Power Dynamics in Newborn Screening

Master thesis submitted to the University of Bern

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Foreword

Writing this master thesis has been a challenging yet rewarding journey, and I am incredibly grateful for the support and guidance I have received along the way. First, I would like to thank my supervisor, Prof. Dr. Rudolf Blankart for his support. His expertise has shaped the direction and quality of this research. I would also like to thank Sandra Gillner for her support and assistance. Her commitment and feedback have been invaluable. Furthermore, I am grateful to my interview partners and the great conversations I was able to have.

Abstract

Newborn screening (NBS) has been spreading around the world since the 1960s. In various countries, there are great differences in which diseases are included in the screening panel. In recent years, spinal muscular atrophy (SMA) has been included in screening panels in some countries, while others are still in the process of decision-making. Two of these countries are Switzerland and Ireland. Both countries have applied for the inclusion of SMA and a decision is expected before the end of 2023. This thesis examines SMA as an illustrative case to highlight the contrasting policy-making processes between Switzerland and Ireland. It delves into the identification of key actors involved and explores the influence of power dynamics on the overall decision-making process. For the thesis, interviews were conducted with experts in both countries who participate in the policy-making process. It was found that Switzerland and Ireland differ both in their actors and in their process for including new diseases in NBS. While Switzerland follows a clearly defined application process, where non-state actors have almost no influence, non-state actors in Ireland are much more integrated in the decision-making process. Furthermore, in Switzerland, the laboratory responsible for NBS is much more involved in the process.

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List of abbreviations

AL	Analysenliste (Analysis list)
BAG	Bundesamt für Gesundheit (Federal office of public health)
CHI	Childrens hospital Ireland
DBS	Dry blood spot
EAG	Expert advisory group
EAMGK	Eidgenössische Kommission für Analysen, Mittel und Gegenstände (Federal Commission for Analyses, Means and Objects)
GPs	General practitioners
GUMEK	Eidgenössische Kommission für genetische Untersuchungen beim Menschen (Federal Commission for Genetic Testing in Humans)
GUMG	Bundesgesetz über genetische Untersuchungen beim Menschen (Federal Act on Genetic Testing in Humans)
GUMV	Verordnung über genetische Untersuchungen beim Menschen (Ordinance on Genetic Testing in Humans)
HIQA	Health Information and Quality Authority
HSE	Health service executive
HTA	Health technology assessment
KVG	Bundesgesetz über die Krankenversicherung (Federal Health Insurance Act)
KVL	Krankenpflege-Leistungsverordnung (Nursing Service Ordinance)
KVV	Verordnung über die Krankenversicherung (Health Insurance Ordinance)
NBS	Newborn screening
NEK	Nationale Ethik Kommission (National Ethics Commission)
NNBSL	National Newborn Bloodspot Screening Laboratory
NSAC	National screening advisory committee
PCR	polymerase chain reaction
PKU	Phenylketonuria
SCID	Severe Combined Immunodeficiency
SMA	Spinal muscular atrophy
SMN	Survival motor neuron
TD	Teachta Dàla (members of lower house of Irish Parliament)

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1 Introduction

1.1 Newborn Screening: A vital public health tool

Newborn screening (NBS) is a vital public health initiative designed to identify genetic or metabolic disorders in infants. It is currently one of the most effective population-based measures of secondary prevention in childhood (Nennstiel, 2021). The primary objective of NBS is to detect conditions at an early stage, with the intention of weakening potential health implications and delivering timely interventions. This proactive approach greatly enhances the prospects of a healthier future for these newborns, while also providing the opportunity to treat certain diseases in an asymptomatic stage, thereby improving population health (Remec et al., 2021). Utilizing methods such as blood tests or other screening techniques, NBS assumes a central role in the prompt identification and intervention, presenting invaluable possibilities for effective treatment and comprehensive management of conditions that might not manifest at birth.

1.2 Research problem and research question

As NBS programs continue to expand worldwide, countries have devised distinct policies to tackle the challenges associated with their implementation. Among these challenges is the decision-making process concerning the inclusion of specific conditions in the screening panel, as well as the consideration of whether diseases should be included in the screening panel even if they are not treatable. This process of inclusion is influenced by various factors. Power plays a significant role in shaping policy processes and outcomes. It influences these processes through various means, including the dynamics between different actors, the establishment of trust, and the deliberate inclusion and exclusion of certain issues or individuals by policy-makers (Mwisongo, Nabyonga-Orem, Yao, & Dovlo, 2016). However, despite its evident benefits in comprehending health policy dynamics and formulating efficient strategies for successful implementation, the explicit examination of power in literature concerning health policies remains limited. A deeper understanding of health policy can be achieved by exploring power relations, ultimately paving the way for the development of more effective measures and interventions (Erasmus & Gilson, 2008).

In recent years, the inclusion of Spinal Muscular Atrophy (SMA) in the NBS panel has gained significant attention due to the availability of new treatments and potential benefits

for affected infants. SMA is a genetic disorder characterized by the absence of a protein called functional survival motor neuron (SMN), due to an autosomal recessive inheritance pattern (Dangouloff et al., 2021). The SMN protein has, among other things, a central role in the development of the peripheral nervous system in the last months of pregnancy and the first months of life. The absence of the SMN protein leads to the early demise of motoneurons, which are nerve cells, responsible for muscular movement (Müller-Felber et al., 2021). The extension of NBS panels to include SMA constitutes the first primary genetic screening program. The timing of when the treatment for SMA is initiated is crucial for its clinical effectiveness (Müller-Felber et al., 2021). For this reason, there are worldwide efforts to include SMA in the general NBS. The policy-making for the inclusion of SMA in the screening panel does differ across countries. Some countries, like for instance Germany, already have included SMA in the NBS panel. Other countries are in the middle of the implementation process.

For instance, in Switzerland the Paediatric Neuromuscular Reference Centres (Myosuisse), in close collaboration with the Swiss laboratory responsible for NBS, have submitted an application for the inclusion of SMA in the NBS panel to the Federal Office of Public Health, Bundesamt für Gesundheit (BAG) in February 2023 (Schweizerische Muskelgesellschaft, 2023). The same goes for Ireland, where an application has been submitted and a health-economy analysis is under way and expected to be completed in 2023. In Ireland the National Screening Advisory Committee (NSAC) is an independent advisory committee which advises the Minister and Department of Health on population-based screening program. In December of 2022, the NSAC recommended to the Minister of Health to include a group of conditions to the National NBS programme. In their recommendation, the committee identified SMA as the next priority condition to undergo a thorough Health Technology Assessment (HTA) conducted by the Health Information and Quality Authority (HIQA) (National Screening Advisory Committee, 2022).

To better understand the power dynamics associated with the implementation of new disorders in the NBS panel, this thesis will focus on Switzerland and Ireland. These countries have been chosen due to their parallel stage of SMA implementation in their respective NBS panels. The comparative analysis of Switzerland and Ireland can provide valuable insights for policy development and decision-making processes in the field of NBS.

Switzerland and Ireland have healthcare systems with notable distinctions in their structures, financing, and delivery of services. In Switzerland, mandatory health insurance is a fundamental aspect, ensuring that every resident possesses coverage. The Swiss system embraces a combination of public and private healthcare providers, employing a decentralized approach where cantons[1] hold considerable autonomy in managing and organizing healthcare services. Private insurance companies offer a variety of coverage options, granting individuals the freedom to select their provider. This competitive environment fosters diverse choices for insured individuals.

Ireland follows a predominantly public healthcare system. The Health Service Executive (HSE) takes charge of providing and overseeing healthcare services in the country. The system's funding comes from general taxation and user charges. While public healthcare is the main coverage option, private health insurance is also prevalent, and many individuals have both public and private coverage. Public healthcare services in Ireland are delivered through a network of public hospitals and primary care centers. Accessing specialist care typically necessitates referrals from primary care physicians, underscoring the importance of general practitioners as the gateway to specialized services.

These differing healthcare systems highlight variations in terms of insurance coverage, healthcare financing, and the balance between public and private sector involvement. Understanding these differences is essential when examining the implementation of new disorders in NBS panels, as it helps to identify potential factors influencing decision-making, resource allocation, and stakeholder involvement within each system.

The research question of this master thesis is going to be: *How do power dynamics shape the decision-making processes of including new disorders in newborn screening panels in Switzerland and Ireland and who are relevant actors in this process?*

1.3 Significance of study

Given the improved diagnostic-analytical possibilities and new therapeutic options for serious congenital diseases, doctors, patient organisations and politicians are calling for

[1] Cantons are constituent states of the Swiss Confederation

NBS to be extended to an increasing number of target diseases (Nennstiel, 2021). In light of this pressing demand, this thesis on power dynamics in NBS, specifically the inclusion of new diseases to the screening panel, holds great significance. By investigating the power dynamics at play during the decision-making process of expanding the screening panel, this research sheds light on the complexities involved in balancing medical advancements, ethical considerations, and resource allocation. Understanding these dynamics is crucial to ensure the optimal implementation and effectiveness of NBS programs.

1.4 Objective of the study

This thesis focuses on examining power dynamics within the context of NBS, with a specific emphasis on the process of expanding the NBS panel. To illustrate this, the inclusion of SMA in NBS serves as a case study. The core objective of this study is to gain a understanding of the power dynamics that come into play during the policy-making process for the inclusion of a new disease in the NBS panel. The comparative approach between Switzerland and Ireland serves as a framework to provide insights into different strategies and approaches that may influence the policy-making process. It enables the identification of similarities, differences, and potential best practices that can enhance the understanding of power dynamics within NBS policy making. By unraveling these dynamics, the research aims to contribute to a deeper understanding of how decisions are made, power is distributed, and various factors come into play during the expansion of the NBS panel.

2 Literature review

2.1 Background of newborn screening

NBS is a vital public health initiative designed to identify genetic or metabolic disorders in infants. The primary objective of NBS is to detect these conditions at an early stage, with the intention of minimizing potential health implications and delivering timely interventions. This proactive approach not only greatly enhances the prospects of a healthier and more promising future for newborns but also leads to an increase in population health, provides cost efficiency, and enables the treatment of certain diseases even before they become symptomatic (Woerner et al., 2021). Utilizing methods such as

blood tests or other screening techniques, NBS assumes a central role in the identification and intervention, presenting invaluable possibilities for effective treatment and management of conditions that might not manifest at birth. As NBS policies continue to evolve, the inclusion of SMA represents a significant stride forward in ensuring timely identification and proactive intervention for this genetic disorder, illuminating a path towards improved outcomes for affected infants.

In Europe, NBS via dried blood spot (DBS) was first implemented in the 1960s with the screening for phenylketonuria (PKU). The list of screened conditions grew over time, due to the introduction of new technologies, as tandem mass spectrometry and molecular technologies, which made it possible to screen for 40-50 conditions using a single blood spot (Loeber et al., 2021). When it comes to including new diseases in the screening panel, the ethical requirements must be examined in addition to the evidence (Nennstiel, 2021). The fundamental road map for creating a screening policy that takes into account prevalence, diagnosis, treatability, etc. is the Wilson and Younger criteria from 1968 (Loeber et al., 2021). These criteria are still applicable today and some countries, such as Germany based their screening criteria on these principles (Nennstiel, 2021). Apart from ethical and evidence-based criteria, the question remains how new diseases are nominated for inclusion in the NBS panel, i.e. how certain diseases get on the political agenda.

According to Jansen et al. (2017) there are two different approaches when it comes to expanding NBS panel. First, the structured horizon scanning approach generally involves an independent body that conducts horizon scanning to identify a range of relevant conditions that should be evaluated for inclusion in the NBS program. This approach supports the expansion of NBS through an evidence-based process (Jansen et al., 2017). Second, the *ad hoc* approach is characterised by the incorporation of new conditions for assessment, driven by advancements in treatment possibilities, expanded disease definitions, enhanced comprehension of pathophysiology, and dedicated advocacy efforts. (Jansen et al., 2017). In this thesis, one objective is going to be to determine which of these two approaches was employed for decision-making purposes to determine the inclusion for new diseases in the NBS panel of Switzerland and Ireland.

2.2 Power dynamics in health policy

When it comes to health policy, where stakeholders, institutions, and diverse interests interweave, understanding power dynamics becomes of great importance in order to shape equitable and effective healthcare systems. Although power is a key factor in the implementation of health policy processes, explicit research on health policy implementation is rare in the existing literature (Topp & Schaaf et al., 2021). Power, in the context of policy making, is commonly perceived as a relational concept that involves one or more actors exerting control or influence over others (Buse et al., 2012). For the present thesis, this understanding of power will be utilized.

In Order to understand the relationship of power and policy, this section will provide overview of power theories concerning health policy making. To begin, this thesis adopts the power triangle framework developed by Walt and Gilson, which highlights the dimensions essential for comprehending power dynamics in policy-making. Building upon this foundation, the subsequent section delves into theories that examine the distribution of power within the policy-making process.

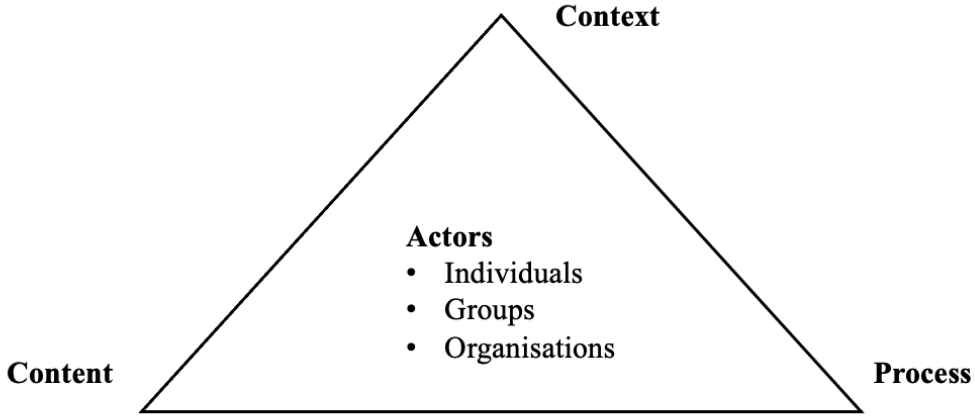

Figure 1 Policy analysis triangle (Walt & Gilson 1994, p. 354, own representation)

Walt and Gilson introduced a conceptual framework called the policy triangle that provides understanding of health policy formulation and implementation. According to Walt and Gilson, a significant oversight in health policy lies in its disproportionate emphasis on the specifics of reform content while overlooking the crucial role of the other components of the triangle, namely context, actors, and process (Walt & Gilson, 1994).

Context refers to factors that shape policy development, such as historical, cultural, and social factors, as well as the existing healthcare system. As for actors in health policies, it is important to acknowledge the presence of non-state actors such as advocacy groups or physicians' networks. These actors, although not driven by the pursuit of formal political power for themselves, strive to exert their influence over those who hold such power. They frequently integrate themselves into networks aimed at deliberating and making decisions on policies. In certain cases, governments may even fund such groups themselves (Buse et al., 2012.). An example may be the establishment of expert commissions through the government. The process component of the policy triangle involves the people, organisations, and steps involved in creating and implementing policies. It includes policymakers, interest groups, and other stakeholders who shape the policy. This component recognizes the dynamic nature of policy development, considering negotiations, power dynamics, and interactions among different actors throughout the policy cycle (Walt & Gilson, 1994).

This thesis follows the policy triangle framework, incorporating all its components and acknowledging their significance. In the theoretical section, the context is explored through an examination of the healthcare systems in Ireland and Switzerland and through shedding light on the key actors involved in NBS. To explore the process component, qualitative research is conducted through interviews, encompassing both state and non-state actors. The aim is to gain valuable insights into the policy development process, including the dynamics of power, specifically regarding NBS, when it comes to the inclusion of further diseases.

In order to analyse the dynamics of power within the context of health policy, it is important to establish a definition of what power entails. According to Steven Lukes (2005) there are three dimensions of political power.

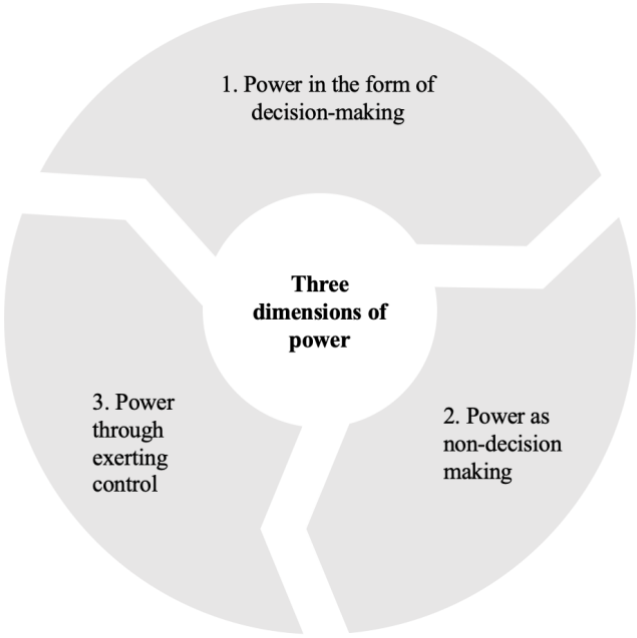

Figure 2 Three dimensions of power (own representation)

Lukes three dimensions of power expand upon two existing concepts of power and introduce a third dimension (Buse, 2012). The first dimension of power, power in form of decision-making, revolves around direct control and influence, focusing on visible actions such as decision-making, policy formulation, and the exercise of authority (Buse, 2012). This initial theory of power, often attributed to Robert Dahl, posits that "A has power over B to the extent that he can get B to do something that B would not otherwise do (Dahl, 1957, p. 203)". Dahl's definition captures the core essence of power dynamics. It underscored the fundamental notion that one party, referred to as A, possesses the capability to wield influence and force another party, symbolized as B into behaving in ways, they would not have chosen autonomously. This definition sheds light on the imbalances within power. Dahl argued that different groups in society, even groups considered as weak, could influence the political system and the policy making process. Only a few people had direct influence over key decisions, most had indirect influence (Buse, 2012). Critics of Dahl's analysis contend that his approach fails to account for crucial aspects of power. These critics argue that Dahl overlooks the potential for dominant groups to shape the political agenda according to their preferences, thereby limiting the range of issues up for discussion (Buse, 2012). Power can thus be exercised not only through explicit decision-making, but also through the ability to shape the boundaries of what is worth considering in political discourse.

Baratz and Bachrach's work, *The Faces of Power*, emerged as a response to Dahl's perspective on power. The authors argued that relying on a single dimension for power, which focuses on the power to choose from a pre-existing list of options, falls short in explaining aspects such as the determinants of decision-making and the construction of the available choices (Bachrach, Baratz, 1962). The second dimension of power, power as non-decision making, expands upon the traditional understanding of power beyond decision-making and highlights the role of agenda-setting. It concerns more subtle forms of power, involving the ability to shape political agendas, control discourse, and determine what is considered acceptable or legitimate. Manipulation, information control, and influence over decision-making processes are key mechanisms in this dimension (Buse, 2012). In other words, power is exercised by determining which issues are bought to the forefront of public attention and which remain neglected or excluded from the agendas.

The third dimension, power through exerting control, introduced by Steven Lukes, explores power dynamics at a deeper level. It goes beyond the observable processes of decision-making and agenda-setting, delving into the realm of people's beliefs, values, and perceptions. By shaping the political consciousness and social reality of individuals or groups, power holders seek to align thoughts, desires, and identities with their own interests (Lukes, 2005). Lukes suggests that the dimension of power involving control is particularly deceptive and harmful. In this form, power operates by influencing people's perceptions, thoughts, and preferences, leading them to accept their place within the existing social order. This occurs because individuals either cannot envision or are unable to imagine any alternative to the current system, perceive it as natural and unchangeable, or believe it to be inherently beneficial and ordained by a higher power. By shaping people's mindset in this way, this insidious form of power discourages objections and reinforces the status quo (Buse, 2012).

This thesis builds upon the established framework of the three dimensions of power to undertake an exploration of power dynamics within the context of NBS in Switzerland and Ireland. The primary objective is to identify the most relevant theory of power for each respective country. Through the utilization of this analytical framework, the research aims to gain understanding of the interplay of power dynamics in health policy,

particularly in relation to the policy-making process for inclusion of a new diseases to NBS panels in Switzerland and Ireland.

To gain a understanding of power dynamics within the context of NBS, it is important to consider the divergent healthcare systems in Switzerland and Ireland. Switzerland adopts a statutory health system, while Ireland operates under a national health system. Recognizing these contrasting frameworks is crucial for examining the potential variations in power dynamics within these systems.

When comparing national health care systems to statutory health insurance systems, the distribution of power is a key differentiating factor. In national healthcare systems, the government assumes the responsibility of providing healthcare services to the entire population, resulting in centralized control (Egger, 2012). This concentration of power enables governments to effectively coordinate resources, regulate service delivery, and negotiate with healthcare providers (Egger, 2012). However, statutory health insurance systems distribute power among various stakeholders, including government bodies, private insurers, and healthcare providers (Thomson et al., 2013). This approach creates a more complex and decentralized healthcare landscape.

In statutory health insurance systems, the concept of redistribution extends beyond the division between the healthy and the sick. It covers a broader spectrum, encompassing redistribution among individuals of varying ages, marital status, and income levels. This means that the financial burdens and benefits are shared among the insured population, with the aim of ensuring fairness and equity (Bandelow et al., 2023). In contrast, national health systems are based on the idea that health care is a social right of citizenship and that the entire population has a right to health care in case of illness, regardless of their social and financial situation. (Wendt, 2013). Thus, while access to national health systems is defined as a right of citizenship, the right to support in health insurance systems must be acquired by paying contributions (Wendt, 2013).

2.3 Overview of newborn screening policies in Switzerland and Ireland

When examining the process of policy development for incorporating new conditions into a country's NBS panel, it is important to thoroughly consider the healthcare systems specific to each country. Therefore, this section offers an overview of the NBS policies

in Switzerland and Ireland, taking into account the distinctive characteristics of their respective healthcare systems. It will also explore the comparability between the two countries in terms of the indications currently being screened, the management of the screening process, and the organization of funding.

2.3.1 Healthcare in Switzerland

In Switzerland, the BAG is the national authority responsible for public health matters, healthcare policy, and coordination of health-related issues at the federal level. The compulsory health insurance system is a fundamental aspect, ensuring that every resident possesses coverage. The Swiss system embraces a combination of public and private healthcare providers, employing a decentralized approach where cantons hold considerable autonomy in managing and organizing healthcare services (Brock, 2022). Private insurance companies offer a variety of coverage options, granting individuals the freedom to select their provider. This competitive environment fosters diverse choices for insured individuals. The Swiss healthcare system has a unique organizational structure characterized by a combination of federalism, liberalism, corporatism, and the influence of direct democracy (Brock, 2022). At the federal level, the legal framework for mandatory health insurance, accreditation of healthcare professionals, regulations for medical devices, and epidemiological measures are established. However, the main responsibility for healthcare lies with the cantons. The majority of hospitals is owned by the respective canton, and approximately half of the funding for the inpatient sector is provided by the cantons. Additionally, the cantons are responsible for organizing long-term care and emergency systems (Konferenz der kantonalen Gesundheitsdirektorinnen und -direktoren, 2023).

Financing in the Swiss healthcare system are highly decentralized. The main financiers are mandatory health insurance providers, which operate as private nonprofit companies in a competitive market (Brock, 2022). A distinctive feature of Swiss democracy is the high level of direct citizen participation in political decision-making processes. Almost all healthcare-related legislative proposals can be influenced through nationwide referendums. Many competencies in healthcare provision are vested in the cantons, and many significant healthcare policy issues are decided directly by the local population (Brock, 2022).

2.3.2 Newborn Screening in Switzerland

NBS has existed for the last 50 years in Switzerland. A few drops of blood are obtained from the heel of every newborn and placed on a filter paper. This blood sampling procedure, which is carried out on the fourth day after birth, takes into account the need for certain screening biomarkers to accumulate and achieve high sensitivity, while also considering the potential lack of specificity in some biomarkers shortly after birth (Sluka, 2022). In 2005 the decision was made to centralize NBS in Switzerland. Since then there is only one NBS lab in Switzerland, the Children's Hospital of Zurich, *Kinderspital Zürich*. The Children's Hospital of Zurich carries out the NBSs not only for Switzerland but also for Lichtenstein. The hospital processes a volume of 1600-1700 filter papers on a weekly basis. (Sluka, 2022). As of now, ten metabolic and hormonal diseases are tested in the Swiss NBS:

1. Phenylketonuria (PKU)
2. Hypothyroidism
3. Medium-Chain Acyl-CoA Dehydrogenase (MCADD)
4. Congenital Adrenal Hyperplasia (CAH), Galactosaemia
5. Biotinidase Deficiency
6. Cystic Fibrosis (CF)
7. Glutaric acidemia type 1 (GA1)
8. Maple syrup urine disease (MSUD)
9. Severe Combined Immunodeficiency (SCID)
10. Severe T-cell lymphopenia

SCID was the last disease to be included in the Swiss NBS in 2019 (Neugeborenenscreening Schweiz, 2023). Experts from the *Kinderspital of Zürich* submitted the application for the NBS of SCID to the BAG in December 2015, and after various revisions it was approved three years later (Steinmann & Baumgartner, 2022).

The BAG is the responsible authority for the expansion of the NBS panel. Two commissions are consulted and advise the BAG in its decision-making. The *Eidgenössische Kommission für genetische Untersuchungen beim Menschen* (GUMEK) functions as a non-parliamentary commission that has been assigned the responsibility of monitoring the progress in scientific and practical aspects of genetic investigations. Its

role includes giving recommendations and ensuring timely identification of problems and legislative gaps (Bundesamt für Gesundheit, 2022). The *Nationale Ethik Kommission* (NEK) is an independent panel of experts that is consulted by the BAG on the expansion of the NBS panel (Bundesamt für Gesundheit, 2023). Both commissions consist of experts in the corresponding area and are elected by the federal council.

In Switzerland NBS is reimbursed through the health insurance. The legal foundation governing the reimbursement of NBS is specified in Swiss health insurance act, the *Bundesgesetz über die Krankenversicherung* (KVG) article 26, which concerns mandatory services within the scope of generally recommended preventive measures, similar to routine childhood vaccinations. The corresponding regulations can be found in the Swiss Health Insurance Benefits Ordinance, known as *Verordnung über die Krankenversicherung* (KVV) article 33, subparagraph d, as well as article 53. Additionally, the reimbursement is also governed by the Swiss health insurance benefits Ordinance, the Krankenpflege-Leistungsverordnung (KVL) article 12, subparagraph e (Steinmann & Baumgartner, 2022).

The inclusion of SMA in Switzerland's screening panel was requested in February 2023. The application, submitted by the Paediatric Neuromuscular Reference Centres (Myosuisse), is currently under review by the BAG (Muskelgesellschaft, 2023a). Myosuisse is a network of all professionals and organisations working for people with a neuromuscular disease in Switzerland and is part of the Swiss Muscle Society, the *Schweizerische Muskelgesellschaft* (Schweizerische Muskelgesellschaft, 2023b). The Swiss Muscle Society is a nonprofit organisation that represents the interests and concerns of people with a muscle disease in Switzerland (Schweizerische Muskelgesellschaft, 2023c).

2.3.3 Healthcare in Ireland

The Irish health care sector is structured as a two-tier, dual system with the HSE, financed by public taxes, constituting the first structural level, the second one being voluntary private health insurances. However, the state sector only covers a few basic services: Ireland is the only Western European country where primary health care is not part of the state health service(Szabó, 2020). Citizens have to pay for it out of their own pockets, unless they are exempt. General practitioners (GPs) are not obliged to accept patients

(Szabó, 2020). While public health care is the primary coverage option, voluntary private health insurance is also widely used, with many people having both public and private coverage. Private health insurances offer services that are complementary to the public health care system, but also partly overlap with it (Szabó, 2020). Public healthcare services are provided through a network of hospitals and primary care centres. Accessing specialized care usually requires referrals from primary care physicians, highlighting the crucial role of GPs as the entry point to specialized services (Irish Life Health, 2023).

The Irish government has recently introduced a reform initiative called Sláintecare. The objective is to enhance healthcare accessibility for all, improve coordination between primary, community, and hospital care, and decentralize decision-making (Sicari & Sutherland, 2023) . However, the healthcare system's intricacy and lack of transparency have made these reforms complex. One key concern revolves around the coexistence of public and private healthcare sectors, with private patients enjoying easier access to services, raising apprehensions about a two-tier system (Sicari & Sutherland, 2023). To address these issues, the government plans to establish regional health areas for more localized decision-making (Sicari & Sutherland, 2023).

2.3.4 Newborn Screening in Ireland

NBS is commonly referred to as the “heel prick” test in Ireland. The first NBS for PKU in Ireland was conducted in 1966. The Irish NBS program was one of the first in the world, implemented just four years after the first NBS in the US. The HSE oversees the National Newborn Bloodspot Screening Laboratory (NNBSL) at Children’s Health Ireland (CHI), which is the only NBS laboratory in Ireland and manages the coordination of screening processes (Health Service Executive, 2022). Ireland currently has nine diseases included in their NBS panel.

1. Phenylketonuria (PKU)
2. Homocystinuria (HCU)
3. Classical Galactosaemia (CGal)
4. Maple Syrup Urine Disease (MSUD)
5. Congenital Hypothyroidism (CHT)
6. Cystic Fibrosis (CF)
7. Glutaric Aciduria Type 1 (GA1)

8. Medium Chain Acyl CoA Dehydrogenase deficiency (MCADD)
9. Adenosine Deaminase-Deficient Severe Combined Immunodeficiency (ADA-SCID)

ADA-SCID was the most recent addition to the screening panel in 2022 (Health Service Executive, 2022). The diseases included in Ireland's NBS have been chosen based on their higher incidence among the Irish population and their alignment with international criteria for NBS (Health Service Executive, 2022). These criteria include treatability of the conditions, availability of a widely applicable test, reliability of the test with minimal false positive and false negative results, a significant occurrence of the conditions in the community justifying screening, and cost-effectiveness of the screening process (Health Service Executive, 2022).

NBS is an essential component of the public health service extended to newborn infants. As a result, there are no expenses incurred by the parents for the screening service (Health Service Executive, 2023). The screening usually takes place between the third and fifth day of the newborn's life. It involves a blood test taken from the baby's heel (Health Service Executive, 2023). The screening process requires the collaboration of multiple entities responsible for collecting, transporting, analyzing samples, and documenting results, as well as referring and managing infants identified with any of the conditions. The HSE and the *Assistant National Director of Health and Wellbeing – Public Health and Child Health*, who leads the NNBSL Governance Group, hold the primary responsibility for ensuring that all infants are offered screening according to established protocols and procedures. The director of the NNBSL oversees the daily coordination and management of the program (Health Service Executive, 2022).

In 2019, the NSAC was created as an autonomous advisory committee with the purpose of participating in the development and evaluation of population-based screening programs in Ireland (Department of Health, 2023). The NSAC's primary responsibility is to offer guidance and recommendations to the Minister for Health and the Department of Health regarding new screening initiatives and proposed modifications to existing screening programs, including NBS (Health Information and Quality Authority, 2021).

In December 2022, the NSAC released their meeting report, suggesting the expansion of NBS for five diseases, with SMA being recommended as the initial priority for inclusion in the screening panel. The report highlighted that a HTA should be conducted by the HIQA specifically for SMA. The NSAC stated their intention to formally request the HTA for SMA to assess its feasibility for integration into the NBS program. (National Screening Advisory Committee, 2023).

2.3.5 Comparing Newborn Screening Programs of Switzerland and Ireland

The comparison of NBS policies in Switzerland and Ireland reveal both similarities and differences in various aspects. First, Switzerland operates a decentralized healthcare system, with a combination of public and private providers and autonomy given to the cantons in managing healthcare services. In contrast, Ireland has a dual-tier healthcare system, with public and private sectors co-existing. Public healthcare is the primary coverage option, but private insurance is widely used. The Irish government aims to enhance accessibility and decentralize decision making of healthcare through the reform of Sláintecare. The funding for healthcare in Switzerland primarily comes from mandatory health insurance providers, in Ireland the Public health service is primarily funded through taxes.

In Switzerland, reimbursement for the NBS is governed by the Swiss health insurance act. NBS is reimbursed through the health insurance. In Ireland, the reimbursement for NBS is covered as part of the public health service. In terms of similarities of NBS, both countries employ heel-prick blood sample tests, taken shortly after birth. Both countries have one laboratory responsible for analysing the samples. Additionally both countries are planning to expand their NBS to include SMA. The process of expanding the screening panel differs between Switzerland and Ireland, primarily due to variations in the entities involved in the application process and recommendation. In Switzerland, the application for including SMA in the screening panel, was submitted by *Myosuisse*, which is part of the non-profit organisation *Schweizerische Muskelgesellschaft*, to the BAG. In Ireland the NSAC, an independent advisory committee, recommend a HTA, conducted by the HIQA, to assess feasibility of the inclusion of SMA to the screening panel.

3 Research method

3.1 Research design

This study adopts a qualitative research approach to examine power dynamics within the policy-making processes for the inclusion new diseases in NBS panels in Switzerland and Ireland. Specifically, it focuses on the inclusion of SMA in the screening panel. This thesis seeks to enhance our understanding of power dynamics in the context of policy-making for NBS. Through the analysis of interview data and the comparative examination of policy processes, valuable insights can be gained, contributing to the existing body of knowledge on this topic.

3.2 Data collection method

In addition to a literature and document review, the primary research method involves conducting interviews with key actors involved in the policy-making process of NBS in Switzerland and Ireland, including both state and non-state actors. Through these interviews, the study aims to gain insights into the different forms of power and its expression in policy-making. Comparing the different policy-making processes of these two countries provides a valuable opportunity to examine similarities and differences, shedding light on the various factors that influence policy-making.

3.3 Data analysis method

In order to answer the research question *"How do power dynamics shape the decision-making processes of including new disorders in newborn screening panels in Switzerland and Ireland and who are relevant actors in this process?"* a thematic analysis will be utilized to examine the interview transcripts, facilitating the identification of recurring themes and patterns pertaining to power dynamics. This allows conducting a detailed and practical analysis through interviews with experts. In this way, opinions and motives of individuals involved in NBS policy-making can be taken into account. Based on the important topics in the literature review, questions were derived that were discussed in semi-structured interviews with state and non-state actors from the NBS policy area. There were two interview partners from each country.

Interview partner 1, based in Switzerland, holds a prominent position in the clinical area of NBS. Additionally, they contribute to an advisory committee associated with the

authorities. This dual role classifies the interview partner as both, a state- and a non-state-actor, providing valuable perspective from different spheres.

Interview partner 2[2], based in Switzerland, serves within a government agency in charge of the execution process. Their involvement allows insight into the execution process from an authoritative point of view.

Interview partner 3, based in Ireland, has a leadership position within a patient organisation, while also practicing in an advisory committee. Their expertise in both fields provides a singular viewpoint by combining the knowledge gained from working with governmental organisations and patient advocacy.

Interview partner 4, based in Ireland, holds a leadership position within a patient organisation. Their experience within this organization contribute to a understanding of NBS from a patient-oriented viewpoint.

The Interviews were transcribed and coded. The codes were made based on the important themes found through the theory. First, the framework proposed by Walt and Gilson, known as the policy triangle, was utilized to analyze the different aspects of policy-making. To gain a deeper understanding of the actors involved, various codes such as 'agenda', 'power', 'challenges', and 'collaboration' were assigned to the interview quotes. These codes allowed for a more exploration of the perspectives and insights shared by the interview partners. To get deeper understanding of the process part, codes like ‘application’, ‘requirements’, ‘indication’, and ‘improvement’ were used. Similarly, the process part of the policy-making was examined using codes like 'application', 'requirements', 'indication', and 'improvement'[3]. This coding approach aimed to uncover the intricacies of policy implementation and shed light on key factors influencing the process. To analyze power dynamics, the three dimensions of power were used as codes. Similar or comparable codes were then grouped together. From these units, the particular characteristics of each category and the possible relationships between the categories can

[2] The conversation with interview partner 2 was conducted in German. Quotes were translated to English. View appendix for original quotes and their translation.
[3] Ad hoc vs. horizontal screening approach by Jansen et al. 2014.

be better captured (Saldaña, 2014). A qualitative content analysis according to Mayring was used to analyse the data (Mayring, 2019).

4 Results

4.1 Results Switzerland

4.1.1 Relevant actors in Swiss NBS

The field of actors in Swiss NBS consists of state and non-state actors. On one hand, non-state actors include patients and their families, patient organisations, physicians, the pharmaceutical industry, and the laboratory responsible for NBS in Switzerland. Patients, their families, patient organisations and physicians share a common interest in the screening and potential treatment of specific diseases. Their primary objective is to achieve improved health outcomes for their family members or patients. The pharmaceutical industry has a financial incentive to advocate for the inclusion of specific diseases in the NBS panel. When a pharmaceutical company offers a treatment for a particular disease, their interest in having that disease included in the panel increases, as it enables them to market and sell their medication.

> *"It usually starts with the interested patients or physicians. So you have a given disease. Let's take SMA and there is now a new disease modifying treatments. So there is a rising interest of the industry that offers these treatments. But at the same time, of course, also of the patients that profit from these. And their parents and their patients-organization that profit from these treatments that future newborns will be screened and treated earlier." (Interview partner 1, 00:05:12)*

On the other hand, there are state actors of the BAG, including its commissions, the GUMEK, the NEK, and the *Eidgenössische Kommission für Analysen, Mittel und Gegenstände* (EAMGK). The EAMGK is responsible for evaluating new or existing positions in the *Analysenliste* (AL) upon request submitted to the EAMGK. The AL encompasses the specific analyses provided by medical laboratories that are covered by mandatory health insurance.

The involvement of the NEK in expanding screening panels differs depending on the target population. Generally, when it comes to screenings for adults, the NEK does not have a role in the process. However, the situation is different for NBS. As newborns lack the capacity for discernment to decide whether they want to be screened or not, the NEK is involved in expanding the NBS panel. The BAG, the GUMEK, and the NEK are collectively invested in ensuring high-quality healthcare while also being mindful of the costs for Switzerland. The strategy for the years 2020 to 2030, *Gesundheit 2030* describes the political agenda of the BAG.

> *"The aim is to have good healthcare in place, and the costs are always a matter of concern, ensuring they remain moderate and affordable, so that it can be paid for. That is certainly also an important aspect." (Interview partner 2, 00:01:30)*

All stakeholder groups involved in the field of NBS in Switzerland are generally supportive of the expansion of the NBS panel. This collaborative approach is driven by the shared goal of improving early detection and intervention for a wider range of genetic disorders. However, there are also some critical voices within the stakeholder community who raise concerns specifically regarding the costs of the expansion of the NBS panel.

> *"There is a general concordance that it [inclusion of SMA to the NBS panel] makes sense. […] Among some of the actors, there may be doubts about what happens to the costs or how Switzerland is going to finance it. But that's not against newborn screening in general." (Interview partner 1, 00:15:20)*

The BAG has to take into account the importance of considering costs in relation to the expansion of the NBS panel. The BAG places great emphasis on securing long-term funding for new screenings.

> *"It was important to us that the financing is guaranteed for a longer period of time, because this affects the whole of Switzerland, which includes between 80 and 90,000 newborns every year." (Interview partner 2, 00:05:30)*

Since NBS is funded through mandatory health insurance, the The Swiss Health and Accident Insurance Directorate, known as the *Direktion für Kranken- und*

Unfallversicherung holds influence in the process of expanding the NBS program. If a new disease is to be included in the NBS panel, an application must be submitted simultaneously to both the BAG and the EAMGK.

> *"Ideally, the applications are then submitted in parallel, precisely also for the inclusion of this preventive measure." (Interview partner 2, 00:07:02)*

The involvement of the EAMGK ensures a smooth process of financing the costs that arise from screening, such as costs for the screening kit or staff.

> *"Maybe it is a bit more political there [EAMGK], because we also have the cost-cutting measures that have to be implemented now. […] as soon as reimbursement is discussed, it becomes a bit more political. (Interview partner 2, 00:25:50)*

Relevant stakeholder groups in Switzerland show strong support for NBS. There is general consensus among stakeholders about the significance of disease testing through NBS. However, on the government side, there is a need to balance these interests with the consideration of cost.

4.1.2 Swiss policy-making process of NBS

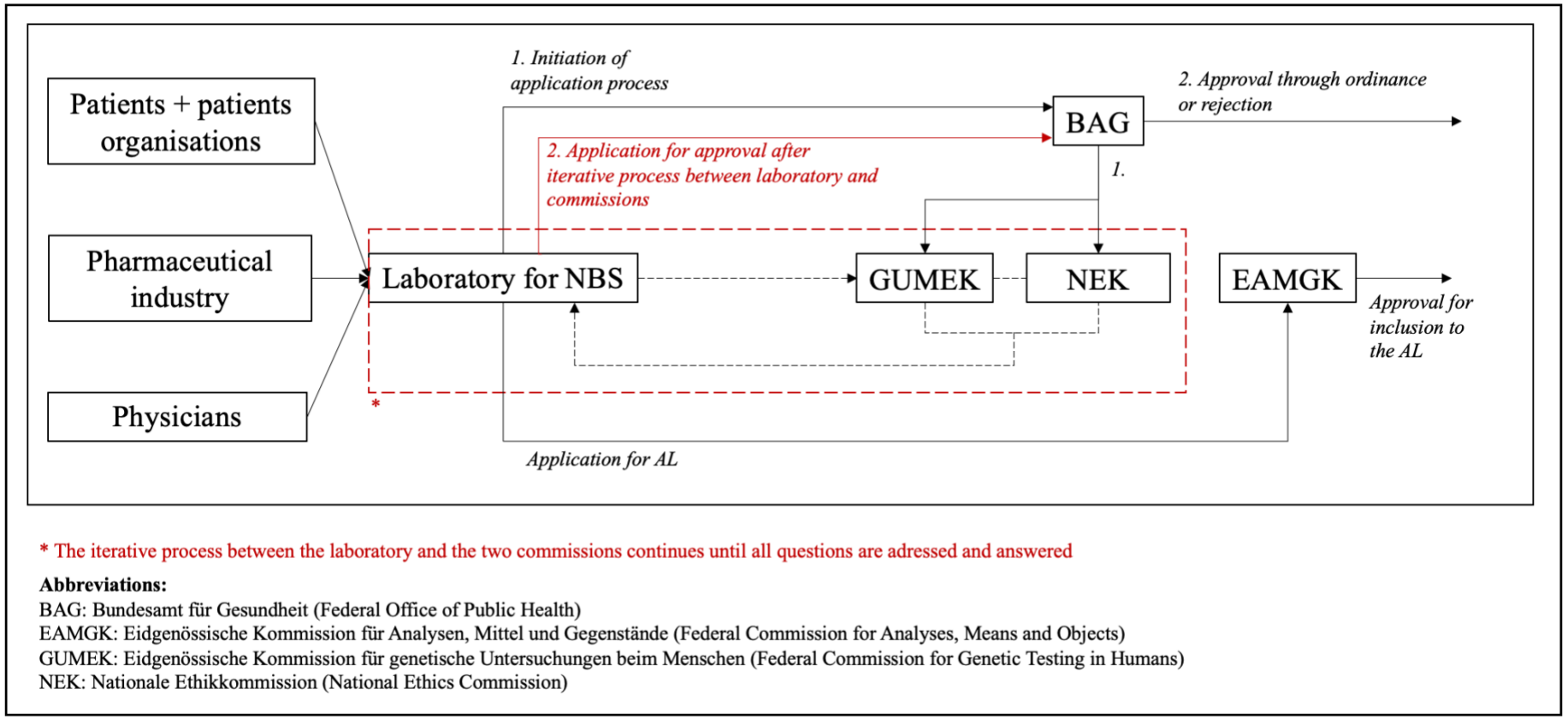

Figure 3 Relevant actors and policy-making process of NBS in Switzerland (own representation)

To secure approval from the BAG for adding a new disease to the NBS panel, a specific application process must be followed. The regulatory framework governing genetic screening in humans is established by the *Federal act and Ordinance on Genetic Testing* (GUMG SR 810.12; GUMV SR 810.122.1). Article 30 of the GUMG specifies the criteria that the application concept must meet and outlines the procedures for initiation, consultation, and approval.

> *"Well, that is simply a well-defined, clear procedure. All the legal provisions are defined in the law and the ordinance, […] the whole procedure, which documents have to be submitted etc., it has to be checked by us, by the experts. And then, if the requirements are met, it can be approved or it has to be approved. […] It is a decision based on professional criteria. And especially the newborn screening, so if there has ever been a success story of a screening, then it is certainly the newborn screening. Because such a life-saving intervention can be made so early." (Interview partner 2, 00:02:08)*

Typically, the application process for adding a new disease to the NBS panel starts with rising interest of various actors, such as the pharmaceutical industry, patients, patients' organisations, or physicians specializing in a particular disease, due to a new modifying treatment. When such interest is present, these actors initiate the application process by approaching the NBS laboratory.

> *"The next step they do is they come to us, to the screening lab and ask, can you do that? […] and I usually say, yes, of course. Then you have to apply to the BAG." (Interview partner 1, 00:07:30)*

When the laboratory is approached by patient organisations, physicians or a pharmaceutical company, they first discuss the possibilities of conducting a screening for the disease they want to put on the NBS panel.

> *"The most important factors are A) you have to have a test that is sensitive and specific of that. And B) you need the disease modifying treatment that is available and will be paid by the insurance. Because, you know, I would never agree to put something into screening if afterwards the e-file or the health insurance won't pay*

for the treatment. Because that's terrible for the parents." (Interview partner 1, 00:24:30)

In the case of SMA, the laboratory was approached by Myosuisse and neuro paediatrician who advocated for SMA to be added to the NBS panel. As a next step it is necessary to submit an application to the BAG. The application should highlight the possibility of early treatment or prevention of the disease being investigated. It should also demonstrate that the screening method provides reliable results, ensuring appropriate genetic counselling and a suitable duration for the screening program. When an application reaches the BAG, it undergoes an evaluation to ensure its completeness and comprehensibility. If the application meets these criteria, the BAG proceeds to submit it to the GUMEK and the NEK for review. Once both commissions have assessed the application, it is returned to the applicants and the laboratory for additional statements and inputs. The right to be heard before a decision is made, is granted. This process may the several rounds between the commissions and the applicants.

"If the application is complete and what they [applicants] want to do has been explained in a comprehensible way […] then the application is submitted to the two commissions and these two actors are also involved. The application is then sent back to the applicants for their comments. The right to be heard before any decision is made is granted. They can comment again on points that the commissions have raised." (Interview partner 2, 00:04:30)

In addition, the laboratory sends a second application to the EAMGK in order for the screening of the new disease to be added to the AL and be reimbursed through the mandatory health insurance.

"[…] you also have to submit a separate submission to get the additional cost that you create just to make the test. So, kit costs, more personnel, whatever that is needed in the central screening laboratory. There you have to make a separate submission to the Analysenliste, also part of the BAG, but another section and another process to raise the price. And they again, look, is everything fine with what

you are testing? Do you do the proper quality control and so on." (Interview partner 1, 00:09:00)

Once all questions regarding the application have been answered, the BAG and the EAMGK approve the two applications and an official order is issued by the BAG, confirming the inclusion of the disease in the NBS. It is worth noting that due to the committees' meeting schedule, the evaluation process may extend over a considerable duration, typically occurring five to six times per year. Figure 3 shows the policy making process for adding an additional disease to the NBS panel in Switzerland and all relevant actors included in the process.

4.1.3 Power dynamics in Swiss NBS policy-making of NBS

In Switzerland, the addition of a new disease to the NBS panel follows a legally prescribed process. The law and constitution outline the specific requirements that must be met to obtain an official order for inclusion. The initiation of the application process involves non-state actors exclusively, until the submission to the BAG. If there is a disease modifying treatment, the pharmaceutical industry has an interest in adding a new disease to the panel. This interest can serve as a driving force behind initiating the application process.

"I think the case of SMA shows that pharma can be dominant. In the way that if they have a treatment, of course they are interested in bringing that along. And I was a little bit surprised that they didn't push to get it done faster." (Interview partner 1, 00:31:30)

The laboratory holds a significant role in the process as well, as it is responsible for conducting all NBSs in Switzerland. After the BAG issues the official order to add a new disease to the panel, the laboratory must be prepared to implement the necessary testing. Moreover, the laboratory holds an important role in the process and exercises decision-making authority as a gatekeeper for including diseases in the NBS panel. They have the power to accept or reject interest groups' requests to start the application process. It is noteworthy that, up to this point, the laboratory has not turned down any proposals from interest groups. This demonstrates their willingness to embrace and accommodate new additions to the panel, ensuring a evolving screening program.

"Certainly the newborn screening lab is a dominant actor in the way that they have to be ready to implement the test. We have not turned down anything you know saying don't do it. But there is some dominance there." (Interview partner 1, 00:30:40)

Once the application is submitted to the BAG, the involvement of state actors begins. The decision and power on whether a disease is included in the NBS panel lies with the BAG.

"Of course they [BAG] can turn it up or down or up. But, you know, it seems like they are in line with what we from the screening lab and the involved physician community think on how screening is done in Switzerland." (Interview partner 1, 00:32:05)

It is crucial to note that these committees operate autonomously and are separate from the parliamentary structure, ensuring their impartiality and expertise in the evaluation process. The BAG committees (GUMEK and NEK) are actively involved in the decision-making process as they are consulted by the BAG and tasked with reviewing the application put forth by the laboratory. Their expertise and input hold significant influence on the final decision regarding the inclusion of a disease in the NBS panel. The EAMGK reviews the application for the AL, but is not directly consulted by the BAG.

"We have the GUMEK, which is a really good expert commission that advises us on these issues and gives its opinion. Of course, it can be critical, that's quite clear. It can be very sympathetic and it can also be very critical. I guess it is the same with the EAMGK. There it is perhaps even a bit more political, because we also have the cost-cutting measures that now have to be implemented." (Interview partner 2, 00:26:00)

As previously mentioned, there have been no instances of actor groups actively opposing the addition of new diseases to the NBS panel. However, it is worth noting that within the committees, there are some voices that express awareness and consideration for the costs associated with adding new diseases to the NBS panel.

"Switzerland is rather conservative in implementing new screening tests. You see that by the number of tests that we have and compare this to other countries. So we are still very conservative in a way. And trying to only introduce diseases that fulfill the old, Younger and Wilson criteria of 1968." (Interview partner 1, 00:26:00)

"And now the BAG, of course we talk to them and they share, and also the GUMEK and the NEK, they share the conservative line that Switzerland is going." Interview partner 1 00:33:20)

Significant decision-making power lies with the state actors involved in the NBS process in Switzerland. Once an application is submitted to the BAG, the involvement of non-state actors in the decision-making process is limited. The authority to evaluate and determine the inclusion of diseases in the NBS panel rests primarily with the state actors, ensuring a centralized and regulated approach to decision-making in NBS.

"Of course, they [non-state actors] were all consulted in the processing of the articles of law and ordinances. That is clear. They were able to express themselves there. There is also a consultation report, both on the ordinance and the law. There you can see what the various parties or professional societies or other associations have said about the individual articles." (Interview partner 2, 00:27:50)

As of now the health insurance covers the costs of tests and treatments for diseases identified through NBS. However, the capacity to add additional diseases to the NBS panel requires careful consideration.

"Now it is paid for, but how many of such additional expensive treatments can Switzerland afford as a country, or is Switzerland willing to afford? And that's a discussion that should be held in a much bigger public and not among the specialists. That's not us to decide, nor the specialists, nor the patients. It's the population that needs to be aware of these of these potential rising costs." (Interview partner 1, 00:25:54)

The power dynamics within Swiss NBS policy-making involve multiple actors. The application process is initiated by non-state actors, who express interest in expanding the

NBS panel. The laboratory holds a role as the gatekeeper, deciding whether to accept or reject requests for inclusion. The final decision rests with the BAG while independent committees like GUMEK and NEK provide valuable expertise and influence. The consideration of costs and the capacity to incorporate new diseases into the panel needs ongoing evaluation. The involvement of state actors and the compliance with the legal frameworks ensure a centralized and transparent approach to decision-making in NBS policy.

4.2 Results Ireland

4.2.1 Relevant actors in Irish NBS

Similar to Switzerland, the non-state actors involved in Ireland's NBS encompass a range of participants, such as patients, patient organisations, physicians, and the pharmaceutical industry. In Ireland, physicians are actively urging for the inclusion of various diseases, including SMA, into the NBS. Furthermore, patients and their families are advocating for the inclusion of specific diseases in the NBS panel, the approval of treatments, and improved accessibility to these treatments. In certain instances, patient organisations may establish collaborations with pharmaceutical companies. This partnership serves as a beneficial arrangement for patient organisations, which often face resource limitations. Collaborating with pharmaceutical companies provides them with much-needed support. Simultaneously, pharmaceutical industries perceive patient organisations as valuable allies, as they cannot directly communicate with patients. Through patient organisations, they gain indirect access to patients and the opportunity to establish contact. The shared objective between these two entities is a mutual desire to obtain approval for treatments.

> *"They [pharmaceutical company] see us as an ally because they can't talk directly to patients, but they see us as a way for them to be able to kind of hear what's happening on the ground. […] You know, if we're campaigning for drugs and they're trying to sell the drugs, you know, we can all work together. And obviously there are ethical boundaries that they have to be very careful about. But we've worked very hard to be a good partner opposite them. And, you know, we work, I think, very well together. There's, you know, mutual respect and we are the smaller party of the relationship. But they're very careful and our goals are very aligned." (Interview partner 3, 00:05:00)*

The involvement of patient families also plays an important role. They actively contribute by writing letters to their local Teachta Dàla[4] (TDs) to generate awareness about NBS and specific disease. These letters carry significant influence, as Irish TDs are particularly attuned to local concerns and interests. This direct engagement by patient families adds to the collective efforts to advocate for the NBS and brings attention to the importance of addressing specific diseases within the parliament.

> *"As part of the government, on the one hand are agencies, but there also are public representatives which in Ireland are called TDs. […] Ireland's very small and it has been accused of having quite a clientelist system. So politicians will work hard to look after local interests. So if you can get a family in the middle of nowhere to write to their local TD to bring up the issue of newborn screening or access to drugs or whatever, invariably that will raise a parliamentary question." (Interview partner 3, 00:10:00)*

> *«We [patient families and patient organisations] will write to TDs frequently and whenever we kind of are in the middle of a campaign. So right now, for example, to coincide with International Neonatal Screening Day, we wrote to eight. Eight TDs who had been sympathetic.» (Interview partner 3, 00:11:54)*

State actors include the NSAC, the HIQA, the EAG, and the Minister for Health. New proposals for population-based screening or suggested changes to current programs must be processed by NSAC. The NSAC issues open call for proposals, with the first one being issued in 2021 and the second in 2022. These open calls serve as opportunities for interested actors, including the HSE, professional organisations, special interest groups, and private citizens, to submit their proposals. This approach ensures that a wide range of actors can contribute their insights and perspectives to the decision-making process related to population-based screening in Ireland. HIQA plays an important role in assessing and monitoring the quality and safety of healthcare services, as well as providing recommendations for improvements in the Irish healthcare system. The EAG refers to a multidisciplinary Expert Advisory Group convened by HIQA. This group

[4] TDs are members of the lower house of the Irish Parliament.

consists of experts from various relevant fields who provide their specialized knowledge and insights to support HIQA in conducting reviews and assessments.

4.2.2 Irish policy-making process of NBS

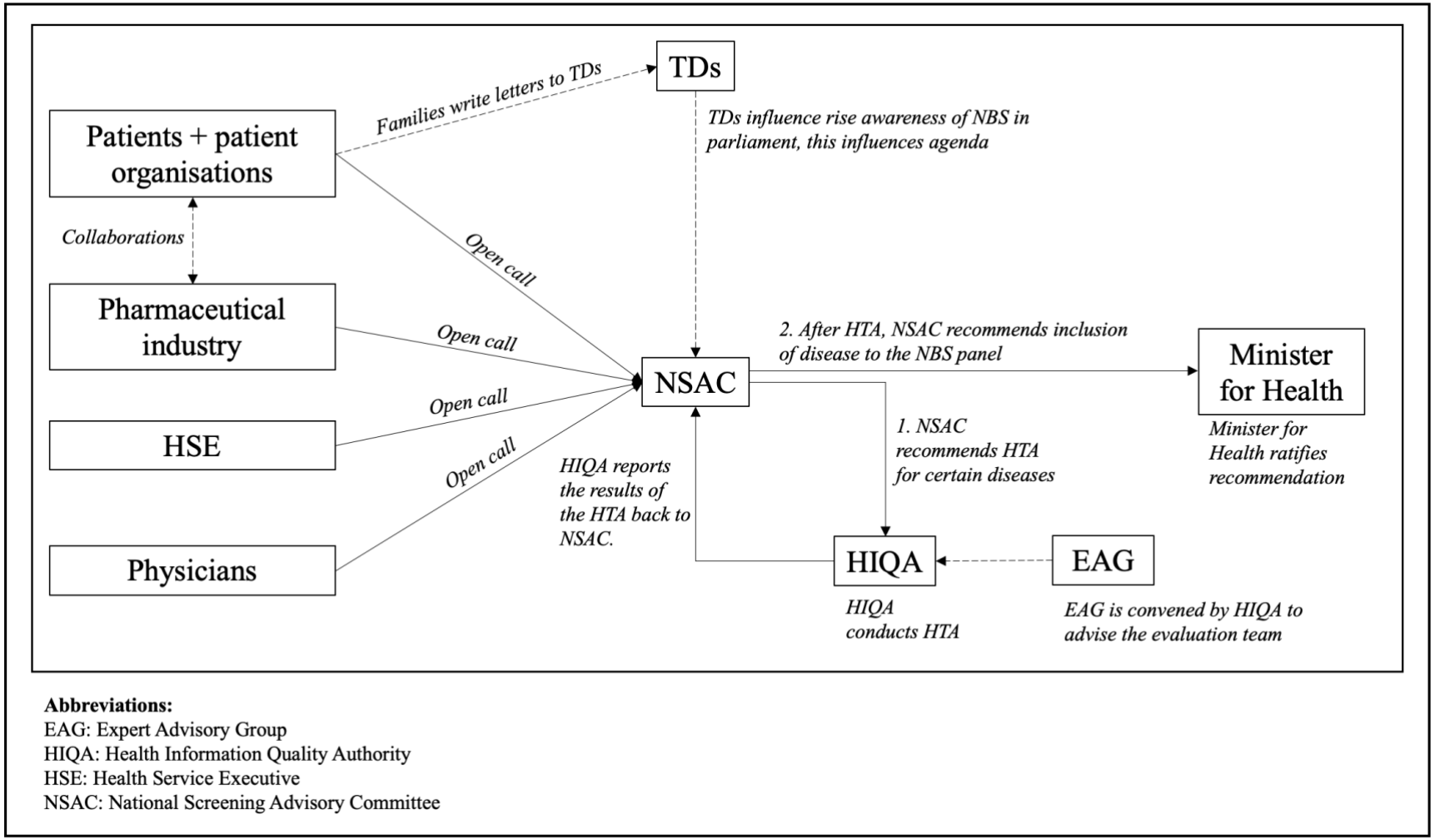

Figure 4 Relevant actors and pclicy-making process of NBS in Ireland (own representation)

In Ireland, the expansion of the NBS panel typically occurs when a new modifying treatment becomes available. This was evident in the case of SMA, where the approval of the Biogen treatment, SPINRAZA®, played an important role. As soon as this treatment received approval, patients and their families began advocating for the inclusion of SMA in the NBS panel. Their advocacy efforts were further strengthened by the formation of patient organisations dedicated to raising awareness and advocating the cause. As a result, submissions were made to propose the inclusion of SMA in the NBS during the open call conducted by NSAC. They conduct open call for submission annually, allowing interested groups to submit applications for the inclusion of new diseases in screenings across Ireland. This also includes NBS. This process ensures that different stakeholders have the opportunity to advocate for the incorporation of diseases into the screening programs. In December 2022, a total of 55 submissions were presented to the NSAC, 18 of which were about NBS. Within this subset, six submissions were focused on advocating for the inclusion of SMA in the NBS panel.

> *"They [pharmaceutical companies] would have the opportunity, as anybody in the public does, to make a submission to ask for something to be added to the to the panels." (Interview partner 4, 00:04:24)*

> *"The NSAC periodically reviews conditions to add or to discuss with respect to newborn screening or screening generally. And if they then decide that something should be considered for inclusion, for example, in the heel prick test, then they would refer that to HIQA who do the HTA. And then HIQA will come back with its decision. And then the NSAC's role is pretty much over. Then they hit a decision. It is either we recommend or not, and then the Minister for Health will ratify that. That's kind of how it works. The NSAC did an open call for consideration of conditions." (Interview partner 3, 00:18:08)*

Following the submissions, NSAC decides, which diseases they recommend for a HTA. The HTA is conducted by the Health Information and Quality Authority, known as HIQA.

> *"That [recommendation of NSAC] goes to a different organ of the state, to the Health Information Quality Authority, and they conduct a HTA. […] The actual panel is made up of a number of experts right across screening and across*

communications. And there are two patient representatives on that board as well. So it's, all of their individual experiences, would inform the health technology assessment." (Interview partner 4, 00:01:54)

To support the review process, HIQA convenes a multidisciplinary expert advisory group (EAG). There are approximately 20 people on the EAG which is currently working on the SMA case, one of them being interview partner 3.

"In January they [HIQA] put together the expert advisory group. They put together the outline of the HTA which they shared with the expert advisory group. And we came back with recommendations and comments regarding the chapter headings and some of the way that they were thinking about it. We met once. We'll meet again in August. The HTA is expected to be concluded by the end of the year. And we are expecting that it will obviously be approved. So that's where we are.» (Interview partner 4, 00:20:27)

Once the HTA is conducted, the results are reported back to the NSAC. Based on these results, the NSAC formulates a recommendation regarding the inclusion of a specific disease in the NBS panel. This recommendation is then forwarded to the Minister for Health. the Minister for Health ratifies the NSAC's recommendation, thus paving the way for its implementation.

4.2.3 Power dynamics in Irish NBS policy-making of NBS

Prior to the NSAC's open call, power dynamics are already in motion. Families of patients, as well as the patients themselves, have the opportunity to express their concerns by writing letters to their local TDs. This allows them to influence the political discourse within the parliament. TDs can raise "Parliamentary questions" that are openly published, and answered by the Minister for Health. Following the 2022 International Neonatal Screening Day, where numerous families of SMA patients wrote to their TDs and invited them to an awareness event, several TDs posed inquiries about the inclusion of SMA in the NBS panel within the parliamentary.

So, for example, we picked eight of those sympathetic ones [TDs] in the week before the neonatal screening day, and all eight of them raised questions in our parliament." (Interview partner 3, 00:13:22)

The collaboration between patient organisations and pharmaceutical companies is another example of power dynamics that shape the political agenda. Pharmaceutical companies, driven by their goal of selling their treatments, often forge partnerships with patient organisations, providing financial support to some of them. In return, pharmaceutical companies gain insights into the experiences and needs of patients. These collaborations are also important for the patient organisations, as they raise their visibility within the government but also provide the organisations with access to the policy-making process. However, due to the government's scepticism towards such collaborations, it becomes essential for these partnerships to maintain a high level of transparency.

"Because the pharmaceutical companies take us seriously and they will, for example, put some money behind us to work with, say, PR companies. We put some campaigns together there to help us to get our website up and running and things like that. So all stuff like that helped us to from a collective of families to an actual organization. Because of their help, the government do now see us on the landscape as part of the equation. Um so when we went for SMA to be included, I was invited to join the expert advisory group. […] So the government at the same time, they are hyper, hyper, hyper sensitive about our relationship with pharmaceutical companies. I think they're worried that we could be pawns for the pharmaceutical companies because we're so desperate. We have no money and all we want is to see the drugs approved. And there is a skepticism. But I do believe that some of the government agencies, they're incredibly sensitive that there would be any question of unethical behavior. And so I go out of my way to be as uber transparent, which is why, you know, any [pharma company] funding or anything like that, it's all on our website published in our annual returns." (Interview partner 3, 00:07:04)

The NSAC, as a state actor, plays an important role in determining whether a new disease should be included in the NBS panel. As the primary authority in this process, the NSAC holds significant influence over the decision-making process.

"Well, I guess, the most power probably lies with NSAC. Once they make their recommendation. And then the power, would transfer to the senior management team in the health service to allocate resources, to bring this new test online. I guess the first decision point is, is NSAC's decision point. And my understanding is that they work as a collective." (Interview partner 4, 00:22:12)

The decision-making process within the NSAC lacks transparency, limiting the ability of non-state actors to exert influence. Non-state actors have no opportunity to participate in the process, and their attempts to engage with NSAC members are rejected.

«Well, the NSAC had a very strong chair. […] They ruled that committee with an iron fist. I tell you, I, for example, they had two patient representatives on the NSAC. One of them happened to be from the patient advocacy group for sickle cell. And because that person was sitting on the NSAC to give a kind of patient advocacy view to the questions and challenges that they face, I reached out to them. Right. So I'm a PHE. You're the PHE representative on the NSAC. Um, you know, thought it'd be a good idea if we could have a talk or whatever. Out of the question. They would not engage with me whatsoever. I was told in no uncertain manner that you're to bring your views to the committee. But that's where it ends. So I was really surprised. It is very closed. Almost secretive." (Interview partner 3, 00:34:22)

"The meetings and stuff of NSAC are all held in private. I think that they would be better served to actually, they publish minutes, but I think they would in fact be better served by making these things public and by inviting the relevant organisations in. HIQA, […] when they're coming towards the end of an assessment, they will convene a panel [EAG] to go through and make sure that everybody is happy or to provide input. But no such convening is done within with NSAC. And I think that as with all of these things, transparency would, would improve the situation. It's a closed door. There isn't any opportunity for the, the patient organisations or industry to present any information or anything, at the time of their decision making. Now today, everything that's gone through assessment has been approved. I guess the trouble will arise when the first thing does not get approved for addition. Then there will be much harder questions asked as to who's

making the decisions and what information they are using to make their decision" (Interview partner 4, 00:24:58)

After NSAC gives a recommendation to add a new disease to the NBS panel and receives ratification from the Minister of Health, the next crucial step is implementation. In Ireland, the NBS panel was last expanded in 2022, when ADA SCID was included for screening, utilizing an enzyme test. It is worth noting that while ADA SCID can be detected through an enzyme test, other forms of SCID require the use of a polymerase chain reaction (PCR) test for detection. As of now, this form of testing is not possible in the Irish NBS laboratory.

"So Ireland approved SCID back in 2022. But that was only the form ADA SCID, which was the form of SCID that could be could be picked up through the emzyne test. The other forms of SCID rely on a PCR test similar to the test for SMA. A genetic Test. So they approved. ADA SCID. Then at the beginning of this year, they recommended SMA should go for HTA and they approved wider SCID. So those SCIDs that need to be done using a PCR but in their approval, if you read the small print, they say that they recommend the approval of testing for the wider varieties of SCID, which relies on a test similar to SMA, which they believe both tests could be done in in a quite an efficient economic manner when the new hospital laboratory opens. And so here we are six months after they approved the wider form of SCID and it hasn't been implemented and we're still waiting for a SMA. But chances are this time next year SMA will be approved. But it won't be implemented. It won't be implemented, I would estimate, until possibly 2026. Interview partner 3, 00:45:18)

"It's just a slow process and the much bigger challenge is actually implementing then a recommendation. So even though something might be recommended by NSAC for inclusion, it doesn't mean that it actually is included within the health service. One condition was approved at the beginning of this year and they haven't even started to try and bring that online within the actual health service because they're out of lab capacity. They don't have the space for the new instrumentation that's required." Interview partner 4, 00:11:25)

The delay in implementing recommended diseases within the NBS panel poses a significant setback for patients and their families who have been advocating for the inclusion of these conditions. Patient organizations, in particular, face an intolerable situation due to these delays and are pushing for a solution.

> *"We're pushing very hard, that's just not good enough. That's just not good enough. We can't wait another three years. That's going to be another 25 babies potentially sent home and lives ruined. It needs to be approved. There's no excuse." Interview partner 3, 00:46:45)*

The implementation delay of recommended diseases within the NBS panel poses a significant challenge for the policy-making process of NBS in Ireland. Patient organisations are actively advocating for a resolution to tackle these delays, and it is yet to be determined how this issue will be addressed.

5 Discussion

5.1 Discussion of findings in relation to existing literature

5.1.1 Initiation of expansion of NBS panel

According to Jansen et al. (2017) there are two different possibilities on how the process of expanding a NBS panel can be initiated. First, the structural horizontal scanning approach and second the ad hoc approach. In Ireland, the NSAC functions as an independent body, as described in the structural horizontal scanning approach. NSAC has an annual open call where individuals and groups can suggest diseases for inclusion in the panel. The NSAC then selects some of these proposed diseases and recommends them for HTA by HIQA. However in the case of SMA, in both countries the most important factor that led to the application for SMA to be included in the NBS panel was the availability of a modifying treatment for the disease. In Ireland, patient organisations started campaigning, when SPINRAZA® was approved, which lead to eight applications during the NSAC open call. In Switzerland, there is no body similar to the NSAC. In the case of SMA the most important factor that lead to the application was also the availability of modifying treatments. In Switzerland it is a requirement for submitting an

application, as it is mandatory to have a modifying treatment available for the disease in question.

5.1.2 Policy triangle

This thesis examined the policy-making processes regarding the inclusion of new diseases in the NBS panel in both Switzerland and Ireland. In line with the policy triangle framework by Walt and Gilson (1994), this thesis incorporated not only an analysis of the content of a policy itself, but also placed equal significance to the contextual factors (health care system), key actors involved, and the decision-making process itself. When comparing these factors within the NBS policy-making process of Switzerland and Ireland, three main differences arise.

1. Initiation of the submission/application phase

In both Switzerland and Ireland, the initial step towards the inclusion of a new disease in the NBS panel typically involves non-state actors such as patient organizations, pharmaceutical companies, and physicians. These actors play a crucial role in identifying and advocating for the addition of specific diseases to the screening program. However, in Ireland, the HSE has the authority to initiate a submission for the open call of the NSAC. This means that the HSE can actively propose and advocate for the inclusion of specific diseases in the NBS panel. On the other hand, in Switzerland, the BAG does not have the same possibility to initiate an application. The BAG's role is more focused on policy implementation rather than actively proposing additions to the screening panel.

In addition, in Switzerland it is also necessary to submit a second application to the EAMGK, to make sure that the screening of a new disease is financially covered. This is not the case in Ireland.

2. Influence of patient organisations and advocacy groups

In Ireland, patient organisations and advocacy groups have a greater opportunity to influence the policy-making process compared to Switzerland. In Ireland, patients and their families have the possibility to influence the NBS policy-making process by writing letters to their local TDs. By reaching out to their elected representatives, patients and their families can contribute to the ongoing discussions and decision-making within the parliament, raising awareness and emphasizing the importance of certain diseases in the

NBS panel. The case of SMA shows, how effective this approach can be. In addition, Ireland has the EAG, which has a direct influence on the decision-making process. The EAG may include representatives of patient organisations. This inclusion ensures that the perspectives and needs of patients and their organisations are considered during the decision-making process. Additionally, the EAG is assembled for each case, meaning that there is a specific EAG for each condition being considered for inclusion in NBS program. In contrast, Switzerland relies on the NEK and the GUMEK to make decisions regarding each application. Patient organizations and advocacy groups do not have a direct influence on these committees in the Swiss system. In addition, in Switzerland, patient organisations and advocacy groups do not have the possibility to influence the decision-making process regarding the inclusion of new diseases in the NBS panel. While they were consulted during the development of articles of law and ordinances related to NBS, their influence specifically on the inclusion of diseases is limited. They do not have a direct role in determining which diseases are included in the screening panel.

3. Role of laboratories in policy-making

The Swiss NBS policy-making process involves a significant role for the NBS laboratory. The laboratory itself takes responsibility for the application process, which includes proposing and justifying the inclusion of a disease in the screening panel. They initiate the application process when approached by non-state actors with the intention to include a new disease in the screening panel. The close involvement of laboratories from the beginning simplifies implementation of diseases into the screening panel. This early involvement facilitates discussions regarding the feasibility of implementation. In contrast, the laboratory's role in the Irish NBS policy-making process is not explicitly mentioned. This may explain the recent delay in implementing recommended diseases to the NBS panel.

5.1.3 Three dimensions of power

Building upon the three dimensions of power by Lukes (2005), this thesis also incorporates a power dynamics analysis of the NBS policy-making processes of Switzerland and Ireland.

1. Power as decision making

According to Dahl (1957) there are multiple actors within a policy-making process. While some of these actors may not be traditionally perceived as the most dominant or influential, they still exert significant influence over the process. In both Switzerland and Ireland, besides the dominant state actors, like the BAG, the NEK, the GUMEK, and the EAMGK in Switzerland and the HSE, NSAC, and HIQA, there are also non-state actors that are very important in the policy-making process. These non-state actors are namely patients, their families, patients organisations, physicians, and the pharmaceutical companies. As previously stated, these non-state actors have more possibilities to engage with the policy-making process in Ireland, as for example through the EAG, than they have in Switzerland.

In Switzerland, the inclusion of a new disease follows a strict application process prescribed by law without much room for discussion or inputs from non-state actors. Switzerland follows a power as policy making approach, where dominant state actors have the biggest influence in the process of including a new disease in the NBS panel.

2. Power as non-decision making

The second dimension of power, as outlined by Bachrach and Baratz (1962), posits that significant processes and discussions occur before the decision-making stage. These pre-decision processes play a crucial role in shaping the political agenda and have a considerable impact on the final outcomes. Moreover, they mention there may be actors, trying to prevent a policy to be introduced on the agenda in the first place, or actively work to obstruct its acceptance even after it has been proposed. Neither in Switzerland nor in Ireland could actors be identified who are trying to prevent an extension of the NBS from being put on the political agenda. However, during the discussions regarding the potential addition of a new disease to the NBS panel, certain voices within the Swiss committees responsible for evaluating the application may express concerns regarding the associated costs of introducing a new disease. Up until now, despite these concerns being raised, they have not been able to prevent the inclusion of a disease in the NBS panel.

In Ireland, the decision-making process within the NSAC is perceived to have limited transparency. Consequently, it is not possible to address the question whether there are

any voices within the NSAC attempting to obstruct the addition of diseases to the NBS panel. In terms of influencing the political agenda, Ireland offers more opportunities for non-state actors to do so than Switzerland. In Ireland, non-state actors have the possibility to engage with their local TDs and raise the awareness of certain topics. This approach has shown to be very influential, as the case of SMA has shown. TDs can bring these issues to the parliament for further discussion, putting them on the political agenda.

3. Power through control

The third dimension of power, as conceptualised by Lukes (2005) is arguably the most abstract dimension of power and also the one most difficult to measure. According to Lukes it goes beyond the observable processes of decision-making and agenda-setting. This dimension concerns the shaping of the political consciousness and social reality of individuals or groups. Indeed, a significant way in which political consciousness is shaped can be seen when patient organisations invite TDs to awareness days regarding a certain disease, in the hope that these representatives will raise and discuss issues related to NBS or specific diseases in the parliament. In Switzerland a similar event, including neuro paediatricians and pharmaceutical companies, was held, which lead to the application for inclusion of SMA in the NBS panel. This event may also have shaped the social reality for some stakeholders present at the event, leading to the application in the end.

On another note, the Irish government is attentive to the potential shaping of political consciousness when collaborations between pharmaceutical companies and patient organizations take place. They are particularly cautious about such alliances, and as a result, patient organizations proactively disclose these collaborations in a transparent manner.

Regarding the political consciousness of NBS panel expansion, it appears that there might not be a widespread discussion among the general Swiss and Irish citizens. Typically, individuals become aware of NBS when they directly encounter it, either through patient organizations or when they become parents. However, based on insights from interview partner 1, it is deemed crucial that a political consciousness about NBS develops within the broader public in the future.

5.2 Limitations

This thesis had the aim to identify relevant actors within the policy-making process of the expansion of NBS panels in Switzerland and Ireland. In addition, the goal was to analyse power dynamics within this process. According to Walt and Gilson's policy triangle, investigating the context of a policy is crucial. As such, health systems were mentioned as examples. However, when examining NBS policies in Switzerland and Ireland, it becomes evident that the health system did not offer significant insights for the analysis.

Another limitation of this thesis lies in the number of conducted interviews, as having more interviews from diverse stakeholders might have provided a clear understanding of the power dynamics at play.

In the case of the Swiss interviews, no interviews were conducted with members of patient organisations. On the other hand, the Irish interviews involved representatives from patient organizations, offering a unique perspective. Comparing the views of patient organizations from both countries could have provided interesting insights.

Additionally, obtaining an interview with a member of the NSAC in Ireland would have provided valuable information about the decision-making process within the most significant authority in Irish NBS policy-making.

Finally, including interviews with pharmaceutical companies involved in modifying treatments would have added a crucial dimension to the research, shedding light on their perspectives and influence in the policy-making process.

6 Conclusion

6.1 Summary and key findings

The most relevant actors within policy-making for the inclusion of new diseases in the NBS panel of Switzerland and Ireland were identified within this thesis. Both countries have stat and non-state actors who influence the decision making process. The main difference between the involved actors is that in Switzerland, the laboratory responsible for the implementation of NBS is a key actor within the process. The laboratory, in close

collaboration with other non-state actors, is the applicant for the inclusion of a new disease to the NBS panel. This is not the case in Ireland. Including the laboratory is an important step, as this ensures that once a disease is added to the NBS panel, the implementation runs smoothly and without any issues.

Another notable difference in actors lies in the advisory committees within the policy making process. Switzerland has the GUMEK and the NEK who are responsible for the evaluation of each application. In Ireland, HIQA conducts the HTA of each disease, but in addition there is a EAG specifically formed for the HTA for each disease, ensuring that experts with relevant expertise in the specific disease are present during the assessment process.

There is a clear difference in the involvement of patient organisations between the policy making of NBS in Switzerland and Ireland. In Switzerland, patient organisations were consulted during the development of articles of law and ordinances related to NBS, but they have smaller influence in the policy-making process of including a new disease to the NBS compared to Ireland.

The initiation for the process of adding a new disease to the NBS panel in the case of SMA in both countries was driven by the availability of new modifying treatments indicating a leaning towards the *ad hoc* approach. However, it is important to note that in Ireland, the NSAC, as a government-formed body, is responsible for the inclusion of new diseases in screening panels, aligning with the horizontal screening approach. Overall, Switzerland distinctly follows the ad hoc approach, while Ireland, on the other hand, exhibits a combination of both the ad hoc and horizontal screening approaches.

Altogether, all three dimensions of power are present in the NBS policy making process in both Switzerland and Ireland. Due to the strict applications process in Switzerland, the first dimension is probably more present in this country. This also leads to the fact, that the second dimension, concerning agenda setting is more present in Ireland. The third dimension, regarding political consciousness is difficult to observe. However, in both countries, Switzerland and Ireland, there were awareness events, shaping the political consciousness of relevant stakeholders, be it state actors like the TDs in Ireland, or non-

state actors like physicians and pharmaceutical companies, that then played a big role in the initiation of the application process.

6.2 Future directions for research

For future research within the field of policy-making of NBS, it is essential to involve pharmaceutical companies in the research process through interviews. Including these stakeholders will offer a different perspective and provide insights that might not have been previously considered.

Moreover, further research should focus on the implementation of recommended and approved diseases into the NBS panel. Understanding the reasons for delays in Ireland's implementation process is crucial to prevent such setbacks in the future, ensuring that patients and their families do not have to endure prolonged waiting periods after campaigning for a disease to be accepted in the NBS panel. Research on this topic will help patients benefit from effective screening processes, leading to timely diagnoses that can ultimately be life-changing.

7 Declaration of independency

I hereby declare that I have written this thesis independently and have not used any auxiliary materials other than those indicated. All passages taken verbatim or in spirit from sources have been marked as such. I am aware that otherwise the Senate is entitled to withdraw the degree conferred on the basis of this thesis in accordance with the University Act.

Bern, 25 July 2023 Alissa Müller

8 Declaration of consent for the publication of the master thesis

I hereby declare that I consent to the publication of the Master's thesis I have written on the KPM homepage in the event of a grade of 5.0 or higher. The thesis is publicly accessible.

Bern, 25 July 2023 Alissa Müller

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