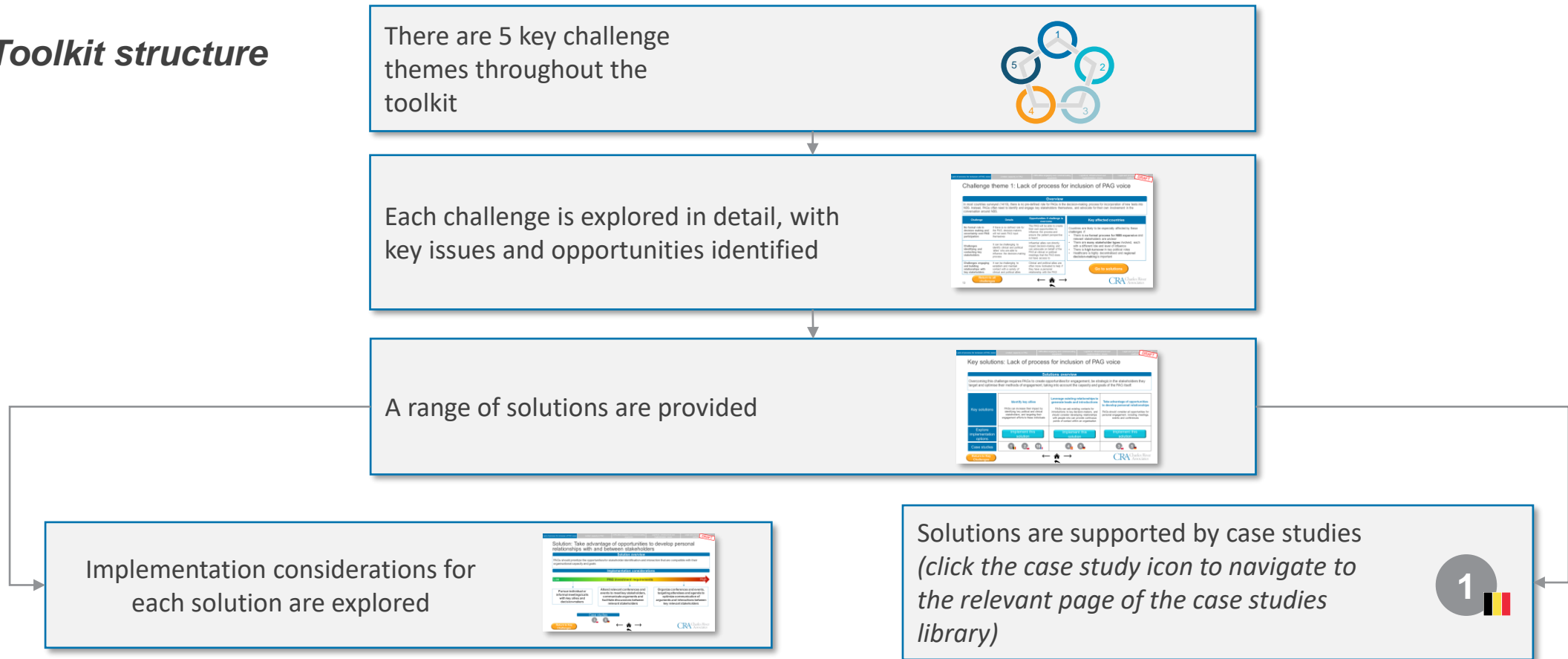


**SMA Europe**

**CRA** Charles River  
Associates

# This toolkit will support PAGs in addressing challenges by proposing solutions based on successful activities carried out by other PAGs

## Toolkit structure



Click here at any time to return to the home page with links to all challenge categories and resource libraries



# Challenge theme 1: Lack of process for inclusion of PAG voice

## Overview

In most countries surveyed (14/18), there is no pre-defined role for PAGs in the decision-making process for incorporation of new tests into NBS. Instead, PAGs often need to identify and engage key stakeholders themselves, and advocate for their own involvement in the conversation around NBS.

Challenge	Details	Opportunities if challenge is overcome	Key affected countries
<b>No formal role in decision making and uncertainty over PAG participation</b>	If there is no defined role for the PAG, decision-makers will not seek PAG input themselves	The PAG will be able to create their own opportunities to influence the process and ensure the patient perspective is heard	Countries are likely to be especially affected by these challenges if: <ul style="list-style-type: none"> <li>• There is <b>no formal process for NBS expansion</b> and relevant stakeholders are unclear</li> <li>• There are <b>many stakeholder types</b> involved, each with a different role and level of influence</li> <li>• There is <b>high turnover</b> in key political roles</li> <li>• Healthcare is highly decentralised and <b>regional decision-making</b> is important</li> </ul>
<b>Challenges identifying and contacting key stakeholders</b>	It can be challenging to identify clinical and political 'allies' who are able to influence the decision-making process	Influential allies can directly impact decision-making and can advocate on behalf of the PAG at clinical or political meetings that the PAG does not have access to	
<b>Challenges engaging and building relationships with key stakeholders</b>	It can be challenging to establish and maintain contact with a variety of clinical and political allies	Clinical and political allies are often more motivated to help if they have a personal relationship with the PAG	

[Go to solutions](#)









[Return to all challenges](#)



# Key solutions: Lack of process for inclusion of PAG voice

## Solutions overview

To overcome these challenges PAGs need to create opportunities for engagement, be strategic in the stakeholders they target and optimise their methods of engagement, taking into account the capacity and goals of the PAG itself. The process for NBS expansion and the key stakeholder involved should be mapped out early to identify key individuals/organisations to develop long-term relationships with

<b>Key solutions</b>	<p><b>Identify key decision-makers</b></p> <p>PAGs can increase their impact by mapping the process, identifying key political and clinical stakeholders, and targeting their engagement efforts to these individuals</p>	<p><b>Leverage existing relationships to generate leads and introductions</b></p> <p>PAGs can ask existing contacts for introductions to key decision-makers, and should consider developing relationships with people who can provide continuous points of contact within an organisation</p>	<p><b>Take advantage of opportunities to develop personal relationships</b></p> <p>PAGs should consider all opportunities for personal engagement, including meetings, events and conferences</p>
<b>Explore implementation options</b>	<p>Implement this solution</p>	<p>Implement this solution</p>	<p>Implement this solution</p>
<b>Case studies</b>	<p>1  2  16  18 </p>	<p>4  5 </p>	<p>3  5 </p>

Return to Key Challenges



# Solution: Identify key allies within stakeholder organisations in order to target engagement efforts appropriately

## Solution overview

Many PAGs have had success through collaborating with a few highly motivated and knowledgeable stakeholders, who can advocate for NBS or have influence over decision-making. PAGs should aim to identify these individuals and target their engagement.

## Implementation considerations

**Is it possible to proactively identify specific individuals who are likely to be engaged and supportive, for example politicians or civil servants with:**

- A relevant medical/health background
- An interest in healthcare policy as evidenced by regular participation in relevant committees and debates, or prior work with PAGs or advocating for patients on policy issues
- Personal/family experience of rare or genetic disorders

**Is it possible to proactively identify specific individuals who are likely to be particularly influential?**

- Clinicians with particular influence or highly relevant experience (e.g. cross-functional expertise in neurology/genetics, current involvement in pilot programs, role in hospital management committee, history of engagement with policy issues)
- Politicians or civil servants who are likely to be involved in their organisation long term, and could provide a constant point of contact
- Politicians, civil servants, clinicians or hospital/laboratory staff with key decision-making power relating to NBS

## Actions

No

Aim to map out the process for NBS in your country to identify potential targets

Contact a wide range of potentially influential stakeholders and follow up with those who respond positively

Yes

Target engagement efforts on these stakeholders

## Case studies



Return to Key Challenges

# Solution: Leverage existing contacts and relationships to generate leads and introductions

## Solution overview

PAGs can improve their chances of engagement with key stakeholders if the initial contact is facilitated through an existing relationship. Within political organisations, it can be beneficial to maintain relationships with people who are likely to keep their jobs through periods of political change (such as key policy advisors or civil servants) in addition to targeting the main decision-makers.

## Implementation considerations

### Potential relationships

What relationships does the PAG have already?

Can these be leveraged to initiate contact with a target stakeholder?

PAG for other diseases/umbrella organisations who have previously engaged successfully with the stakeholder

Clinicians who could facilitate an introduction to hospital management/testing laboratories

Remaining political contacts who could facilitate an introduction to a new key decision-maker after political change/elections

## Case studies

4



5



Return to Key Challenges





# Solution: Take advantage of opportunities to develop personal relationships with and between stakeholders

## Solution overview

PAGs should prioritize the opportunities for stakeholder identification and interaction that are compatible with their organisational capacity and goals

## Implementation considerations



## Case studies



[Return to Key Challenges](#)



# Challenge theme 2: Limited capacity in PAG

## Overview

PAGs are often small organisations and may lack the financial and human resources for large scale advocacy work or political/clinical expertise

Challenge	Details	Opportunities if challenge is overcome	Key affected countries
<b>PAGs are small organisations</b>	Some PAGs have a small number of members, each of whom has limited time to dedicate to the campaign	PAGs can increase their influence through using their time in the most efficient way, or partnering with other groups with similar goals	<p>Countries are likely to be especially affected by these challenges if:</p> <ul style="list-style-type: none"> <li>• The PAG is <b>small, new or has particularly limited funds</b></li> <li>• The process for NBS expansion is <b>particularly complex</b></li> </ul>
<b>PAGs have limited funds</b>	PAGs may be unable to perform activities that require significant funding such as organizing events	PAGs can identify activities that are within their budget and perform these to create impact	
<b>PAGs lack internal expertise on processes relating to NBS</b>	PAGs may not know how to get involved in the NBS process, or be aware of the key political, financial or clinical arguments for the expansion of NBS	PAGs can identify the most promising opportunities for engagement with the process and communicate impactful arguments	

[Go to solutions](#)


[Return to all challenges](#)



# Key solutions: Limited capacity in PAG

## Solutions overview

Overcoming these challenges requires the PAG to both find opportunities to increase their capacity and capabilities, and also to consider ways in which they can maximise the impact of the resources they have available, either through targeted advocacy or by identifying key allies to support common goals they may share

<p>Key solutions</p>	<p><b>Identify organisations with similar goals who may be able to support</b></p> <p>PAGs can increase their capacity and bring in additional expertise by engaging with other organisations</p>	<p><b>Identify resources developed by other groups that can be re-used</b></p> <p>Resources developed by other PAGs can be valuable tools, although groups should consider whether they are applicable to the local healthcare environment</p>	<p><b>Identify knowledge gaps in PAG and reach out to individuals who may be able to fill them</b></p> <p>Bringing experts on board is critical for highly technical issues, but can also save PAGs trying to learn everything themselves</p>
<p>Explore implementation options</p>	<p>Implement this solution</p>	<p>Implement this solution</p>	<p>Implement this solution</p>
<p>Case studies</p>	<p>4  5  6 </p>	<p>11 </p>	<p>1  8  2 </p>

Return to Key Challenges



# Solution: Identify organisations with similar goals who may be able to provide additional support

## Solution overview

Many PAGs have had success through collaborating with other patient groups with similar interests and goals in order to increase their capacity or capabilities, this can take multiple forms, such as financial support or by providing additional volunteers or expertise

## Implementation considerations

### Consider which organisations exist in your country

*Examples include:*

Broad rare disease patient groups

Other specific neuromuscular disease patient groups or groups which can be screened with the same test (e.g. SMA and Primary Immunodeficiency (PID), Severe Combined Immunodeficiency (SCID))

### Prioritise target organisations

*There is a higher chance of success if:*

Your PAG already has a relationship with the target organisation

The organisation already has experience advocating for NBS

The group is considering NBS for other diseases which would benefit from the precedent of SMA screening

### Communicate shared goals

*Ensure the organisation understands that:*

SMA NBS has been successfully implemented in many countries

Testing technologies are well-developed and have been trialled in multiple programmes

There are an increasing number of therapies for rare diseases becoming available and there will be increasing demand in the future to expand NBS programs, expanding NBS can allow new processes and infrastructure to be established to have a more efficient process for NBS expansion in future

## Case studies

4



5



6



Return to Key Challenges

# Solution: Identify resources developed by other groups and re-use for local advocacy where appropriate

## Solution overview

Many PAGs have developed resources during their own advocacy work, which can be shared with PAGs in other countries who may be earlier in the process of BNS expansion. However, you should consider carefully whether the information is applicable in your country.

## Implementation considerations

Identify what type of resource is appropriate for your goal and audience

*Example goals*

Trying to convince a member of parliament to raise the issue of NBS in government

Trying to convince the MoH to implement a NBS program

*Useful resource types*

Patient and clinician testimonies and videos  
Countries with resources: Greece, France

Detailed white papers and clinical/economic evidence  
Countries with resources: Portugal

Consider relevance to local situation

*Select and modify resources carefully to ensure relevance to your local situation*

Select case studies and examples from countries with a similar healthcare system and treatments available where possible

Consider adding any additional local data such as, patient numbers, estimated incidence, number of hospitals/ testing centres etc. to demonstrate the need for NBS and how a programme could be implemented e.g. how many newborns would be tested, how many new cases would you expect in a year, how many maternity centres/neonatal hospitals are there, how many infants are born at home?

## Case studies

11

Return to Key Challenges



# Solution: Identify knowledge gaps in PAG and reach out to individuals who may be able to fill them

## Solution overview

PAGs in several surveyed countries have benefitted from the support of individuals who have specific expertise needed to overcome a challenge. In particular, since PAGs are normally new to NBS advocacy, it can be helpful to engage clinicians, civil servants or policy-makers who are familiar with the processes and procedures involved, and can guide the PAG through the technical aspects of the process.

## Implementation considerations

### Example knowledge gap

We want to propose a pilot but don't know how to design a clinical trial

We have a meeting with a politician but don't know how to explain the clinical data

We want to engage but don't know how the process of NBS expansion works

We want to push forward with a pilot but don't know how to organise the project

### Example supporter

Clinicians or scientists with previous experience of NBS trials or clinical trials or an interest in diagnostic/ the disease area  
Alternatively documents with trial protocols from other countries/regions could be used to support a local trial design

Clinicians with personal interest and relevant expertise, who will be able to communicate clinical arguments to decision-makers convincingly

Politicians/decision-makers with personal interest in rare diseases or NBS, who may be willing to advise the PAG on opportunities for engagement with the process

Technical/ clinical project managers or trial administrators\*

\*note this solution is most relevant to PAGs who are funding/running at least part of the project themselves, and are able to fund additional administrative support

## Case studies



Return to Key Challenges

# Challenge theme 3: Difficulties engaging and communicating arguments

## Overview

PAGs may struggle to communicate the key arguments needed with the different stakeholders involved in decision-making, or may experience push-back against the arguments they present

Challenge	Details	Opportunities if challenge is overcome	Key affected countries
<b>PAGs may not know which key arguments to present to which stakeholder</b>	When multiple stakeholders are involved, it is important to have targeted communication strategies	The PAG can ensure that key information is delivered to the right people, facilitating the decision-making process	<p>Countries are likely to be especially affected by these challenges if:</p> <ul style="list-style-type: none"> <li>• There are <b>many stakeholder types</b> involved, each with a different role and level of influence</li> <li>• There is <b>limited NBS in the country currently</b>, and <b>stakeholders are unfamiliar</b> with the process and benefits</li> <li>• The government or healthcare system is facing <b>other major challenges</b>, particular if related to budget limitations</li> </ul>
<b>Stakeholders may push back with economic, clinical or implementation-based concerns</b>	PAGs may need to quickly develop responses to stakeholder questions in order to maintain momentum	If PAGs are prepared to respond, stakeholder concerns can be effectively addressed	
<b>PAGs need to manage complex relationships between stakeholders and the media</b>	The media can be a powerful tool for advocacy, but interaction needs to be carefully managed to avoid antagonising decision-makers	Effective use of the media can increase public awareness and political pressure without placing 'blame' on decision-makers	

[Go to solutions](#)

[See supporting messages](#)






[Return to all challenges](#)



# Key solutions: Difficulties engaging and communicating arguments

## Solutions overview

Overcoming these challenges requires the PAG to both find opportunities to increase their capacity, and also to consider ways in which they can maximise the impact of the resources they have available by identifying potential areas of pushback they may receive and to prepare appropriate responses tailored to specific stakeholders

Key solutions	<p><b>Consider the underlying motivations and situation of your audience</b></p> <p>Stakeholders such as politicians may consider decisions in the context of their own underlying goals such as re-election – strategic timing and incorporating their goals into arguments for NBS can increase impact</p>	<p><b>Collect compelling evidence and highlight the broader value of NBS</b></p> <p>By understand the types of evidence that are likely to be compelling for decision-makers, PAGs can maximise their impact and chance of success</p>	<p><b>Prepare for push-back</b></p> <p>By being aware of potential objections, including those raised by stakeholders in other countries, PAGs can act, prepare responses and maintain the momentum of the discussion</p>	<p><b>Engage strategically with the media to avoid backlash</b></p> <p>Careful use of the media can increase public awareness and political pressure without damaging relationships between the PAG and key stakeholders</p>
	<p>Explore implementation options</p>	<p>Implement this solution</p>	<p>Implement this solution</p>	<p>Implement this solution</p>
	<p>Case studies</p>	<p>7 </p>	<p>1  2  9 </p>	<p>6 </p>

Return to Key Challenges





# Solution: Consider the underlying motivations of your audience

## Solution overview

Communication of arguments for NBS can be more effective if they are linked to the goals and current situation or context of the audience

## Implementation considerations

### Example goals/situations

The MoH is about to approve or has recently approved a **new treatment for your condition**

The government is **approaching an election**

A **new government** has just been elected

The government has **implemented laws or policies** relating to healthcare access

### Opportunities to increase impact

Highlight the fact that NBS can result in **better outcomes from the approved treatment**, since children can be treated earlier\*

Highlight effective public awareness campaigns and the potential for progress towards NBS to be presented as a 'success' that could **improve public perceptions**

Engage with the technicians who are writing the electoral programme at this stage

Engage with politicians and civil servants who are involved in writing the **governmental agreement**, to ensure NBS is considered in new policies

Quote policies and politicians directly to increase impact of PAG arguments, highlighting their **previous commitments**

\*If the treatment has age restrictions, identifying patients earlier may lead to more patients being treated and therefore higher costs overall – PAGs should prepare for this push-back (see solution 3)

## Case studies

7



## Other relevant challenge themes

Lack of process for inclusion of PAG voice

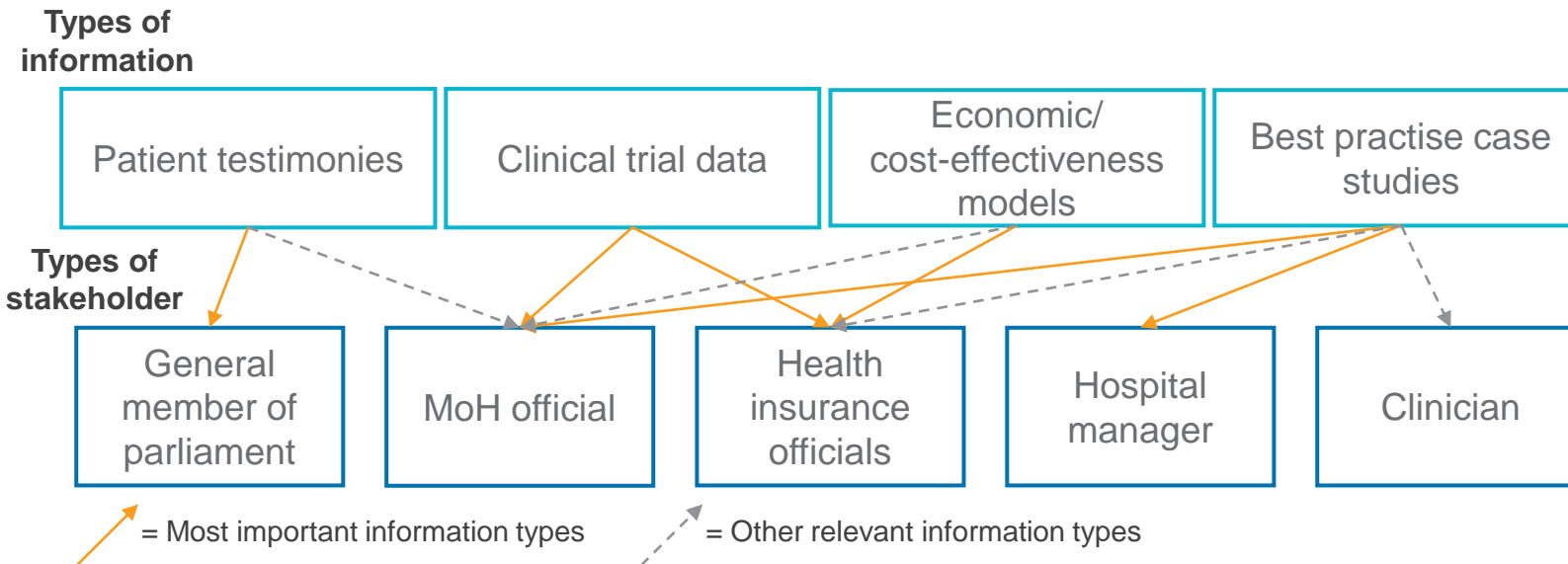
Return to Key Challenges

# Solution: Use data to highlight the broader impacts beyond the patient: to families, health care systems and society

## Solution overview

When communicating with a stakeholder, it is important to find a balance between highlighting the information that is most important to them, but not ignoring the other 'pieces of the puzzle'. Aim to ensure the most relevant information is communicated first, and then provide supporting arguments, particularly relating to the wider context of disease management.

## Implementation considerations



It is often valuable to identify the most important information and put this in the context of the broader management of the condition e.g. social and caregiver costs associated with disability care for children and adults

Note that cost-effectiveness analyses showing that NBS leads to cost savings across the health system are available to support your evidence package.

[https://www.valueinhealthjournal.com/article/S1098-3015\(22\)02065-4/fulltext#secsectitle0140](https://www.valueinhealthjournal.com/article/S1098-3015(22)02065-4/fulltext#secsectitle0140)

## Case studies



Return to Key Challenges

# Solution: Prepare for push-back

## Solution overview

Anticipating possible stakeholder concerns can allow the PAG to prepare effective responses. PAGs should understand the concerns raised in countries with similar healthcare systems, and consider whether their own decision-makers are likely to respond similarly.

## Implementation considerations

Country characteristic	Limited equipment and testing infrastructure	No previous genetic testing for newborns in country, lack of understanding of the test	Limited number of specialist doctors in country
Example objections	<b>“The equipment is too expensive”</b>	<b>“Genetic testing is unethical”</b>	<b>“We can’t handle any more patients”</b>
PAG responses	<ul style="list-style-type: none"> <li>Highlight potential for other tests to be performed on the same equipment in future, to further improve healthcare without significant additional setup cost</li> </ul>	<ul style="list-style-type: none"> <li>In some countries, the PAG may need to proactively explain the concept of genetic tests in NBS, how this differs from pre-natal genetic testing and how the results are only used to make decision about how best to treat the child’s disease</li> </ul>	<ul style="list-style-type: none"> <li>Explain that since all patients will eventually develop symptoms, screening just allows earlier identification and greater potential for access to better treatment outcomes that may reduce the amount of care needed in the long term</li> </ul>
	<ul style="list-style-type: none"> <li>Highlight overall cost savings associated with better management that can help to offset initial setup costs (reduced treatment costs over patient lifetime, reduced social care costs, ability of parents of continue working)</li> </ul>		

## Case studies

6



Return to Key Challenges

# Solution: Engage strategically with the media to avoid backlash

## Solution overview

The media can be a powerful advocacy tool and can help to raise public and political awareness of the need for NBS. However, engagement needs to be strategic, since publicly 'blaming' decision-makers for the current situation can turn them against the campaign.

## Implementation considerations

**Before engaging with the media, consider what your aim is and whether media engagement is the best way to support it.**

**Examples of appropriate media engagement opportunities may include:**

- We are struggling to get any engagement with decision-makers and need to increase pressure
- We have been involved in the successful early treatment of a child and need to publicise the story to ensure it has an impact
- We are trying to fundraise and need to increase public exposure and awareness

If you are already engaged in discussions with decision-makers and feel that progress is being made, it may be better to focus your efforts on effective targeted communication and relationship-building with decision-makers.

**If you do feel that a media campaign will be beneficial, consider your strategy carefully:**

- Try to avoid direct criticism of the health system or any organisations who may be involved in decision-making, since it is important to maintain positive relationships with decision makers, and criticism could lead to fewer opportunities to engage with decision makers
- Utilise individual patient stories, ideally from your own country, to appeal to audiences
- Try to ensure that you have the capacity to maintain any email or social media accounts associated with the campaign in the medium to long term, to avoid losing momentum
- Aim to educate journalists directly, to ensure that the information is presented accurately

## Case studies

10



[Return to Key Challenges](#)

# Challenge theme 4: Logistical, infrastructural and implementation issues

## Overview

In countries with limited previous experience in genetic testing, there may be significant barriers associated with a lack of necessary infrastructure. In some cases, implementation issues may be a major area of push-back during the decision-making process, whereas in other countries they can pose separate challenges after the authorities have agreed to approve a testing programme.

Challenge	Details	Opportunities if challenge is overcome	Key affected countries
<b>Lack of necessary infrastructure in the country</b>	Even countries with established NBS programmes may not have the equipment or expertise in place to easily establish a genetic test as part of the national programme	Establishing the infrastructure for NBS can have significant costs so addressing this issue removes a major economic barrier to implementation	Countries are likely to be especially affected by these challenges if: <ul style="list-style-type: none"> <li>• The <b>current NBS panel is limited</b>, especially if it <b>does not currently include any genetic tests</b></li> <li>• Current NBS tests are conducted at a number of <b>local labs rather than being centralised</b></li> <li>• Healthcare <b>budgets are particularly limited</b></li> </ul>
<b>Lack of expertise in the country</b>		Specialist lab technicians can help to drive decision-making and implementation at national and local levels	
<b>Lack of established logistics for sample processing</b>		Once logistics are in place, national rollout becomes much easier	

[Go to solutions](#)







[Return to all challenges](#)



# Key solutions: Logistical, infrastructural and implementation issues

## Solutions overview

Implementation issues can be difficult for PAGs to address, but it is important to keep this aspect of the process in mind and make the most of any opportunities to increase feasibility

<p>Key solutions</p>	<p><b>Identify what is the infrastructure gaps and aim to identify potential sources of financial support</b></p> <p>PAGs may be able to raise money for key pieces of equipment needed to initiate a pilot, or co-fund a pilot more broadly</p>	<p><b>Aim and plan for scale-up during pilot programs</b></p> <p>Gradually scaling up a pilot programme to include a second hospital or region can be an effective way to establish necessary logistics and demonstrate the feasibility of national roll-out</p>	<p><b>Engage with technical staff</b></p> <p>Engaging with lab staff directly can allow the PAG to understand and address implementation issues; several PAGs have also had success bringing lab staff on board to explain the feasibility of implementing testing to decision-makers</p>
<p>Explore implementation options</p>	<p>Implement this solution</p>	<p>Implement this solution</p>	<p>Implement this solution</p>
<p>Case studies</p>	<p>5  13 </p>	<p>12 </p>	<p>2  3  9 </p>

Return to Key Challenges



# Solution: Identify what is the infrastructure gaps and aim to identify potential sources of financial support

## Solution overview

A lack of equipment and limited budget to expand infrastructure is a common payer objection in countries where limited genetic testing is currently available. Some PAGs surveyed have been able to provide the necessary equipment for a pilot through their regular fundraising efforts or through a targeted campaign.

## Implementation considerations

### 1. Understand the needs

Before committing to this strategy, speak to clinicians, technicians and hospital managers to understand what is needed. Consider:

- How many tests do we need to have capacity for?
- What other genetic or biomarker tests are currently being run, and how will the new testing equipment need to be integrated with existing lab setups? Are appropriate sample already being taken (e.g. bloodspot/ heel prick tests)
- Is there a need for genetic counsellors to enable testing? Do these roles currently exist in sufficient numbers?
- Will testing be run at individual hospitals or at a central lab?
- How will samples be collected and delivered?
- How much time needs to be factored in for set-up before testing can start?
- How would the referral route to specialists work for infants identified by NBS?

### 2. Optimise your fundraising

If a targeted fundraising campaign is required, consider:

- How can we engage with the media to increase awareness but without alienating or 'blaming' policymakers?
- Is there any relevant campaigning or advertising experience within the PAG that we can leverage?
- Can we involve other organisations with similar goals (e.g. campaigning for a joint NBS programme covering multiple conditions)?

## Case studies

5



13



Return to Key Challenges

# Solution: Aim and plan for scale-up during pilot programs

## Solution overview

A pilot programme is a good way to demonstrate the feasibility of a NBS programme. While starting small, a NBS pilot can be broadened over time to include more centres and can provide valuable experience that scaling-up to a full national programme is feasible in your country.

## Implementation considerations

### Situation: Individual regions have a lot of influence over their own healthcare systems

- Focussing on engagement with a single regional government can be the most efficient approach in the early stages of a project.
- Consider whether expansion to additional hospitals within the region is possible
- Be prepared to engage with other regions or countries who may become interested in setting up a pilot once there is a precedent in your region/country

### Situation: Current NBS tests are run at a centralised lab

- If NBS samples are already being tested for other diseases at a centralised lab, the infrastructural barriers to programme expansion are reduced
- PAGs can consider capitalising on this and reach out to additional clinicians and hospital managers, highlighting that there are minimal additional administrative requirements

## Case studies

12

Return to Key Challenges





# Solution: Engage with technical staff

## Solution overview

Laboratory staff can be a valuable ally when advocating for NBS as they can provide clinical, logistical and implementation-based inputs and explanations during discussions. Building a relationship with the technicians doing the testing can also help to reduce potential barriers when trying to scale up the number of tests during a pilot to demonstrate the feasibility of wider roll-out.

## Implementation considerations

### Laboratory staff...

...can act as **credible advocates for the feasibility of implementation** which can be powerful during discussions with decision-makers

...can **support a realistic and feasible trial/pilot design**, reducing the likelihood of implementation problems later on

...can **facilitate the expansion of a pilot** to include additional sites in order to demonstrate feasibility of roll out. However, this is likely to increase lab workload, so a positive relationship can be important in these negotiations

Consider leveraging existing relationships with clinicians or hospital managers who may be able to introduce you when aiming to engage with lab technical staff

## Case studies



Return to Key Challenges

# Challenge theme 5: Legal and procedural delays and inefficiencies

## Overview

In some countries, there may be specific legal barriers that prevent post-natal genetic tests being included in NBS panels. There may also be inherent inefficiencies or delays in the decision-making process that a PAG has little ability to influence

Challenge	Details	Opportunities if challenge is overcome	Key affected countries
There may be laws in place that prevent genetic tests being included in NBS panels	Laws relating to genetic information can cause issues for screening	If there are significant legal issues, resolving these will be critical if screening is to be possible	Countries are likely to be especially affected by these challenges if: <ul style="list-style-type: none"> <li>• The <b>current NBS panel is limited</b>, especially if it <b>does not currently include any genetic tests</b></li> <li>• There is <b>no formal process</b> for inclusion of new tests on the NBS panel</li> </ul>
The decision-making process may be lengthy and inefficient	Decisions relating to public sector health services can take a long time to reach which can be frustrating for PAGs	If the PAG can facilitate the process, your condition may be included on panels sooner and screening targets are more likely to be met	

[Go to solutions](#)





[Return to all challenges](#)



# Key solutions: Legal and procedural delays and inefficiencies

## Solutions overview

Delays and inefficiencies are a common part of decision-making in many countries. The advice throughout this toolkit relating to maximising the impact and efficacy of advocacy work should be applied to facilitate the decision-making process wherever possible. If specific laws are likely to be problematic for adoption of NBS, targeted advocacy may be needed alongside more general NBS advocacy.

Key solutions	<p><b>Identify specific legal barriers and advocate for change</b></p> <p>PAGs may need to first target major legal changes that will be required for progress towards establishment of NBS</p>	<p><b>Continue to make decision-makers aware of the benefits of NBS and the key barriers being faced</b></p> <p>PAGs can ensure that issues around NBS are not forgotten or deprioritized over time by continuing to engage in with the key decision makers</p>
Explore implementation options	<p>Implement this solution</p>	<p>Implement this solution</p>
Case studies	<p>7  14 </p>	<p>17  15 </p>

[Return to Key Challenges](#)



# Solution: Identify specific legal barriers and advocate for change

## Solution overview

In some countries surveyed, specific legal issues have been barriers to approval of NBS. In these cases, PAGs may need to advocate more generally for policy change before launching a specific NBS campaign.

## Implementation considerations

Recommendations:

### Understand the legal barriers

Examine the broader policy landscape (e.g. rare disease, diagnostic, patient rights policies) If necessary, identify stakeholders (e.g. policymakers) who can help to explain the laws and policies that are causing barriers

### Identify and engage new allies

Legislation that prevents genetic screening can be fairly general in scope and is therefore likely to be causing problems for other groups who are advocating on issues relating to genetics. These groups can work together to facilitate change.

### Target the legislation directly

Advocacy efforts need to be specifically targeted to reforming the legislation, although arguments relating to the benefits of NBS can be part of the evidence package

Leverage broader legal frameworks in your country e.g rights for patients with rare diseases etc to provide context and justification for adding NBS

### Use existing policy processes

Members of Parliament can raise Parliamentary questions and suggest Health Committee hearings which can lead to proposals for legal amendments

## Case studies

7



14



Return to Key Challenges

# Solution: Continue to make decision-makers aware of the benefits of NBS and the key barriers being faced

## Solution overview

In some cases, there is little PAGs can do to address delays and inefficiencies within the decision-making process. In these cases, it is important to limit the impact these delays have on overall progress by maintaining consistent communication and pressure to ensure the discussions about NBS does not get side-lined, deprioritised or forgotten.

## Implementation considerations

Aim to maintain:	Purpose	Example activities
Contact with political or clinical allies	By keeping in contact with key allies, PAGs can ensure that when the current issue or delay is resolved, they do not have to re-establish contact and re-engage the stakeholder in order to push forward	<ul style="list-style-type: none"> <li>Send regular updates on PAG activities and disease news to allies</li> </ul>
Awareness of the key issues	By continuing to make their voices heard, PAGs can ensure that issues around NBS are not forgotten, even if no specific progress is currently possible	<ul style="list-style-type: none"> <li>Encourage PAG members to continue contacting local politicians</li> <li>Utilise the media when there is key news to share</li> </ul>
Up-to-date arguments and resources	By ensuring resources are up-to-date, PAGs will be ready to engage in the next stage of the process as soon as progress is made	<ul style="list-style-type: none"> <li>Continue to engage with other PAGs and with scientific progress in diagnosis and treatment of the disease, and ensure that materials contain the most recent data and case studies</li> </ul>

## Case studies

15



Return to Key Challenges



## **Supporting Case Studies**

**CRA** Charles River  
Associates

# Case Study Library

This Case Study Library includes examples of activities that PAGs across Europe have conducted as part of their NBS advocacy work and their experiences

For more information, PAGs are encouraged to reach out to the organizations listed in the case studies

[See here for a list of SMA Europe member organisations with further contact details](#)



**Proactive identification of key political stakeholders can lead to the development of long-term relationships to further the goal of NBS expansion**

<b>Organisation</b>	SMA Belgium
<b>Current status of NBS</b>	Implemented in some regions
<b>Challenge</b>	There are formal regional processes for the expansion of NBS in Belgium, but the role of PAGs remains uncertain, with no clear pathway through which PAGs can engage with the decision-making process
<b>Action</b>	The PAG identified an Member of Parliament with a physiotherapy background who was sensitive to issues around SMA and targeted engagement efforts accordingly.
<b>Impact</b>	The identified stakeholder became a key advocate for the PAG, including raising issues around reimbursement of therapies with the minister and working with the PAG to develop their arguments to include a more holistic view of SMA management across the lifetime of the patients which would resonate with decision-makers

**Challenge themes:**

Lack of process for inclusion of PAG voice

Limited capacity in PAG







## Proactive identification of key clinical allies can provide vital technical support to make the case for NBS

<b>Organisation</b>	Fundacja SMA / SMA Foundation Poland
<b>Current status of NBS</b>	National screening implemented
<b>Challenge</b>	There is no fixed process for expansion of NBS in Poland so there is a need for coordination of stakeholders to develop a comprehensive proposal and evidence package for NBS
<b>Action</b>	The PAG identified a specialist paediatric neurologist/clinical geneticist who was ideally positioned to support both advocacy and implementation, and organized press conferences and meetings to provide a platform where the clinicians could engage with key decision-makers at the MoH
<b>Impact</b>	Key opinion leader clinicians were highly engaged in the process and were able to develop a detailed plan for SMA NBS in collaboration with technical staff at the central lab, which was a key part of the evidence package

### Challenge themes:

Lack of process for inclusion of PAG voice

Logistical, infrastructural and implementation issues

Difficulties engaging and communicating arguments

Limited capacity in PAG





## Engagement with influential stakeholders through conference attendance to facilitate PAGs and key allies to make the case for NBS

<b>Organisation</b>	Fundacja SMA / SMA Foundation Poland
<b>Current status of NBS</b>	National screening implemented
<b>Challenge</b>	There is no fixed process for expansion of NBS in Poland so there is a need for informal engagement and coordination of stakeholders
<b>Action</b>	Members of the PAG attended an academic conference and were able to discuss SMA testing in detail with a key decision maker from the national screening labs, which have significant autonomy in Poland
<b>Impact</b>	The PAG developed a personal relationship with the stakeholder and were able to clearly communicate key arguments directly to a key decision maker

### Challenge themes:

Lack of process for inclusion of PAG voice

Logistical, infrastructural and implementation issues





## Leveraging relationships with other organisations with similar goals can allow PAGs to utilise existing relationships and expertise to accelerate PAG advocacy work

<b>Organisation</b>	FundAME
<b>Current status of NBS</b>	Pilot only
<b>Challenge</b>	Engaging with many regional stakeholders across the country is challenging for the PAG, given limited capacity and variety of organisational structures, so targeting a few national stakeholders is key
<b>Action</b>	The PAG leveraged the greater political power of an umbrella rare disease organisation (FEDER), who already had relationships with national politicians, to set up meetings with key stakeholders to discuss screening for SMA
<b>Impact</b>	The national process to incorporate SMA into the NBS panel has now started, although the necessary regulatory steps are expected to be lengthy

### Challenge themes:

Lack of process for inclusion of PAG voice

Limited capacity in PAG





## The development of personal relationships with key stakeholder can support NBS expansion even when formal processes are in place

<b>Organisation</b>	Initiative SMA
<b>Current status of NBS</b>	National screening implemented
<b>Challenge</b>	There was limited funding available to support an early pilot study for SMA screening in Germany
<b>Action</b>	The PAG were able to leverage personal relationships and interactions with the pharmaceutical industry to generate funding for the pilot, demonstrating a role for this type of engagement in the wider process of NBS expansion even in countries with highly formalized approval processes.
<b>Impact</b>	The pilot was funded – although pilot data was not available at the point of approval in Germany, the results contributed to the pool of European evidence available for other countries to use

### Challenge themes:

Lack of process for inclusion of PAG voice

Limited capacity in PAG

Logistical, infrastructural and implementation issues





## Collaboration with other PAGs can serve to highlight the wider benefits of investment in testing infrastructure beyond just one condition

<b>Organisation</b>	Asociatia SMACARE
<b>Current status of NBS</b>	Pilot in development
<b>Challenge</b>	There is a lack of equipment, staff and testing capacity in the country, which limits the ability to add new tests to the panel
<b>Action</b>	The PAG highlighted the cost-effectiveness of screening as an argument for investment in the infrastructure, and collaborate with the National Alliance of Rare Diseases to highlight potential future cost savings in other diseases which would be accessible with investment in testing capabilities.
<b>Impact</b>	This evidence, as part of the overall campaign, has contributed to ongoing progress towards NBS in a country where there are many infrastructural barriers

### Challenge themes:

Difficulties engaging and communicating arguments

Limited capacity in PAG





## Strategic timing and tailoring of arguments to reflect the local political and legal environment can support engagement where there is limited experience of NBS

<b>Organisation</b>	Asociatia SMACARE
<b>Current status of NBS</b>	Pilot in development
<b>Challenge</b>	There is no clear process for PAG involvement in the decision-making process, and very little NBS is currently implemented so there is limited political precedent or experience
<b>Action</b>	The PAG was strategic with the timing of their engagement efforts, and aimed to push harder at times when politicians were already aware of the issues around SMA, such as the approval of therapies. They also tried to 'speak the language' of the Ministry, leveraging legal frameworks around the right to health in Romania
<b>Impact</b>	PAGs were able to engage effectively with the MoH, leading to a clear understanding of the objections, which can now be addressed

### Challenge themes:

Difficulties engaging and communicating arguments

Legal and procedural delays and inefficiencies





## Utilising expertise and prior experience of NBS pilots and programmes in other countries can support the case for NBS

<b>Organisation</b>	Asociatia SMACARE
<b>Current status of NBS</b>	Pilot in development
<b>Challenge</b>	With limited current testing and significant budget constraints, there is significant pressure on local pilots to be highly successful in order to convince decision-makers to implement screening
<b>Action</b>	The PAG were involved in initial discussions around development of the pilot, and the pilot team developed good relationships with KOLs in screening such as Dr Laurent Servais (Belgium) who was able to support the development of a robust protocol and methodology
<b>Impact</b>	Pilot design ongoing – impact yet to be seen

Challenge themes:

Limited capacity in PAG





## Leveraging existing testing capabilities and protocols developed for other conditions can accelerate implementation of approved programmes

<b>Organisation</b>	Initiative SMA
<b>Current status of NBS</b>	National screening implemented
<b>Challenge</b>	Germany protocols allow testing labs 6 months to begin implementing the test after formal inclusion on the national programme – the PAG was targeting earlier roll-out of screening
<b>Action</b>	The PAG leveraged the recent roll out of testing for SCID and sickle cell prior to SMA, in order to highlight existing capabilities and protocols for PCR-based genetic testing in Germany
<b>Impact</b>	In this case, implementation still took the permitted 6 months

### Challenge themes:

Difficulties engaging and communicating arguments

Logistical, infrastructural and implementation issues







## Effective use of the media can raise significant public and political awareness of NBS, however it is important to maintain the momentum to support full implementation

<b>Organisation</b>	SMA Belgium / 'Team Pia'
<b>Current status of NBS</b>	Implemented in some regions
<b>Challenge</b>	Many diseases were on a 'waiting list' for inclusion on NBS panels, so the PAG needed to convince decision-makers that SMA was a priority
<b>Action</b>	The 'Team Pia' campaign was initiated by parents hoping to crowd-fund treatment with Zolgensma for their baby. The campaign was extremely successful in raising money and massively increased awareness of the disease. SMA Belgium were able to use the same story to campaign for screening, since Pia's case highlighted the problems of late diagnosis leading to treatment ineligibility.
<b>Impact</b>	The campaign raised a lot of money directly, and contributed to the introduction of NBS in Wallonia. However, the web pages and resources to raise awareness and funds have not been maintained, so the Flanders region has not been able to similarly benefit.

### Challenge themes:

Logistical, infrastructural and implementation issues



**Many PAGs have developed their own detailed resources which can be used to develop local arguments for NBS**

<b>Organisation</b>	APN - Associação Portuguesa de Neuromusculares
<b>Current status of NBS</b>	Pilot in development
<b>Challenge</b>	Several PAGs did not have access to a consolidated evidence package that could be used to support discussions
<b>Action</b>	APN developed a white paper with input from scientists, doctors, pharmaceutical companies and the ethics committee and included insights and comparisons from across Europe, Canada and Australia. APN have received requests from other SMA Europe member organisations to share the white paper, and have been able to do so.
<b>Impact</b>	The white paper was shared with the authorities in Portugal and facilitated approval of a pilot study. It is also being used by other PAGs including Greece and Turkey.

**Challenge themes:** Limited capacity in PAG



## Steadily scaling up within a pilot programme is a viable strategy to achieve if full national NBS from day one is difficult to achieve

<b>Organisation</b>	AFM-Téléthon
<b>Current status of NBS</b>	Pilot in development
<b>Challenge</b>	In countries with regional healthcare, it can be challenging to implement a national programme – involving multiple regions or hospitals in the pilot can help to demonstrate the feasibility of integrating multiple centres into the program
<b>Action</b>	AFM-Téléthon have been closely involved in funding and designing the pilot scheme in France. The first phase of the pilot will include two hospitals in Strasbourg and Bordeaux, and the PAG have seen increased interest from other regions now that there is precedent. The hope is to integrate additional regions after the first year of the pilot, and that this approach will make progress towards a national program easier after the pilot.
<b>Impact</b>	Ongoing, impact yet to be determined

### Challenge themes:

Logistical, infrastructural and implementation issues

## Building the supporting infrastructure for diagnosis and treatment can support the case for broader expansion of NBS

<b>Organisation</b>	MDA Hellas
<b>Current status of NBS</b>	No program currently available
<b>Challenge</b>	In Greece, treatment of SMA was limited by a lack of specialist centres where specialist doctors and cross-functional teams could provide the best care for newly diagnosed patients
<b>Action</b>	MDA Hellas worked to set up three paediatric neuromuscular units in hospitals across Greece and are now developing a centre to support patients transitioning into adulthood. The PAG have also worked to fill infrastructural gaps such as patient registries, and have engaged with pharmaceutical companies to increase the number of clinical trials available in Greece.
<b>Impact</b>	There are currently two clinical trials for DMD and one for ALS available through the neuromuscular disease centres. A trial for SMA screening is expected to follow.

### Challenge themes:

Logistical, infrastructural and implementation issues

## When advocating for legal change it is important to identify the broader concerns of politicians and to educate them on the NBS process and the value it can bring

<b>Organisation</b>	AFM-Téléthon
<b>Current status of NBS</b>	Pilot in development
<b>Challenge</b>	A major law regarding genetic information presented a barrier to implementation of genetic NBS in France
<b>Action</b>	AFM-Téléthon were involved in advocating for legal change along with other groups who were affected by this legislation. In discussions with politicians, it was important to clearly explain the process, including highlighting the fact that NBS for SMA would involve testing for a single gene after birth, rather than sequencing full genomes, or pre-natal testing.
<b>Impact</b>	These discussions helped to overcome politicians' fears around genetic testing, which stemmed from a lack of understanding of the process. There was a change in the law in 2021, allowing AFM-Téléthon to progress with planning a pilot scheme for SMA testing.

### Challenge themes:

Legal and procedural delays and inefficiencies

Lack of process for inclusion of PAG voice



## When long delays for NBS approval are expected it is important to continue to make key stakeholders aware of the issues to ensure the issue continues to be raised

<b>Organisation</b>	SMA Ireland
<b>Current status of NBS</b>	Discussions around a possible pilot underway
<b>Challenge</b>	In Ireland, changes in the health system typically happen slowly and SMA Ireland are a small organisation with limited leverage to push forward with a pilot independently
<b>Action</b>	SMA Ireland called on members to write to their individual MPs and public representatives, raising questions around SMA screening and management more generally
<b>Impact</b>	The PAG were able to maintain contact with politicians and ensure awareness of the issues continued despite inertia in the health system

### Challenge themes:

Legal and procedural delays and inefficiencies



## Identification of key political allies with similar interests can be highly impactful in overcoming legal/ political barriers to NBS

<b>Organisation</b>	AFM-Téléthon
<b>Current status of NBS</b>	Pilot in development
<b>Challenge</b>	AFM-Téléthon faced significant legal barriers to implementation of testing, and until recently had been unable to progress with planning a pilot study
<b>Action</b>	The PAG identified a small department within the MoH which was dedicated to rare diseases. This department had developed a national plan on rare disease, which included objectives relating to diagnosis and NBS, which the PAG was able to leverage in discussions. The department was also able to facilitate introductions to other parts of the Ministry and help to bring key stakeholders together.
<b>Impact</b>	Advocacy and engagement with policy-makers contributed to a change in the law in 2021, allowing AFM-Téléthon to progress with planning a pilot scheme for SMA testing.

### Challenge themes:

Legal and procedural delays and inefficiencies

Lack of process for inclusion of PAG voice





## Raising awareness of the key issues with politicians and the general public can support in overcoming potential legal barriers to NBS

<b>Organisation</b>	SMA Österreich
<b>Current status of NBS</b>	National pilot, originally Jun'21 to May'22; with an extension until end of 2022. A transition into a permanent programme is being planned for January 2023.
<b>Challenge</b>	In Austria, PAGs have very limited influence on changes in the health system and relevant laws, with a national law on the use of genetic testing in humans seen as a key barrier to NBS for SMA SMA Österreich are a small and recently established organisation with limited leverage to push forward with an NBS pilot independently.
<b>Action</b>	The PAG supported local clinical experts in implementation of an NBS pilot by submitting petitions to the MoH and raising awareness of both SMA and NBS with the general public through social media campaigns
<b>Impact</b>	The PAG were able to raise awareness of the importance of SMA early diagnosis & treatment in the public and at MoH level to support the political case for approval of a NBS pilot

Challenge themes:

Legal and procedural delays and inefficiencies







## Raising awareness and communicating with a wide variety of stakeholders can generate increased support for the addition of new tests

<b>Organisation</b>	FundAME
<b>Current status of NBS</b>	Pilot only
<b>Challenge</b>	The PAG are aiming to incorporate screening into the national plan but involving many regional stakeholders from across the country is challenging
<b>Action</b>	Awareness raising, information and communication meetings requesting the implementation of screening with the ministry's screening department, national congressmen, regional deputies, national ombudsman. Support and meetings with the various national pilot/study teams. Mediation with industry to solve support and funding.
<b>Impact</b>	The national process to incorporate SMA into the NBS panel has now started, although the necessary regulatory steps are expected to be lengthy. A resolution proposal has been achieved in the national debate which was supported.

### Challenge themes:

Logistical, infrastructural and implementation issues

Difficulties engaging and communicating arguments

Limited capacity in PAG





## **Appendix**

Key stakeholders in NBS decision making



**SMA Europe**

**CRA** Charles River  
Associates

# Key Stakeholders: DENMARK



Stakeholder	Role	Description	Impact on decision making
<u><a href="#">Sundhedsministeriet</a></u> <i>Ministry of Health</i>	MoH	Political responsibility for organizing Healthcare in Denmark.	High
<u><a href="#">Statens Serum Institut (SSI)</a></u> <i>State Serum Institute</i>	Public health Agency	Responsible for implementation and management of NBS.	High
<u><a href="#">Sundhedsstyrelsen</a></u> <i>Danish Health Authority</i>	Public health Agency	Responsible for screening programs (not only NBS) in Denmark.	High
<u><a href="#">Dansk Pædiatrisk Selskab (DPS)</a></u> <i>Danish Paediatric Society (DPS)</i>	Medical society	Professional association of Danish paediatricians which have actively lobbied for including SMA in the NBS.	Moderate



High



Moderate



Low





# Key Stakeholders: FINLAND

Stakeholder	Role	Description	Impact on decision making
<u><a href="#">Synnynnäisten aineenvaihduntasairauksien seulontakeskus - Saske</a></u> <i>The Saske Screening Center for inborn errors of metabolism</i>	The national centre for NBS	The unit coordinating and performing all NBS lab tests in Finland (located in Turku).	High
<u><a href="#">Sosiaali- Ja Terveysministeriö</a></u> <i>Ministry of Social Affairs and Health</i>	MoH	Responsibility for organizing healthcare including screening activities. Provide high level guidance on NBS and is currently in the process of reforming their NBS process.	High





# Key Stakeholders: FRANCE

Stakeholder	Role	Description	Impact on decision making
<a href="#"><u>Haute Autorité de Santé (HAS)</u></a> <i>High Authority for Health</i>	MoH	National Authority for Health, responsible for disease evaluation and final approval.	High
<a href="#"><u>Centre National de Coordination de Dépistage Néonatal (CNCNDN)</u></a> <i>The National Neonatal Screening Coordination Center</i>	Screening centre (National)	Responsible for running the national NBS program.	Moderate
Centre Régional de Dépistage Néonatal (CRDN) <i>The Regional Neonatal Screening Coordination Center</i>	Screening centres (Regional)	Independent centres in each region responsible for implementing national testing procedures.	Low
<a href="#"><u>ECLAS (ensemble contre l'amyotrophie spinale de type 1)</u></a>	Patient Organization	Type 1 SMA dedicated PAG. Consulted by National Authority for Health (HAS).	Moderate
<a href="#"><u>Familles SMA</u></a>	Patient Organization	Type 2 and 3 SMA dedicated PAG. Consulted by National Authority for Health (HAS).	Moderate
<a href="#"><u>AFM telethon</u></a>	Patient Organization	Neuromuscular Disease dedicated PAG. Consulted by National Authority for Health (HAS). Public affair department of AFM is in contact with French Government.	Moderate



High



Moderate



Low





# Key Stakeholders: GERMANY

Stakeholder	Role	Description	Impact on decision making
<a href="#">Gemeinsamer Bundesausschuss (G-BA)</a> <i>Joint Federal Committee</i>	HTA body	Responsible for full evaluation of any NBS programme.	High
<a href="#">Bundesministerium für Gesundheit (BMG)</a> <i>Federal Ministry of Health</i>	MoH	Final political decision-maker.	Moderate
<a href="#">Deutsche Gesellschaft für Muskelkranke eV – DGM</a> <i>German Society for Muscular Diseases</i>	PAG	Umbrella organisation for neuromuscular diseases (NMD).	Within NMD



High



Moderate



Low



# Key Stakeholders: ICELAND



Stakeholder	Role	Description	Impact on decision making
<a href="#"><u>Embætti landlæknis</u></a> <i>The Directorate of Health</i>	MoH	Overall responsibility in organizing healthcare in Iceland including screening programs.	



High



Moderate



Low



# Key Stakeholders: IRELAND



Stakeholder	Role	Description	Impact on decision making
<a href="#">National Screening Advisory Committee (NSAC)</a>	MoH advisory group	Independent advisory group for the MoH covering all screening services including NBS	High
<a href="#">National Newborn Bloodspot Screening Laboratory (NNBSL)</a>	Screening centre	National laboratory for conducting newborn screening	Moderate







# Key Stakeholders: ITALY

Stakeholder	Role	Description	Impact on decision making
<a href="#">Centro di coordinamento per lo screening neonatale</a> <i>Coordination Centre for Newborn Screening</i>	Evaluation	Responsible for performing disease evaluation for national NBS (part of the national institute of health/ <i>Istituto Superiore di Santità</i> ).	High
<a href="#">Ministero della Salute</a> <i>Ministry of Health</i>	MoH	Provides final approval for NBS expansion.	High
Regional health systems	Implementation	Responsible for implementing tests according to regional infrastructure capabilities.	Moderate



# Key Stakeholders: NETHERLANDS



Stakeholder	Role	Description	Impact on decision making
<a href="#">De Gezondheidsraad</a> <i>Health Council</i>	MoH Advising Committee	An independent scientific advisory committee which advises the MoH on public health and healthcare research, including advising the MoH to include new conditions, such as SMA, in a national screening programme.	High
<a href="#">Rijksinstituut voor Volksgezondheid en Milieu (RIVM)</a> <i>National Institute for Health and Environment</i>	National public health agency	Conducts feasibility studies on the implementation of NBS, as seen with SMA (see link).	High



High



Moderate



Low





# Key Stakeholders: NORWAY

Stakeholder	Role	Description	Impact on decision making
<a href="#"><u>Folkehelseinstituttet</u></a> <i>Norwegian Institute of Public Health</i>	Institute within MoH	Responsible for evaluation of applications for NBS.	High
<a href="#"><u>Helse- og omsorgsdepartementet</u></a> <i>Ministry of Health and Care Services</i>	MoH	Responsible for final approval of an NBS programme.	High
<a href="#"><u>Oslo universitetssykehus</u></a> <i>Oslo University Hospital</i>	Unit responsible for NBS in Norway.	A single lab processes the results for NBS in Norway and therefore have a central role in suggesting new diseases to be included in the NBS.	High
<a href="#"><u>Helsedirektoratet</u></a> <i>Health Directorate</i>	Health Authority	Managing the process after a proposal is submitted and prior to approval.	High



High



Moderate



Low



# Key Stakeholders: POLAND



Stakeholder	Role	Description	Impact on decision making
<a href="#">Agency for Health Technology Assessment and Tariff System (AOTMiT)</a>	HTA body	Responsible for disease evaluation for conditions for NBS	High
<a href="#">Ministerstwo Zdrowia</a> <i>Ministry of Health</i>	MoH	Responsible for providing final approval for NBS expansion.	High
<a href="#">Polskie Towarzystwo Chorób Nerwowo-Mięśniowych</a> <i>Polish Neuromuscular Diseases Association</i>	Medical and patient organisation	Umbrella organisation for neuromuscular diseases	Moderate
<a href="#">Instytut Matki i Dziecka</a> <i>Institute of Mother and Child</i>	Screening Laboratory	National laboratory responsible for processing and reporting NBS test results	Moderate

Note there is no centralised screening lab in Poland





# Key Stakeholders: Portugal

Stakeholder	Role	Description	Impact on decision making
<a href="#"><u>Programa Nacional de Rastreio Neonatal (PNRN)</u></a> <i>National Neonatal Screening Programme</i>	NBS programme organisation	Steering committee for decision making – final approval for inclusion of a disease on the panel comes from the President of the PNRN.	High
<a href="#"><u>Instituto Nacional de Saúde Dr. Ricardo Jorge (INSA)</u></a>	MoH	Public institution under the MoH with autonomy in decision making for diagnostics.	High
<a href="#"><u>Associação Portuguesa de Neuromusculares – APN</u></a>	PAG	Portuguese Neuromuscular Diseases Patient Association.	High
<a href="#"><u>Ministério da Saúde</u></a> <i>Ministry of Health</i>	Minister of health	The MoH is ultimately responsible for approving the budget for an NBS programme.	High
Neonatal Screening, Metabolism and Genetics Unit, Instituto Nacional de Saúde Doutor Ricardo Jorge	Screening lab	Single centralised screening lab for NBS within INSA.	Moderate
<a href="#"><u>SPEDNM</u></a>	Medical Society	Responsible for the submission to INSA of the dossier with the clinical data that supported the national NBS for SMA.	Moderate



# Key Stakeholders: ROMANIA



Stakeholder	Role	Description	Impact on decision making
<u><a href="#">Ministerul Sănătății</a></u> <i>Ministry of Health</i>	MoH	The MoH is the ultimate decision maker in implementation of national screening programs in Romania.	High
<u><a href="#">Comisia De Genetică Medicală</a></u> The Genetic Diseases Committee of the MoH	Specialty Committee	The Committee has a very important advisory role within MoH and their advice is sought by the MoH when new screening programmes are considered.	Moderate
<u><a href="#">Alianța Națională pentru Boli Rare din România</a></u> <i>The Romanian National Alliance for Rare Diseases</i>	PAG umbrella association	The national alliance provides key advocacy on the need for the expansion of NBS in Romania (currently there are only 2/3 disease routinely screened in the country).	Moderate
<u><a href="#">Societatea Romana de Neurologie Pediatrica (SRNP)</a></u> <i>Romanian Society of Pediatric Neurology</i>	Professional Society	The RSPN can be an advocate in supporting the need for NBS in SMA / neuromuscular diseases.	Low



High



Moderate



Low





# Key Stakeholders: RUSSIA

Stakeholder	Role	Description	Impact on decision making
<a href="#">Медико-генетический научный центр имени академика Н.П. Бочкова</a> <i>Medical Genetic Research Centre</i>	Research Centre	Federal research institute, founded with the Ministry of Science and Higher Education and responsible for disease evaluation of new conditions for NBS.	High
<a href="#">Правительство России официальный сайт (government.ru)</a> <i>Government of the RF</i>	Government	Instructs MOH to develop proposal and provide final approval	High
<a href="#">Министерство здравоохранения</a> <i>Ministry of Health</i>	MoH	Prepares proposal for NBS programmes Minister, Deputy Minister, Head of Public Health and Maternal and Child Care Department	High
<a href="#">Министерство финансов</a> <i>Ministry of Finance</i>	Government	Makes decisions on the budget allocated to an NBS programme Key stakeholders: Deputy Minister, Department of Budget Policy for Social Sector and Science	Moderate



High



Moderate



Low





# Key Stakeholders: SPAIN

Stakeholder	Role	Description	Impact on decision making
<a href="#">Agencia de Evaluación de Tecnologías Sanitarias</a> <i>Health Technology Assessment Agency</i>	HTA body	Responsible for performing disease evaluation.	High
Autonomous regional health authorities eg. Agència De Qualitat i Avaluació Sanitàries de Catalunya (AQUAS)	Health Authorities	Determines local NBS panel composition.	High
<a href="#">Ministerio de Sanidad</a> <i>Ministry of Health</i>	MoH	Provides final approval for NBS expansions.	Moderate
<a href="#">Federación ASEM</a>	PAG	Umbrella organisation for neuromuscular diseases.	Low







# Key Stakeholders: SWEDEN

Stakeholder	Role	Description	Impact on decision making
<u><a href="#">Nationella screeningrådet</a></u> <i>National Screening Council</i>	Assessing Committee	Responsible for the evaluation of new conditions for NBS. The committee consists of politicians, clinical experts and HTA representatives.	High
<u><a href="#">Socialstyrelsen</a></u> <i>National Board of Health and Welfare</i>	MoH	Responsible for final approval of any new NBS programme.	High
Individual healthcare regions	Implementation	Responsible for deciding whether to implement a recommended test in each region.	High





# Key Stakeholders: SWITZERLAND

Stakeholder	Role	Description	Impact on decision making
<a href="#">Office fédéral de la santé publique/ Bundesamt für Gesundheit</a> <i>Federal Office of Public Health</i>	MoH	Responsible for disease evaluation and final approval, there are currently no specific screening committee within the FOPH/BAG	High
<a href="#">Kinderspital Zürich</a> <i>Zurich Children's hospital</i>	Screening lab	Single centralised screening laboratory for Switzerland	Moderate



High



Moderate



Low





# Key Stakeholders: UKRAINE

Stakeholder	Role	Description	Impact on decision making
<u>Охматдиту</u> <i>Specialist hospital established by the MoH</i>	Hospital	A centre established by the MoH that provides highly specialized medical care to patients with rare diseases and is the main centre to diagnose and verify tests for rare diseases	High
<u>Комітету з питань здоров'я нації, медичної допомоги та медичного страхування</u> <i>Committee on the Health of the Nation, Health Care and Health Insurance</i>	MoH group	Establishes the laws and regulations around NBS and the budget required for a programme. The work of the Committee is aimed at developing and improving legislation in the field of health care, reforming the health care system, and creating the legal conditions necessary to improve the availability and quality of medical care for the population.	High



High



Moderate



Low





# Key Stakeholders: United Kingdom

Stakeholder	Role	Description	Impact on decision making
<a href="#">National Screening Committee</a>	Advisor to MoH	Responsible for disease evaluation, supports implementation	High
<a href="#">Department of Health and Social Care / Public Health England</a>	MoH	Ministers give final approval, subsequent approval needed from respective ministers in devolved nations	Moderate
<a href="#">NHS</a>	Public health system	Also required to approve changes to screening programmes	Moderate
<a href="#">Muscular Dystrophy UK</a>	PAG	Umbrella patient group for neuromuscular diseases (NMD)	Within NMD
<a href="#">UKAS</a>	Accreditation body	Approves NBS laboratories to conduct screening	Low



High



Moderate



Low

