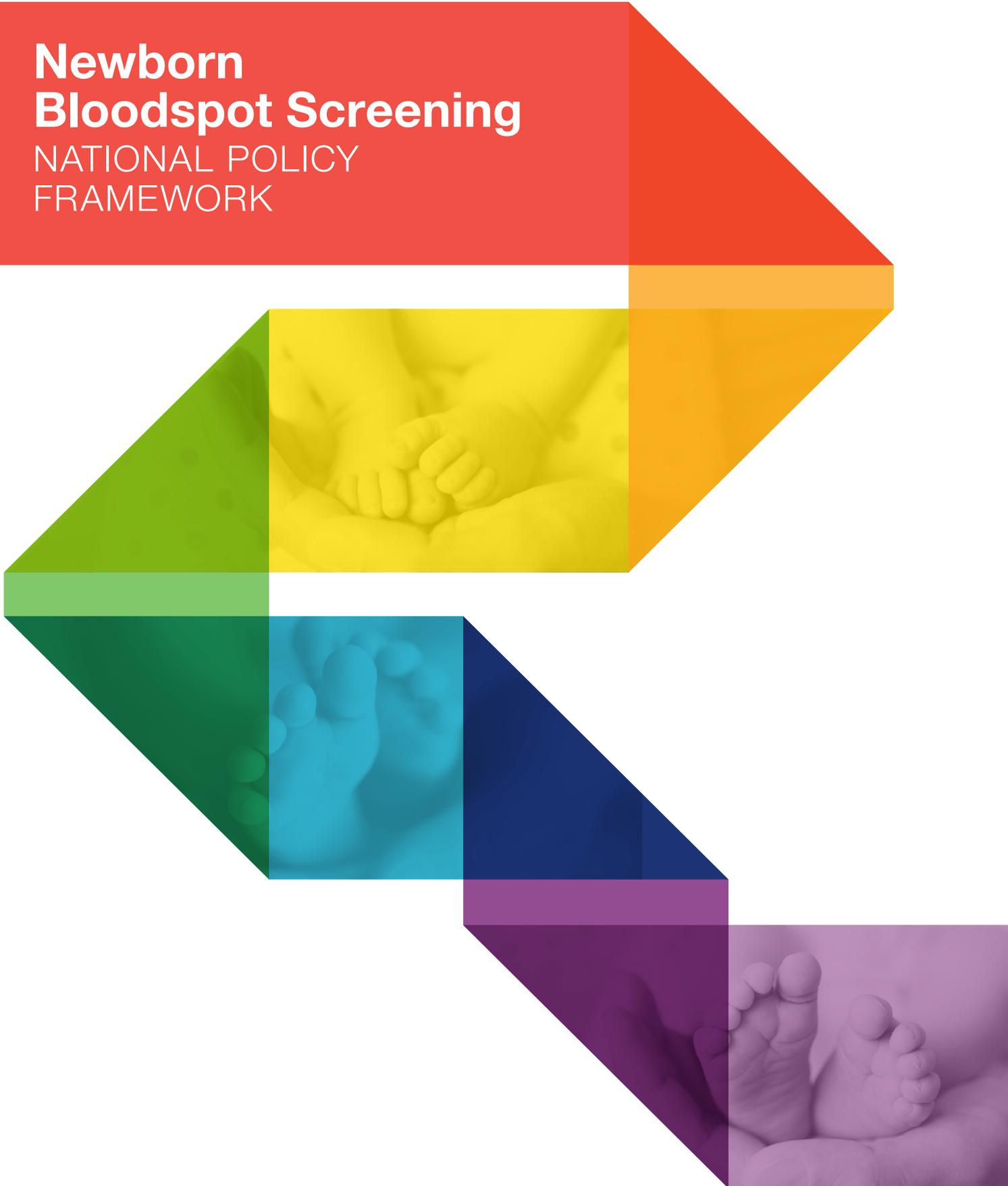




AUSTRALIAN HEALTH MINISTERS'
ADVISORY COUNCIL

Newborn Bloodspot Screening

NATIONAL POLICY
FRAMEWORK





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Newborn Bloodspot Screening National Policy Framework

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Foreword

Newborn bloodspot screening is an exemplar of what a successful population health program can deliver. Since the 1960s, a simple blood test at birth has meant that thousands of Australians have avoided lifelong disability or death from a range of conditions.

Since their inception, newborn bloodspot screening programs have evolved, increasing the number of conditions they screen for and the benefits they offer. This has been achieved thanks to the significant efforts and foresight of those individuals at the helm of these important programs and their highly skilled teams.

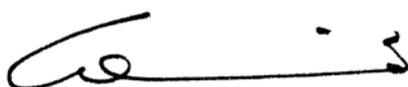
New technologies, treatments and understanding of conditions, coupled with a health care system constantly being asked to deliver more, mean that the newborn bloodspot screening programs are under new and evolving pressures. The changing environment means that now, more than ever, there is a need to reflect on what works well and put in place a clear national framework that enables the programs to successfully navigate the opportunities ahead.

It is with great pleasure that I present to you this landmark Newborn Bloodspot Screening National Policy Framework. For the first time in the newborn bloodspot screening programs' histories, they are united through a nationally agreed vision and way of working. The framework highlights what works well, builds upon the successes of the programs and provides mechanisms to enable the programs to grow and adapt into the future. It provides guidance to those who deliver and support newborn bloodspot screening and also paints a clear picture for the millions of Australian families that participate in the programs.

I am extremely proud of what is presented in this policy framework. It has been carefully crafted over 18 months of intense work and stakeholder engagement. My sincere thanks go to the members of the Newborn Bloodspot Screening Working Group, and the secretariat and project management team. This group of experts has worked collaboratively and productively, investing both professionally and personally in what is presented here.

Importantly, my thanks go to the hundreds of consumers, clinicians, midwives, nurses, policy makers, scientists and others who contributed to the policy framework through the stakeholder workshops and survey. Engagement of those people who are involved in, and affected by, newborn bloodspot screening has been a priority of the working group.

And, of course, my thanks go to those of you who are now responsible for taking this policy from paper into practice. In doing so, you will help to ensure that newborn bloodspot screening in Australia continues to be known as one of the most successful population health initiatives of our time.



Clinical Associate Professor Craig White

Chair, Newborn Bloodspot Screening Working Group



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Summary of policies

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2.2 Program roles and responsibilities are clearly defined, including at the national, state and territory and operational levels.

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- 3.1 All programs are supported by clear and effective governance processes and structures.
- 3.2 Health professionals who support and deliver newborn bloodspot screening take action to promote quality and safety.
- 3.3 Facilities, equipment and work processes are designed for quality and safety.
- 3.4 Programs take action to extend excellence, and prevent and minimise harm.
- 3.5 The programs, including all elements of the screening pathway, are adequately funded and resourced.

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- 5.2 Decisions to add or remove conditions from newborn bloodspot screening programs must be made in line with agreed criteria.

Introduction

The purpose of this policy framework is to provide an overview of the elements that are needed to successfully deliver newborn bloodspot screening in Australia. The policy framework outlines high-level policies and recommended steps that support high-quality and family-focused newborn bloodspot screening. The intended audience for the policy framework is anyone involved in, or affected by, newborn bloodspot screening. This includes clinicians, families, midwives, nurses, policy makers, program managers and scientists.

GUIDING PRINCIPLES

The policy framework is built upon the following key guiding principles:

- Australian newborn bloodspot screening programs work well and protect babies from the effects of life-limiting conditions.
- The environment in which newborn bloodspot screening programs are delivered is changing.
- Clear, national approaches are needed to support programs into the future.
- Any future developments should be focused on conditions and should not be driven by technology.
- Screening must remain high-quality and safe.
- Families should remain the central focus of the programs.

Overview

This policy framework describes what is needed to support the ongoing success of newborn bloodspot screening in Australia. This introduction and the background that follows provide information on newborn bloodspot screening in Australia and the policy framework. The later chapters present five policy areas, which outline:

- guiding principles and a high-level description of the programs (Policy Area 1)
- how the programs are implemented (Policy Area 2)
- what is needed to support high-quality and safe newborn bloodspot screening (Policy Area 3)
- how the programs should be monitored, reviewed and evaluated to ensure they are achieving their aim and objectives (Policy Area 4)
- a decision-making process to enable conditions to be assessed for inclusion in or removal from the programs (Policy Area 5).

Each policy area includes a number of policy statements, which are built on the best available evidence and describe the essential elements of an effective newborn bloodspot screening program. Each policy statement is followed by a context, which outlines why the policy statement is important and defines any key concepts. A number of recommendations are also provided. These recommendations are steps that can be followed to deliver the programs in line with the policy statement. Those people who are supporting and delivering the programs should use the policies and recommendations to inform local approaches.

In most cases, what is presented here already occurs in the programs. This is because the programs have been in operation for more than fifty years and are highly successful. Therefore, the intent of this framework is to capture what works well, identify and respond to any gaps that exist, and put in place a framework that supports future development.

Background

NEWBORN BLOODSPOT SCREENING IN A NUTSHELL

Newborn bloodspot screening has been offered to newborns in all states and territories in Australia since the 1960s. Through the programs, a test is offered to all families in the first few days after the birth of a child. The test is provided at no cost to the families. The midwife or nurse first provides information about the test to families. The family can then decide whether to undergo screening. If they agree, as is almost always the case, the baby's heel is pricked and blood is collected on a special filter paper card.

The bloodspot card, which also includes the baby's name and other details, is sent to a specific laboratory. It is here that the bloodspot is tested to see if the baby is at risk of a range of conditions. If the results of this test suggest the baby is at risk of having one of these conditions, laboratory staff quickly get in touch with health care providers. The health care providers then arrange for the baby to receive urgent care if required or have further testing that will confirm whether the baby does indeed have the condition.

The benefit of the test is that, if a baby is found to have a condition, they are able to then receive treatment or management earlier than would have otherwise been possible. This earlier intervention leads to better health outcomes for the baby and the family. In Australia, about 99 per cent of babies receive newborn bloodspot screening. This means that more than 300 000 babies are screened each year. Of the babies screened, around one in every thousand has a condition that would otherwise have gone undetected.

NEWBORN BLOODSPOT SCREENING IS COMPLEX

While collecting blood on a piece of card and testing it might sound simple, it is not. The science around bloodspot screening is complex. Before a condition is included in the newborn bloodspot screening program, program scientists need to clearly define what a 'normal' or healthy result is and what an 'abnormal' result is— an abnormal result being one that would suggest a baby has the condition. This process takes time but is a vital step, as it then enables every baby's result to be assessed in the same way. Newborn bloodspot screening is also complex because it involves coordinating a great number of people and organisations across Australia doing different jobs to support the families that participate in the programs.

HOW THE PROGRAMS ARE ORGANISED

Newborn bloodspot screening programs in Australia are funded by state and territory governments and operate independently of each other. The testing of dried bloodspots is undertaken by five major laboratories, which are located in New South Wales, Queensland, South Australia, Victoria and Western Australia. For those states and territories with no screening laboratory, dried bloodspots are sent interstate for testing. In addition to these local-level operations, national decision-making and information support mechanisms exist. These mechanisms are detailed within this framework.

A BRIEF HISTORY OF NEWBORN BLOODSPOT SCREENING IN AUSTRALIA

Australian newborn bloodspot screening programs have evolved over time, from screening for a single condition to screening for more than 25 conditions. Newborn bloodspot screening in Australia started in the 1960s with screening for phenylketonuria. This condition has no clinical signs at birth and can lead to severe intellectual disability, with affected babies losing about four IQ points every month while untreated.

The next condition to be added to the programs after phenylketonuria was congenital hypothyroidism. This leads to both intellectual and physical disability if left untreated. Screening for this condition began in two states in 1977, with other states and territories starting screening in the following years. The programs were then further expanded to include cystic fibrosis. This screening started in 1981 in New South Wales, and all other states and territories were screening for the condition by 1999. Another condition added early in the programs' histories is galactosaemia. Screening for this condition started in all states and territories except one in the early 1980s. This remains the case today, with the condition being diagnosed through normal clinical practice rather than newborn bloodspot screening in Victoria.

A huge change in newborn bloodspot screening worldwide came about with the introduction of tandem mass spectrometry into newborn bloodspot screening programs. This technology made it possible to screen for many conditions at the same time using a single blood sample. Australia began using this technology in two states from 1998, and it was used in all programs by 2005. As a result, the number of conditions screened in Australia was vastly increased, in line with what we see today. While this ad-hoc process for expanding screening has worked well in the past, the number of conditions that may now be considered for screening means that it is important for future changes to the range of conditions screened to be based on transparent and robust decision making [1]. This policy framework outlines an agreed method to do exactly that by providing a national approach to assess conditions for inclusion in, or removal from, Australian newborn bloodspot screening programs.

WHAT TO SCREEN

There are a large number of conditions that may affect newborns. It is important to note that it is neither appropriate nor safe to screen for *all* of these conditions. While newborn bloodspot screening is highly effective for the conditions it currently screens, it may be less effective for other conditions. This is because health or wellbeing of the child or family is not always improved by identifying a condition early. Worse still: screening every baby born for a condition could potentially lead to babies receiving intervention that may not be useful or may even harm them. Therefore, careful consideration is needed when deciding which conditions to screen.

WHY A NATIONAL POLICY FRAMEWORK IS REQUIRED

Newborn bloodspot screening is recognised around the world as a highly successful population health program. In Australia, thousands of children have been spared intellectual and other disabilities, and in some cases death, through newborn bloodspot screening. In addition to the improvements in health and wellbeing in individual families, there are considerable benefits at the population level. As such, there is value in supporting these programs into the future.

While the programs are currently successful, more and more pressures are being placed on them. More is being learnt about the conditions being screened for as well as about conditions not currently included in the programs. The technology related to the testing and management of conditions is also rapidly changing. This means that there are calls to include other conditions in the programs. In addition, across health care, there has been a shift in the way that families and consumers are involved in their care.

In addition to the changing environment, there is also greater recognition of what it takes to implement a screening program. Australian governments have outlined the key elements of a screening program in the *Population Based Screening Framework* [5], which did not exist when newborn bloodspot screening commenced. Given this, and the above pressures, there is the need for a national policy framework that reflects what currently works well, while also supporting the newborn bloodspot screening programs so that they can continue to be successful into the future.

BENEFITS OF A NATIONAL POLICY FRAMEWORK

The most effective screening programs are those that are implemented nationally [2]. A national approach can enhance consistency, cost effectiveness, transparency and accountability. This national policy framework has been established to capitalise on these benefits while supporting local implementation. Further, this framework recommends, for the first time, a national governance approach for newborn bloodspot screening to support policy direction, monitoring and decision making. In so doing, it supports equity across Australia and minimises duplication of effort.

This policy framework encourages those working in the programs to share ideas, pool data and work together to inform the future direction of newborn bloodspot screening. Given the rarity of the conditions detected through newborn bloodspot screening, information and data must be shared in this way and considered nationally. Only in doing so is it possible to accurately consider the clinical benefits as well as the risks and harms of screening for a particular condition. A national approach also enables individual programs to assess their performance, share successes and identify areas for improvement. Given these factors, the policy framework will benefit families, programs and governments.

HOW THE POLICY FRAMEWORK WAS DEVELOPED

The policy framework was developed by the Newborn Bloodspot Screening Working Group on behalf of the Standing Committee on Screening (SCoS). SCoS is the national committee that provides advice to the heads of all health departments in Australia on national screening issues. Early in the process, the working group recognised that the success of the policy framework relied on working with the range of people and organisations that are interested in newborn bloodspot screening, including families. As such, the policy development process involved two consultation workshops and a consultation survey to maximise the opportunities for people to provide input.

In addition to stakeholder input, and the expertise on the working group, the policy framework was informed by the existing practices of newborn bloodspot screening programs in Australia. The working group also considered relevant policy documents, including: the Australian *Population Based Screening Framework*; frameworks for newborn bloodspot screening programs from other countries; policies for other screening programs in Australia and overseas; and the work of the Australian Commission on Safety and Quality in Health Care. The policy framework also draws upon relevant academic literature. The working group used this range of information to develop policies that take into account the health system within which newborn bloodspot screening is delivered. As such, what is presented here is a well informed and considered policy that can be implemented within the local settings.

Further details on the working group and the policy development process are provided in [Appendix A](#).

HOW THIS POLICY FRAMEWORK SHOULD BE USED

This policy framework should be used by any health professional who is involved with newborn bloodspot screening. This includes clinicians, midwives, nurses, policy makers, program managers, scientists and others. Ideally, these individuals and their colleagues should be using this policy framework as a guide to then inform local policies, processes and practice. This framework can also be used by anyone who wishes to know, in more detail, about newborn bloodspot screening.

ONGOING MAINTENANCE OF THE POLICY FRAMEWORK

A useful policy document will be dynamic and able to change and evolve in response to new evidence and ideas. In line with this, the policy framework has been developed for easy review and update. Such review can be completed as needed under the guidance of SCoS.

Policy Area 1: Program overview

A newborn bloodspot screening program encompasses the policies, processes, people and facilities that are involved in providing newborn bloodspot screening. The purpose of this chapter is to provide an overview of the key elements of newborn bloodspot screening programs in Australia. This includes outlining a national aim and objectives, the target population and a screening pathway. The following also recognises newborn bloodspot screening as a population based screening program, and outlines the associated requirements. Finally, guidance is provided for ensuring that newborn bloodspot screening programs are delivered in line with clinical guidelines and legislation.

PART I: AIM AND OBJECTIVES OF NEWBORN BLOODSPOT SCREENING

1.1 All programs are delivered in line with a nationally agreed aim and objectives

Context

A successful screening program requires concerted and organised efforts from multiple stakeholders across the health sector. To support these efforts, it is necessary to have a nationally agreed aim and objectives that stakeholders will work towards and that provide strategic program direction [3, 4]. In doing so, individuals in each of the state and territory programs can focus their efforts and steer newborn bloodspot screening across the country in the one agreed direction.

Recommendation

Programs use the nationally agreed aim and objectives contained in Box 1 to guide local practice. The aim and objectives should inform, and be articulated in, relevant local policies, training, information and other documentation.

BOX 1: NATIONALLY AGREED AIM AND OBJECTIVES FOR NEWBORN BLOODSPOT SCREENING

The aim of newborn bloodspot screening is to improve the health of babies by identifying those at risk of developing a serious condition early, generally before symptoms present, thereby enabling earlier intervention.

The objectives are that newborn bloodspot screening programs:

- provide quality, timely and evidence-informed screening to all newborns in Australia
- enable early detection of individuals at risk of conditions screened, in order to reduce the morbidity and mortality of the relevant conditions
- support referral to enable early diagnosis and management of conditions identified through screening
- maximise program participation and public trust
- encourage strong partnerships across health systems, focused on the delivery of high-quality newborn bloodspot screening
- support timely communication of high-quality information to families on all aspects of newborn bloodspot screening
- develop and learn through continuous improvement processes that assess short- and long- term program performance.

PART II: PROGRAM DESCRIPTION

1.2 Programs are delivered in line with the agreed definition of a population based screening program

Context

It is essential that there is a common language upon which the programs are based. This ensures that families, governments, maternity service providers, scientists and others have a shared understanding of what the programs involve. A cornerstone to this shared understanding is the recognition that newborn bloodspot screening is a population based screening program. In line with this, newborn bloodspot screening refers to the organised, systematic approach of testing all newborns for conditions, generally before symptoms present.

Recommendations

- Newborn bloodspot screening programs are recognised as population based screening programs. Australian governments agree that population based screening programs have:
 - an identified target population
 - invitation and recruitment mechanisms
 - information and support for participants
 - workforce and facilities to enable screening and the interpretation of screened material
 - health care services for the diagnosis and appropriate management of conditions detected through screening and for the follow-up of individuals who have been treated or require monitoring
 - quality improvement processes, including education and training for health care providers and laboratory staff
 - systematic monitoring and evaluation [\[5\]](#).
- In order to adhere to the definition of a population based screening program set out above, a newborn bloodspot screening program should be:
 - understood to encompass all the people, processes, policies and facilities involved in delivering and governing newborn bloodspot screening
 - delivered in line with the policies included within this policy framework.

1.3 The target population for newborn bloodspot screening is all newborn babies in Australia

Context

Australian governments have agreed that an essential component of a screening program is a clearly defined target population [5]. The target population for a screening program is the group of people who will most benefit from screening for the agreed condition(s). Focusing efforts on an agreed target population will ensure that a program delivers the most benefit to the population and resources are concentrated on evidence-informed screening.

Recommendations

- The target population for newborn bloodspot screening is all newborn babies in Australia, noting that there may be circumstances where babies outside of this target population are eligible for screening.
- Maternity service providers must have processes in place to ensure that all families are offered newborn bloodspot screening for their newborn.
- Samples should be collected forty-eight to seventy-two hours after birth, being the optimal time frame for conditions currently included in newborn bloodspot screening programs.
- Local policies must outline approaches to support screening of babies who cannot have a sample collected within the optimal time frame but who may still benefit from screening.

1.4 Newborn bloodspot screening is delivered in line with the agreed screening pathway

Context

A screening pathway is the journey that a family will follow when they participate in a screening program and the work processes that facilitate this journey. For newborn bloodspot screening, the pathway starts at the point a family is first made aware of the program and ends when a normal result is reported to the maternity service provider or when the family is referred to an appropriate health care provider for diagnostic testing and follow-up care. This means that diagnostic testing, condition management and formal psychological support for families all lie outside of the screening pathway. However, programs take steps to support relevant families to access services beyond the scope of the program in a timely manner. Following these steps and having a clearly defined screening pathway means that families can be prepared for what screening might involve outside of the sample being taken. It also enables staff involved in the programs to understand their roles.

Recommendations

- The programs are delivered in line with the national screening pathway, which is outlined at [Appendix B](#).
- Strong links exist between the maternity service providers, laboratories that undertake diagnostic testing and the health care providers involved in follow-up care.

1.5 Newborn bloodspot screening is provided in line with relevant clinical guidelines and legislation to support high-quality screening and care

Context

It is essential that screening and work processes occur along the screening pathway in line with best practice, evidence-informed approaches and the law. To ensure this is the case, all programs are required to operate in line with relevant clinical guidelines, legislation and policies. Doing so supports consistency of practice, which is linked to high-quality outputs [5]. Therefore, adhering to this policy statement protects families, health care providers, laboratory staff, maternity service providers and the programs.

Recommendations

- Newborn bloodspot screening is supported by clinical guidelines to ensure quality of care. These may include, but are not limited to, clinical guidelines relating to:
 - general practice
 - neonatal care
 - obstetrics and midwifery care
 - paediatrics
 - pathology
 - information sharing.
- Newborn bloodspot screening adheres to relevant legislation, standards and policies. These may include, but are not limited to:
 - privacy legislation
 - information legislation
 - health and public health legislation
 - health insurance legislation
 - human tissue acts
 - criminal legislation
 - freedom of information legislation
 - law and other relevant guidance relating to the ethical conduct of research
 - relevant professional organisations' policies and accreditation processes.

Policy Area 2: Program implementation

This policy area provides an outline of how programs should be implemented. In essence, this relates to the components of the program that are physically delivered. Specifically, the following information defines the responsibilities of governments, maternity service providers and laboratories. Policies are also included that highlight the need to maximise participation in the program, minimise inequities and respond to the information needs of families along the screening pathway. The chapter also provides policies on taking and managing the dried bloodspot, from the time screening takes place until the sample is destroyed.

In addressing the above, this policy area aligns newborn bloodspot screening with national guidance. It highlights that screening programs are most effective when they are organised, their components are clearly defined and they are implemented in a consistent manner across a country [5]. Describing how the programs should be implemented and where responsibilities lie enables people delivering the programs to have a clear understanding of their role in the screening process. It also makes it possible for families to know what to expect from the programs. Furthermore, having a clear description of the programs increases transparency and enhances efficiency.

This policy area provides for a baseline level of consistency between the programs. Ideally, screening programs should be managed nationally and implemented in the same way across the country. Adhering to this principle would mean that families are provided exactly the same program, no matter where they live. The current Australian newborn bloodspot screening programs have evolved separately over time. While they all operate successfully, they are managed at the state and territory level and, as a result, there are nuances in the ways in which they are implemented. Therefore, the policies that follow are high-level. They provide a consistent foundation for program implementation but respect the autonomy of individual states and territories. They also provide a starting point for working together to achieve further consistency into the future.

PART I: PROGRAM DESIGN

2.1 Programs are designed and operate to maximise participation in screening and minimise inequities

Context

For a screening program to be effective, the people who will benefit the most from the program must be able to access it. Maximising participation ensures that the greatest number of newborns and families can benefit. This leads to population-wide reductions in morbidity and mortality and increases the cost-effectiveness of the programs.

Recommendations

- Evidence-informed strategies are developed for recruiting people in the target population, including subgroups and populations at risk of being under-screened, such as:
 - Aboriginal and Torres Strait Islander families
 - families living in remote areas
 - families with culturally and linguistically diverse backgrounds
 - premature babies
 - babies in neonatal intensive care units
 - babies who are seriously ill
 - babies born at home or with the assistance of independent midwives.
- Equity of access and outcome are considered and incorporated into the design of the program. This includes considering equity across public and private health care sites; for babies born at home; for families in metropolitan, rural and remote settings; and across state and territory borders.

2.2 Program roles and responsibilities are clearly defined, including at the national, state and territory and operational levels

Context

Newborn bloodspot screening is offered in public and private settings and involves a range of different stakeholders. To ensure the success of newborn bloodspot screening programs and to support families to move seamlessly along the screening pathway, the roles and responsibilities of organisations and individuals must be clearly defined. Doing so means that all individuals that support the screening pathway understand how they contribute and work together to deliver an effective screening program. Having clearly defined responsibilities ensures accountability, supports efficiency and reduces duplication of effort.

Recommendations

- National responsibilities relating to policy direction, decision making and information sharing are described in the national governance structure outlined in *Policy Area 3: Quality and safety*.
- State and territory governments have overall responsibility for the delivery of newborn bloodspot screening within their jurisdictions. Generally, this responsibility includes:
 - overseeing state- or territory-wide policy development and program management
 - provision of program funding
 - monitoring program performance, quality improvement, evaluation and review.
- Roles and responsibilities of maternity service providers, laboratories and other key stakeholders:
 - are agreed by the state or territory government and clearly documented
 - include the roles and responsibilities listed in Box 2.

BOX 2: ROLES AND RESPONSIBILITIES

Program delivery

This should be the responsibility of maternity service providers. It includes the following roles:

- developing relevant operational policies and procedures to support a safe and quality program
- ensuring appropriate information is provided¹ to families
- ensuring all families are offered screening for their baby
- ensuring necessary resources for bloodspot sampling are available
- ensuring midwives, neonatal nurses and relevant staff receive continuing support and education
- ensuring timely transport of dried bloodspots to the laboratory
- ensuring families are aware of the process for handling all results and how these results may be accessed
- ensuring every baby has a recorded screening result or refusal
- nominating a liaison person (e.g. community liaison midwife or nurse unit manager of maternity) to have responsibility for newborn bloodspot screening in each health care setting delivering maternity services
- supporting program-wide quality improvement and safety
- ensuring timely follow-up for babies who have abnormal results or who require further samples to be taken
- ensuring the diagnostic results are provided to the newborn bloodspot screening laboratory for those babies identified as being at increased risk of having a condition².

Dried bloodspot testing and management, and program support

This should be the responsibility of laboratories. It includes the following roles:

- developing information for families, in collaboration with other stakeholders where required, and making it available to maternity service providers
- supplying and distributing bloodspot cards to maternity service providers
- ensuring that both the sample and information contained on the dried bloodspot cards are processed in a timely manner
- managing dried bloodspots, including their retention, storage, secondary use, release and destruction
- ensuring all results are reported to maternity service providers in a timely manner
- reporting delays in receipt of dried bloodspots to maternity service providers
- promptly notifying an appropriate health care provider when the screening result is abnormal
- ensuring that the family of a baby with an abnormal result is contacted by the appropriate health care provider for diagnostic testing in a timely manner
- issuing timely requests for repeat samples to maternity service providers
- providing administration, quality assurance and improvement, and management services to support the effective provision of newborn bloodspot screening
- providing and managing a high-quality, secure information management system to support the program. This includes keeping a record of all screening events and outcomes of diagnostic testing
- reporting program data to governance and oversight agencies for monitoring
- establishing and maintaining strong links with the diagnostic laboratories.

¹ 'Provision' of information within this policy framework involves the delivery of information accompanied by an opportunity to discuss the material. See also policy statement 2.3.

² Where diagnostic testing is not completed by the maternity service provider, it is the responsibility of the relevant health care provider to ensure diagnostic testing results are provided to the laboratory.

PART II: CONSENT

2.3 Appropriate information and support are provided to families at all points along the screening pathway

Context

It is essential that families are provided information and are engaged along the screening pathway. This promotes a culture of openness and transparency, and respects the rights of families to be well informed about the care that they receive [6]. Furthermore, it supports informed consent and decision making by families [5, 7]. In the context of newborn bloodspot screening, given that families are likely to be going through an eventful and demanding time, information may need to be communicated more than once and in different ways [8]. Providing individuals who are making a decision about screening with appropriate information and support will promote trust in the program and encourage transparency [9].

Recommendations

- Families are provided relevant information and support at key time points, including:
 - **Prior to birth**
 - At this point, information should be provided to families on newborn bloodspot screening.
 - Any information provided to families should be accompanied by sufficient time for consideration and discussion with other family members and the opportunity to discuss newborn bloodspot screening with a relevant health care provider for clarification, particularly when material is offered in a written format.
 - **After birth, before the bloodspot sample is collected**
 - At this point, the person collecting the sample should check that families have received information on newborn bloodspot screening, have had opportunity for discussion and clarification, and agree to the test.
 - If this has not yet occurred, information on newborn bloodspot screening should be provided at this time to support consent.
 - **If recalled for further testing**
 - At this point, families should be offered additional relevant information.
 - This should include information on relevant support services, such as psychological support.
- Information on newborn bloodspot screening outlines, at a minimum:
 - conditions for which screening is offered, including a very brief description of each condition or group of conditions
 - rationale for screening
 - the heel-prick process
 - what personal information is collected and stored
 - length of retention of dried bloodspots and screening results
 - possible secondary uses of the dried bloodspot
 - how results may be accessed by families.

- Information on newborn bloodspot screening should be:
 - available through a range of methods and media
 - user-friendly and suitable for a range of audiences, including people from culturally and linguistically diverse populations, Aboriginal and Torres Strait Islander families and other vulnerable groups.
- Collaborative efforts between states and territories should occur to work towards national consistency in the information that is provided to families.

2.4 Participation in the programs is voluntary and supported by processes to enable informed consent or refusal

Context

In Australia, families can consent to or refuse newborn bloodspot screening. Consent can be defined as the decision by an individual to undergo a medical intervention [10]. In the context of newborn bloodspot screening, consent is provided by the family on behalf of a baby, who is not yet able to make this decision himself or herself. Consent is grounded in a range of important ethical concepts, which focus on ensuring that families are able to act in line with their own values. In the context of newborn bloodspot screening, this means that families' rights and interests are respected during the decision-making process.

For consent to be valid, the family must be provided with appropriate information and the opportunity to ask questions before deciding whether to participate. Also, the decision maker must have sufficient capacity to make the decision and must make it voluntarily, without undue pressure. Having in place appropriate processes and information to support informed consent or refusal builds trust in the program and demonstrates a commitment to engaging families in decision making. In addition to protecting the newborn and families, adhering to required standards of consent will minimise the potential for any legal liability or professional standards investigations for both health care providers and maternity service providers.

Recommendations

- The decision to consent to screening is to be voluntarily made by the newborn's family, provided that the consenting individual has sufficient capacity to make the decision.
- Where the newborn's family cannot make this decision, relevant legal requirements must be met to obtain consent or a refusal for screening.
- Each program must have a policy outlining its consent process.
- The consent policy should adhere to any legal requirements and outline, at a minimum:
 - the information supporting consent that should be provided to families, both prior to and following birth. This should align with the recommendations regarding the provision of information outlined under policy statement 2.3
 - the need to provide consent after birth and check that the family has received and understood information on newborn bloodspot screening prior to taking the bloodspot sample
 - the written and/or verbal processes involved in consent being obtained
 - how the consent process is documented in medical records, on the baby's personal health record and, where applicable, on the bloodspot card
 - the purpose of consent and whether it includes consent for the sample to be retained for extended periods of time or be used for other purposes, including but not restricted to de identified research (see policy statement 2.8).

- The policy on consent should be readily accessible to health care providers, and information on consent should be provided in a user-friendly way for families and members of the community.
- All families should be encouraged to keep this information for future reference.
- An archive of past policies should be maintained for at least as long as the blood samples collected under them are stored.

2.5 Processes are in place to manage refusal of screening

Context

As newborn bloodspot screening is voluntary, families may refuse screening for their baby. However, given that newborn bloodspot screening is low-risk and may protect the baby from serious, lifelong disability or death, efforts should be made to ensure that a family's consent to screening is facilitated but not coerced [11]. A family's decision to refuse screening must be respected, and processes must be in place to support families and health care providers in the event that screening is refused.

Recommendations

- Each program must have a policy outlining the steps to be taken when a family refuses to consent to screening. The refusal policy should adhere to any legal requirements and outline at a minimum:
 - the need to reiterate the importance of newborn bloodspot screening to families
 - that families be informed of the risks to the baby should he or she have one of the conditions that would be identified through newborn bloodspot screening
 - options for the family to seek further information from an appropriate health care provider on the implications of not screening, to be provided in a timely manner
 - advice to the family that they should take their baby for medical attention if the baby is unwell and advise the health care provider that the baby has not been screened
 - that refusal must be documented in the baby's medical record and any other relevant records. Where possible, the reason(s) for refusal should be stated
 - the need to complete demographic details on the unused bloodspot card and submit it to the laboratory to document the refusal.

PART III: PROGRAM OPERATIONS

2.6 Processes are in place to support bloodspot sampling and management

Context

'Bloodspot sampling and management' refers to the policies and practices that guide the collection of the bloodspot sample and relevant information, and the provision of the dried bloodspot to the laboratory for analysis. These are fundamental steps in the screening pathway and have implications for the time frame for obtaining a result. In line with this, samples should be taken from all babies whose families consent to screening. This should include babies who are stillborn or die shortly after birth, with consent, in line with relevant coroner's regulations. Screening these babies can support completeness of program data and provide valuable information to the family.

Recommendations

- Local policies exist that guide sample collection and management. These policies outline, at a minimum:
 - the sample collection procedure, including that the sample is taken by an appropriately trained health care provider
 - the ideal time frame for collecting the sample, which is forty-eight to seventy-two hours after birth for the conditions currently included in programs. Collection outside of this time frame may restrict the ability to detect conditions
 - steps to minimise pain and distress to the baby and family
 - protocols for screening a baby:
 - who is born at home
 - who is premature, requires intensive care and/or has received transfusions of blood or other blood products
 - who presents after the recommended time frame for screening
 - when the baby and mother are discharged early, including when the family will return to a remote location
 - any arrangements with the coroner's office to support screening of stillborn babies or babies who die soon after birth
 - that appropriately dried bloodspots should be dispatched by the maternity service provider within twenty-four hours of a sample being taken to facilitate receipt of the sample by the laboratory within one to two days
 - processes to ensure maternity service providers know which dried bloodspots have been received by the screening laboratory
 - management approaches for accessioning the dried bloodspot.

2.7 Processes are in place to ensure that families are followed up, in a timely manner, to confirm an abnormal result and access intervention

Context

For the newborn to experience the health benefits of screening, the family must progress along the screening pathway in a timely manner once a sample is taken. This requires processes to be in place that ensure that the sample is analysed, results are issued and the family is followed up to access diagnostic testing and care within the shortest possible time frame. Ideally, these steps should occur prior to any morbidity and mortality associated with a diagnosed condition. A specific time frame for follow-up is not stipulated here, as the time frame in which appropriate follow-up should be achieved differs for each condition.

Recommendations

- Laboratories must have procedures in place to ensure:
 - the sample and information contained on the dried bloodspot cards are processed in a timely manner
 - nominated health care providers are contacted to organise diagnostic testing for babies at increased risk of having a condition
 - diagnostic testing is completed as soon as possible after referral to an appropriate health care provider. For some conditions, this should be on the same day that an abnormal test result is received.
- Laboratories must have policies for managing:
 - follow-up of unsuitable samples and abnormal results, and requests for repeat samples where needed, in a timely manner
 - a lack of response from maternity service providers after requesting repeat samples or other information
 - cases lost to follow-up.
 - Achieving the above will support the relevant health care provider to then:
 - contact the family
 - arrange for diagnostic testing in the time frame appropriate for the condition
 - develop a management plan in case the baby becomes unwell prior to the results of the diagnostic test becoming available
 - provide the results of diagnostic testing to the newborn bloodspot screening laboratory.

2.8 Policies are in place that guide the retention, storage, use of and access to dried bloodspots

Context

The primary purpose of collecting the dried bloodspot is for screening. However, after being analysed for this purpose, the dried bloodspot and relevant participant information are kept by the laboratory for a number of important reasons, which are outlined below. It is essential that programs maintain clear information about why dried bloodspots are retained, how long they are kept for and by whom, the environment in which they are stored and any other ways in which they may be used, and that this information is made available to families.

This is particularly important given that families are providing consent on behalf of the baby. The purpose of the following recommendations is to protect the privacy of the individual from whom the bloodspot sample was taken and maintain the integrity of the programs.

Recommendations

- Information on the use, retention and storage of dried bloodspots is:
 - communicated with families, including at the time of consent (see policy statement 2.3)
 - easily accessible to families, health care providers, laboratory staff and the public.
- A policy exists that outlines:
 - that the primary purpose of collecting a dried bloodspot is for newborn bloodspot screening
 - uses of the dried bloodspot that support this primary purpose, including:
 - subsequent investigation of any initial screening test results that may have been a false positive or false negative
 - quality assurance and audit
 - assay improvement and validation of tests for conditions currently in the program
 - validation of assays for potential new conditions to be included in the newborn bloodspot screening programs
 - post-mortem testing
 - the approved secondary uses of the dried bloodspot, if any, including:
 - research, subject to adherence to the provisions of the *National Statement on Ethical Conduct in Research Involving Humans* [\[12\]](#)
 - forensic/police investigations
 - coronial investigations, subject to appropriate legal processes
 - the length of time the dried bloodspot is retained, noting that the minimum period is two years in accordance with National Pathology Accreditation Advisory Council requirements
 - if and when the dried bloodspot will be destroyed
 - steps to maintain confidentiality of the dried bloodspots
 - relevant laws, customs and guidelines to support ethical management of dried bloodspots and an individual's privacy
 - approvals required to release the dried bloodspot to the individual screened or their family
 - permissions and approvals required for access by others, including managing requests from:
 - researchers
 - physicians
 - coronial or forensic investigators.

Policy Area 3: Quality and safety

The policies contained in this policy area, in combination with those in *Policy Area 4: Monitoring, evaluation and review*, articulate the key aspects of a quality management plan. The information in this policy area outlines policies and recommendations that support the programs to be governed and organised for quality and safety, family-centred and driven by information and evidence [13]. What is outlined, in many instances, provides a framework for what is already occurring and complements existing quality approaches. In line with this, the following policy area should be considered in conjunction with other relevant quality guidance, including that released by the Australian Commission on Safety and Quality in Health Care and the National Pathology Accreditation Advisory Committee.

Maintaining a high level of quality and program safety is the responsibility of health professionals at every level. It is achieved through constant review and improvement of system performance against national standards [14]. It is not a static process; rather, it requires continuous reflection and adjustment at all levels of the program [15]. Following this continuous quality improvement cycle helps to ensure that screening programs maximise the benefits and minimise the harms to the people they screen. Further, to achieve this goal, a program must have in place approaches for looking inwardly at practice and reflecting outwardly to benchmark or learn from others in the field. In line with this, this chapter sets out policies and steps to support these forms of reflection and quality improvement.

PART I: GOVERNED AND ORGANISED FOR QUALITY AND SAFETY

3.1 All programs are supported by clear and effective governance processes and structures

Context

Governance is essential in supporting newborn bloodspot screening programs and enabling them to grow and develop in a safe and effective way [16]. Governance refers to the way in which people and processes are organised to support decision makers in governments and management roles to guide and lead a program [17]. Its core role is to provide a forum in which relevant people are able to inform policy development and program implementation. To do so, and to monitor trends and identify issues, it is necessary for people within governance processes to have access to program data [17]. In the context of screening, it is recommended that clearly defined governance structures exist at the national, state and territory, management and operational levels [5]. Clear and appropriate governance at these levels supports program clarity, accountability, transparency and informed decision making and reduces duplication of effort [18, 19].

Recommendations

- In addition to the roles and responsibilities outlined under policy statement 2.2, programs adhere to the agreed national governance structures and implement local governance arrangements in line with those outlined in Box 3.

- All committees within the governance structure have clearly defined terms of reference that align with the aim and objectives of the programs and include their:
 - purpose
 - deliverables/responsibilities
 - membership
 - reporting lines
 - time frames for meetings, with meetings occurring on a regular basis.

BOX 3: PROGRAM GOVERNANCE

National governance includes:

- an inter-jurisdictional committee that represents health departments, ideally SCoS, which provides national policy direction and supports program monitoring and decision making
- where necessary, under the remit of SCoS and in line with the decision-making pathway in Policy Area 5: Decision-making process, a time-limited working group to support assessment of conditions
- a separate inter-jurisdictional 'program management committee', consisting of, at a minimum, program management representatives from each state and territory, which supports the consideration of operational issues, information sharing and forward planning.

State and territory governance includes a committee that provides advice and recommendations to health departments on, at a minimum:

- local policy development
- operational issues
- monitoring data and trends, locally and with reference to other programs
- opportunities and strategies for quality improvement
- horizon scanning
- risk management and contingency planning
- identifying issues of national relevance.

The state and territory governance committee should have a multidisciplinary membership that includes consumer representation and other stakeholders involved in the screening pathway in both the private and public sectors, being those individuals with skills and expertise in:

- newborn bloodspot screening program management
- the scientific aspects of newborn bloodspot screening
- government representation
- public health
- maternity services
- midwifery and neonatal nursing
- medical practice (such as paediatricians, neonatologists and obstetricians).

Where dried bloodspots are analysed for another state or territory, the committee should support links between those jurisdictions.

3.2 Health professionals who support and deliver newborn bloodspot screening take action to promote quality and safety

Context

A high-quality and safe screening program is dependent upon the people involved in the programs having the skills and support to undertake their roles. To maintain skills, and remain aware of developments in the evidence on which screening and follow-up care are based, it is essential that staff are continually learning in their roles. This relies on, but is not limited to, people supporting the program taking active steps to continually improve their individual and collective performance [\[20\]](#); and managers and leaders supporting the development of their colleagues [\[21\]](#).

Recommendations

- All professionals delivering or supporting newborn bloodspot screening programs hold relevant skills, expertise, training and certifications.
- To support continuous improvement and development, staff are supported by, at a minimum:
 - regular reviews
 - ongoing training and professional development opportunities
 - monitoring against performance measures [\[21\]](#).
- Underperformance is managed promptly and in line with local policies and processes.
- Maternity service providers, laboratory staff and other health professionals take steps to improve program-wide quality when issues are identified.

3.3 Facilities, equipment and work processes are designed for quality and safety

Context

Newborn bloodspot screening programs are multidisciplinary and are delivered and supported by a range of health care agencies, including the places where samples are taken, the laboratories and the places that provide follow-up care. It also includes the government health departments that support the programs. The current technologies, and those potentially on the horizon, are complex, and related work processes are highly specialised [\[22\]](#). Therefore, it is essential that relevant facilities, equipment and work processes are planned, organised and managed to ensure high-quality screening.

Recommendations

- All elements of a newborn bloodspot screening program meet relevant accreditation requirements, including laboratory standards set by the National Pathology Accreditation Advisory Committee.
- New technologies are rigorously assessed before being introduced.
- Programs are supported by effective information systems, including, at a minimum, a database to capture, analyse and report program data.
- Processes are in place to support partnerships and information sharing between health professionals.

- Work processes are in place to ensure the screening pathway—including communication of information to families; collection of the sample; delivery of the sample to the laboratory; analysis of the sample; reporting of results back to the health care provider, maternity service provider and, if appropriate, the family; and provision of diagnostic test results back to the laboratory where required—is completed in a timely fashion.
- Special efforts are made to ensure quality and safety of the program in rural and remote regions.

3.4 Programs take action to extend excellence, and prevent and minimise harm

Context

Within any health care setting, there is the potential to cause harm. This can include physical harm resulting from a test or procedure and psychological harms due to receiving an abnormal or incorrect result [23]. These harms might include a one-off significant event or repeated smaller events [24, 25]. Screening programs should only be implemented if it is clear that these harms are able to be appropriately minimised and are outweighed by the benefits that can be delivered to individuals and the population [5].

There are currently two approaches to ensure that the benefits of a program outweigh any potential harms. These include an ‘appreciative approach’, whereby excellence is identified and extended into other areas of a program [26, 27]; and a ‘harm minimisation approach’, in which potential harms are actively identified and prevention strategies put in place [28]. Despite these approaches, as with all health care procedures, harms will occur in a small number of cases [29]. It is essential that programs learn from these occasions, provide appropriate support to affected families and put in place further steps to reduce the occurrence of such harms in the future [29].

Recommendations

- Processes are in place to identify, share and encourage excellence across all aspects of the program.
- Risk management plans are developed and regularly reviewed and mitigation strategies put in place where necessary.
- Relevant guidelines are promoted and followed to reduce inappropriate variation.
- Policies exist to manage:
 - complaints
 - missed cases
 - serious incidents and harms.
- These policies should align with local adverse event or critical incident reporting processes, build upon the Australian Open Disclosure Framework [29], and include an outline of:
 - what constitutes a serious incident or harm
 - the importance of having an open disclosure approach
 - staff roles in reporting a serious incident or harm
 - the requirement for:
 - the family to be informed
 - the issue to be reported, including what information should be reported and to whom
 - steps to be put in place to resolve the issue in a timely fashion
 - monitoring to ensure the issue is resolved
 - approaches for supporting families to report serious incidents/harms.

3.5 The programs, including all elements of the screening pathway, are adequately funded and resourced

Context

Newborn bloodspot screening is understood to be a cost-effective approach to minimising the burden of disease on the population [30, 31]. To achieve such cost-effectiveness, investment is required. The programs must be adequately funded and resourced for the work that they do [5, 13]. This resourcing is required across the screening pathway to support aspects such as midwives and nurses providing information to families and collecting the samples; laboratories undertaking the analyses, developing information and supporting the program; and governance and oversight, including within health departments.

Recommendation

Programs are resourced appropriately at all levels to achieve the aim and objectives of the programs and meet the recommendations within this policy framework.

PART II: FAMILY-CENTRED

3.6 Programs are delivered using family-centred approaches

Context

People-centred approaches are recommended throughout health care [13]. In the context of newborn bloodspot screening, these are referred to as ‘family-centred approaches’. A cornerstone of such an approach is being respectful of, and responsive to, the values and preferences of families that are offered screening and participating in the program [32]. Further, a family-centred approach means that consumers, who can provide a family perspective, are involved in program and policy development and health care design. Engaging families in these ways builds trust and influences their satisfaction with the program [33]. Across health care more broadly, it can also lead to better health outcomes as well as improve quality and safety and cost-effectiveness [7, 34–37].

Recommendations

- Consumers, who can provide a family perspective, are engaged in program planning and policy development.
- Information on the programs is family-focused, in line with policy statement 2.3, to enhance a family’s understanding of newborn bloodspot screening and their health literacy more broadly.
- Families’ rights are respected and their engagement in decision making is supported.
- Information on families’ level of satisfaction with the program is sought at key points in time.
- Screening and care is provided that respects and is sensitive to different cultures [13].

3.7 An 'open culture' exists for complaint and incident reporting

Context

This policy statement focuses on the family and their experience, in the rare instance where the screening process results in serious harm. It complements the information at policy statement 3.4 on minimising harm and takes a family-centred approach to complaints management and serious incident or harm reporting. The underlying principle of this policy statement is that, if something goes wrong, families are informed and supported through the resolution process. Similarly, if families wish to make a complaint, or report a serious incident or harm, they are able to do so in a way that best meets their needs [29].

Recommendations

- Information on complaints reporting and management, and the serious incident or harm reporting mechanism, is family-friendly and easily accessible.
- Health care providers and laboratory staff involved in newborn bloodspot screening support families who wish to make a complaint or report an incident.
- Families are made aware of other resources available to them to support them through and after the complaint or incident process.

PART III: DRIVEN BY INFORMATION AND EVIDENCE

3.8 Program information is used to support further development in newborn bloodspot screening

Context

The information and evidence generated by the programs, and how they are used, are important factors in a high-quality screening program [5, 13, 38]. As outlined in *Policy Area 4: Monitoring, evaluation and review*, programs should be continually assessing their performance, including against other programs in Australia and, where appropriate, internationally. In addition, programs should share their experiences and contribute to relevant research. Following these steps will mean that the programs' future developments will be driven by local and international information and evidence.

Recommendations

- The policy statements in this policy area, which relate to quality and safety, are informed by program data.
- Appropriate data on performance should be provided to maternity service providers to support process improvement.
- Coordinated opportunities exist for shared learning between state and territory programs.
- Processes are in place to support the regular review of the latest available evidence of benefits and harms of newborn bloodspot screening.

- Programs support relevant research and development through having consistent policies and processes in place that:
 - govern access to data for research purposes
 - encourage research partnerships between laboratories, health care providers, research agencies and others involved in the programs
 - support horizon scanning to identify key issues facing the future of newborn bloodspot screening
 - highlight strategic research priorities.
- Programs strive to innovate and have processes in place to identify and assess new ideas, integrate them into practice within their own program and assist with integration into other state and territory programs where appropriate.
- Where innovation approaches and changes are implemented that affect the delivery of the program, they must be accompanied by an appropriate communication strategy.

Policy Area 4: Monitoring, evaluation and review

The information within this policy area is essential to ensuring screening programs are high-quality. Specifically, the following content outlines high-level policies to support national, state and territory, and operational level monitoring, evaluation and review. It articulates the need for indicators, which enable assessment of how well the programs meet their aim and objectives. It also outlines the need for a structured approach to ensure data are collected, reported and then used to inform program development. The data generated through monitoring, evaluation and review should be used to inform the steps articulated in *Policy Area 3: Quality and safety*; therefore, the two chapters should be read in parallel.

Assessment of a program's performance is essential for a number of reasons. First, governments and programs have an obligation to the individuals screened, and their families, to ensure that the programs continue to maximise benefits and minimise harms [5]. Second, governments require a program to remain effective and efficient. This requires continual assessment, as the factors that influence the viability of a program change over time. For example, the understanding of a condition can change, and new, more effective, interventions may emerge. Third, performance assessment is an essential part of the quality improvement process and therefore supports a high quality program [39].

Performance assessment is most effective when it occurs at all levels of a program [5]. In the context of newborn bloodspot screening, national monitoring provides a collective picture of the programs. It enables benchmarking and a framework within which programs can share successes and identify and manage areas of underperformance. Program-level assessment of performance is essential so that individual state and territory governments can assess the extent to which they are meeting their commitment to the public and the programs are delivering value for money. Operational-level assessment is essential to monitor and manage the functional aspects of a program. Across each of these levels, to gain an accurate understanding of the programs, it is necessary to consider trends to assess their performance over time [40].

PART I: MONITORING, EVALUATION AND REVIEW

4.1 Formal approaches exist for ongoing monitoring, evaluation and review of newborn bloodspot screening programs

Context

Monitoring, evaluation and review are essential components of a quality screening program [5]. They ensure program transparency and accountability and enable identification of program successes and areas requiring further development. In the context of this policy framework, monitoring refers to the systematic collection, assessment and reporting of data to consider performance against program objectives; evaluation refers to a more comprehensive program-wide review in response to a specific issue or set of circumstances; and review refers to a smaller one-off investigation or consideration of a specific program area or issue. All components of monitoring, evaluation and review are both retrospective, looking back at program performance; and prospective, using program data to inform development.

Recommendation

- Formal approaches exist at the operational, state and national levels to facilitate ongoing monitoring, evaluation and review that are clearly articulated; are supported by agreed policies; and outline steps for quality and safety in line with *Policy Area 3: Quality and safety*. These approaches should align with the following:
 - Program monitoring occurs through analyses of data against agreed program indicators. Monitoring outcomes are reported to program decision makers and relevant stakeholders in line with agreed time frames.
 - Collaborative efforts are made to define and endorse national indicators and data elements to support national monitoring.
 - Evaluation and review occur in line with agreed time frames or circumstances that would necessitate program evaluation or reorientation, or investigation of a specific issue.
 - Evaluation occurs in line with an appropriate plan and includes consumer and stakeholder engagement.
- Short- and long-term outcomes are monitored, evaluated and reviewed.

PART II: PROGRAM INDICATORS

4.2 Appropriate and measurable indicators exist against which data are collected to monitor the newborn bloodspot screening programs

Context

Indicators are measures of performance that enable assessment against the intended objectives of a program. Screening programs must have clear, measurable indicators against which they are monitored and evaluated [5]. These indicators must enable short- and long-term assessment against the programs' aim and objectives and support consideration of a program's performance across the screening pathway. Reporting data against agreed indicators enables monitoring at a point in time and consideration of a program's performance over time. Where indicators are the same across programs, they enable national and international benchmarking. As such, appropriate and measurable national indicators are an essential component of transparent and high-quality newborn bloodspot screening.

Recommendation

- Newborn bloodspot screening indicators are in place to enable monitoring of the programs against their aim and objectives.
- Where possible, programs across Australia should collect data against the same indicators. To assist this, national indicators and data elements should be agreed and defined, ideally through a national data dictionary.
- Indicators should enable comparison over years, to consider trends, and between local and international programs where appropriate.
- Indicators must enable assessment of the programs' success, including their impact upon morbidity and mortality. This should include the measurement of positive (abnormal) diagnosis rates and false positive rates.

- Key areas of importance for developing indicators include:
 - participation/coverage, including of groups at risk of being under-screened
 - timeliness of sampling and testing
 - unsuitable samples
 - short- and long-term program performance
 - diagnostic testing and follow-up care
 - harms and adverse events
 - family satisfaction
 - other aspects of screening performance.
- Examples of the types of indicators that could be used to monitor newborn bloodspot screening can be found within the Human Genetics Society of Australasia's and the Royal Australasian College of Physicians' joint Newborn Bloodspot Testing Policy [\[41\]](#).
- Program indicators are used for program monitoring, evaluation and review. These activities should also be supported by accessing other sources of data and specific projects.

PART III: DATA COLLECTION AND ANALYSIS

4.3 Sustainable and appropriate methods exist for collecting and analysing data against the indicators and supporting program operations

Context

Newborn bloodspot screening spans a range of different health care areas; therefore, data to support monitoring, evaluation and review will be collected by different areas of the health system. While a large proportion of the data will be collected by laboratories, data will also be collected by midwifery service providers, general practitioners, hospitals and private specialist practices. It is therefore necessary to have data information systems and data collection approaches that span across the different settings and ensure accountability for data collection is attributed to the correct stakeholder. An information system refers to the people, technology and processes that collect and analyse data and information [\[42\]](#). Effective information systems are supported by clear policies that enable collection, storage and reporting of complete program-wide data.

Recommendations

- Information systems exist to capture and maintain appropriate data that meet monitoring, evaluation and review requirements and support local operations. This includes capturing data from the laboratories and multiple sources across the health system.
- There are agreed roles and responsibilities to support the information systems. These should define the data collection responsibilities of the different stakeholders that are required to support completeness of program data.
- Local policies and processes are in place to ensure completeness and accuracy of data that flow through the relevant information systems.

- Data are managed securely within the information systems, and policies exist that outline:
 - the steps taken to ensure privacy of data
 - who has access to the data and for what purposes
 - approval processes to access data
 - relevant legislation by which the programs are governed, and which ensure security of the data.
- Data are collected and analysed to support local and national reporting.

PART IV: REPORTING

4.4 Program monitoring data are reported to key stakeholders, including governments and the public, at regular intervals

Context

‘Reporting’ refers to the ways in which data are communicated to the broad range of stakeholders involved in newborn bloodspot screening [43]. This includes data that have been collected against indicators for monitoring, evaluation and review purposes. To align with the continuous quality improvement cycle, program data must be reported to the relevant individuals and governance bodies to ensure successes are shared and any issues are managed promptly. Further, to support transparency and accountability, it is essential that program information is made available to the stakeholders involved in, and affected by, newborn bloodspot screening in a way that is meaningful to them.

Recommendations

- State and territory processes support program reporting to, at a minimum, the public, state and territory health departments, and to national governance groups, in line with the governance arrangements outlined in policy statement 3.1.
- Processes exist to support the timely reporting of the findings of monitoring, evaluation and reviews to relevant stakeholders. These processes should support a swift response to any significant issues identified through monitoring, evaluation and review, and they should promote excellence.
- Public reporting should be done in a way that ensures that the information is both easily accessible and accessible in a way that is meaningful for target audiences.

Policy Area 5: Decision-making process

The purpose of this chapter is to provide a national decision-making process for assessing conditions for inclusion in, or removal from, newborn bloodspot screening programs. This provides a single mechanism for assessment, to which states and territories can contribute. The outcome of this assessment can then be used to inform all states' and territories' decision-making processes regarding the conditions included in their newborn bloodspot screening programs, thereby avoiding duplication of effort. It also supports a continued high level of consistency for conditions screened. A robust decision-making process ensures that a decision to add or remove a condition is only ever reached after careful assessment of the evidence and minimises the extent to which a program can be inappropriately influenced by external pressures [1, 44, 45]. Further, having a clearly articulated process means that the programs are transparent and accountable, and each condition is assessed in the same way. Ultimately, this decision-making process is a key mechanism to support continued success and safeguard the programs into the future.

PART I: DECISION-MAKING PATHWAY

5.1 Decisions to add or remove conditions from newborn bloodspot screening programs must be made in line with a nationally agreed decision-making pathway

Context

International examples of newborn bloodspot screening programs demonstrate that having a cohesive national process for managing the addition or removal of conditions supports an evidence informed approach and promotes consistency and transparency across jurisdictions [46–48]. In line with this, the following recommendations provide a pathway by which conditions can be assessed for inclusion in, or removal from, Australian newborn bloodspot screening programs.

The following pathway aligns with the governance structure outlined in *Policy Area 3: Quality and safety*. In doing so, it draws upon the wealth of knowledge and experience that currently supports newborn bloodspot screening in Australia as well as the expertise of SCoS. Further, in line with best-practice policy development, the following pathway provides opportunities to engage the appropriate representatives and experts, including families, to inform decision making for newborn bloodspot screening.

Recommendation

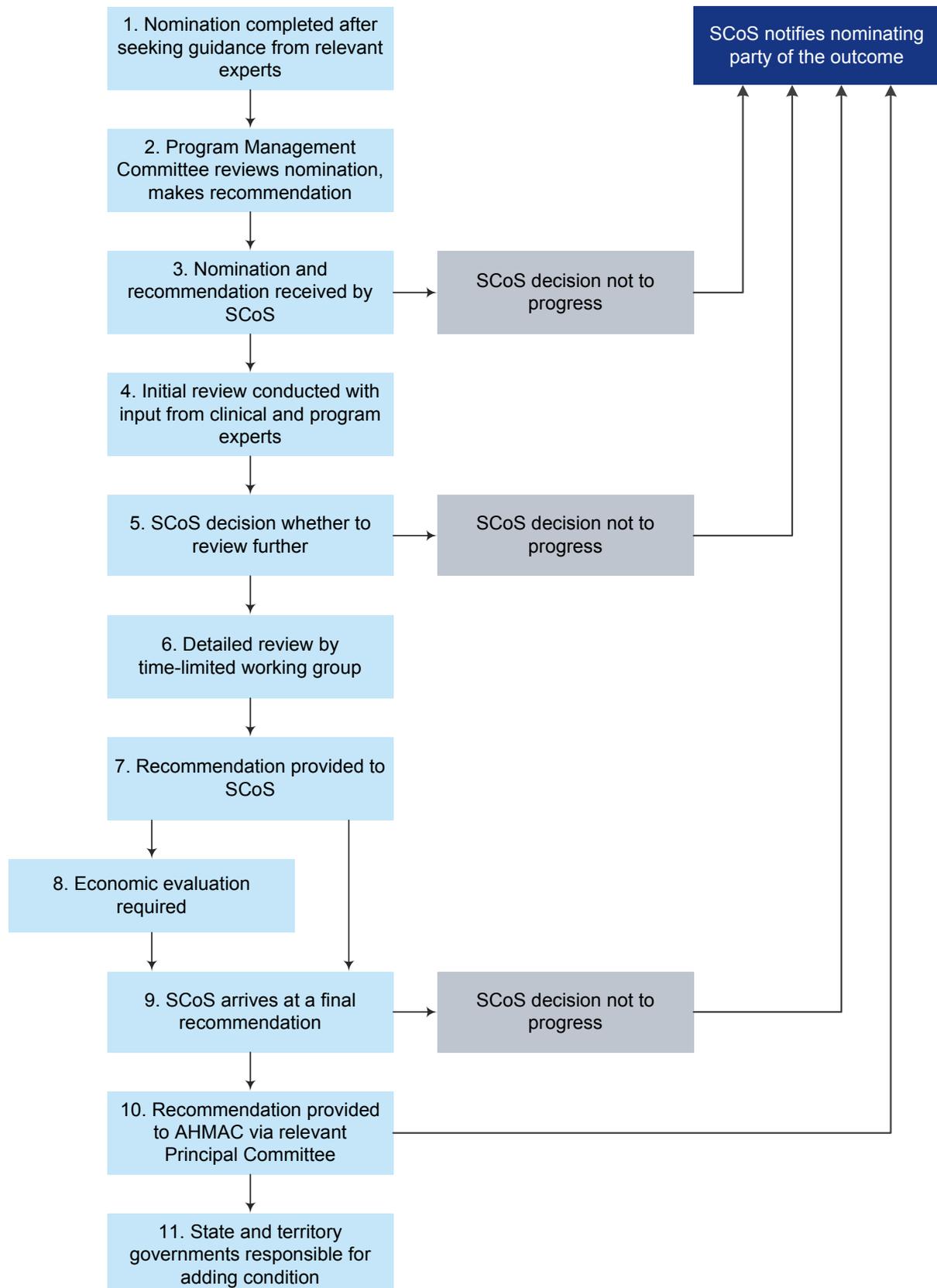
- When a condition is nominated for inclusion in the screening programs, it is to be assessed in line with the following pathway (illustrated in Figure 1):
 1. The condition can be nominated by anyone in Australia by completing the nomination form at Appendix C. It is recommended that a nominee who is not from a newborn bloodspot screening program seeks the advice and guidance of their jurisdiction's newborn bloodspot screening program regarding the required documentation and evidence in order to make a submission for the addition or removal of a condition.
 2. When the nomination form is completed, the submission is forwarded to an inter-jurisdictional Program Management Committee in line with the governance structure outlined at policy statement 3.1. This group considers the nomination and provides a recommendation to SCoS as to whether further assessment of the condition is merited.

3. The nomination form and associated Program Management Committee recommendation are received by SCoS.
 4. If SCoS agrees with a recommendation for further assessment, an initial review is conducted. This review should be informed by relevant clinical and newborn bloodspot screening expertise, either through the standard SCoS briefing process or by seeking further input from the Program Management Committee. This initial review will:
 - a. provide a preliminary assessment of the feasibility/desirability of screening for the identified condition
 - b. be primarily based on the information provided within the nomination form
 - c. determine whether a more detailed review is warranted.
 5. At the completion of the initial review, SCoS determines whether the condition should be reviewed in more detail.
 6. At the request of SCoS, a detailed review of the evidence of screening a condition is conducted against the agreed decision-making criteria (see Part II: Decision-making criteria within this policy area). This assessment should be undertaken by a time-limited working group established by SCoS. This group should be comprised of relevant experts, including but not limited to experts in the condition being assessed and newborn bloodspot screening.
 7. At the completion of the detailed review, a recommendation is made to SCoS as to whether:
 - a. an economic analysis is warranted, and/or
 - b. based on the information assessed, the condition should be added to the programs, in line with the possible recommendations outlined in Box 4.
 8. If deemed necessary by SCoS, an economic analysis is conducted.
 9. SCoS then arrives at a final recommendation from those options outlined in Box 4.
 10. If SCoS recommends screening for a particular condition, the relevant recommendation, accompanied by preliminary cost implications where necessary, will be submitted to the Australian Health Ministers' Advisory Council (AHMAC) for consideration, via the relevant Principal Committee.
 11. If the recommendation is supported by AHMAC, state and territory governments are then responsible for funding and establishing any required governance around adding conditions, taking into account local contexts.
- A condition can be nominated for removal from the programs using the form at [Appendix D](#). To assess removing the condition, the pathway outlined above can be followed. Progression along the pathway may be expedited where appropriate, at the discretion of SCoS.
 - The nominating party should be notified of the outcome of the nomination, including any decision to not review or assess the condition, or the outcome of assessment. Communication with the nominating party is the responsibility of SCoS.

BOX 4: RECOMMENDATIONS THAT CAN BE MADE FOLLOWING ASSESSMENT OF THE EVIDENCE FOR SCREENING A CONDITION

- 1. When considering including a condition in newborn bloodspot screening,** possible recommendations include:
 - Screening is recommended.
 - A pilot is recommended and specific issues flagged for investigation.
 - Based on the current evidence and understanding of a condition, screening is not recommended at this time. However, there may be merit in revisiting this condition in the future if further evidence emerges.
 - Screening is not recommended.
- 2. When considering removing a condition currently screened,** possible recommendations include:
 - Continue screening.
 - Cease screening.

Figure 1: Decision-making pathway for newborn bloodspot screening in Australia



PART II: DECISION-MAKING CRITERIA

5.2 Decisions to add or remove conditions from newborn bloodspot screening programs must be made in line with agreed criteria

Context

There are a number of fundamental principles that support the provision of any screening program to the target population [5]. These principles ensure that screening is provided in line with the latest evidence, guidance and clinical understanding of the conditions. General principles for screening include the following:

- Screening aims to benefit its target population through the early diagnosis of a condition. This then enables early management, generally leading to better health outcomes.
- While there are many benefits to screening, there can also be harm associated with such tests or the subsequent health interventions. Harms may include false positives or false negatives; over-diagnosis; over-treatment; and the emotional impact to the individual and their family that may occur when they receive an abnormal result and then a diagnosis.
- There is a need to ensure that the benefits of screening outweigh any potential harms. In Australia, screening programs endorsed by governments adhere to the Australian *Population Based Screening Framework*. This document enables consideration of the benefits and harms of screening.
- The Australian *Population Based Screening Framework* includes guidance to ensure that screening is provided only for those conditions where there is sufficient evidence to support screening and the benefits outweigh the harms. This guidance is based on the World Health Organization's (WHO) screening principles. These principles are internationally recognised as supporting safe, effective and cost-effective screening programs. These principles are included at [Appendix E \[49\]](#).
- The *Population Based Screening Framework* also includes the principles for implementing and managing screening programs in Australia, as agreed by all Australian governments.

In line with the above general principles for screening, newborn bloodspot screening is provided where:

- there is benefit to the baby from early diagnosis of conditions screened
- the benefit is reasonably balanced against any harms and costs
- there is a reliable test suitable for newborn bloodspot screening
- there is a satisfactory system in place to deal with diagnostic testing and follow-up care of babies with abnormal screening results.

The following decision-making criteria draw upon the above principles to provide a comprehensive set of criteria for assessment relevant to the Australian setting of newborn bloodspot screening.

Recommendation

- The purpose of the criteria, and their sub-points, is to provide a guide for those considering the appropriateness of a condition for inclusion in or removal from newborn bloodspot screening programs.
- All the criteria and sub-points must be considered when assessing a condition. The sub-points are not intended to be 'tick-box' type questions, where a certain number must be met for screening to be recommended. Instead, they provide prompting questions to generate discussion and debate on the full range of issues to be considered when assessing a condition. This aims to support a balanced and considered assessment of the relevant benefits and harms of screening for a condition.

- In the context of newborn bloodspot screening, there are likely to be differing levels of evidence available to assess the criteria and sub-points. Therefore, the best available evidence and information should be drawn upon to explore the following criteria. This should include, where available:
 - high-quality studies
 - international experiences
 - program and condition expertise
 - other relevant sources of information and evidence.

DECISION-MAKING CRITERIA

The condition

1. The condition should be a serious health problem that leads to significant morbidity or mortality.
 - 1.1 What data are there on the incidence of the condition, including in the Australian population? How is this incidence determined—through screening studies, international programs, cases identified clinically, modelled estimates based on data from variant databases or some other means? Are there any known differences in incidence in Australian sub-populations?
 - 1.2 What is the burden of disease associated with the condition, including morbidity and mortality? Does the burden of disease vary between individuals?
2. There should be a benefit to conducting screening in the newborn period.

While the benefit to the baby must always be the first consideration, for some conditions a benefit for the family and/or community, as well as the benefit to the baby, may also be important and warrant consideration. This might include benefits to the family for conditions where there is currently no intervention and which will be likely to lead to early mortality but where a definitive diagnosis might be aided by a screening test.

 - 2.1 What are the known health benefits from early detection that exist, or can be achieved, through screening for the condition? This may include early intervention, prevention of symptoms or reduction in condition severity.
 - 2.2 Why is screening for this condition during the newborn period the most beneficial method of early detection?
 - 2.3 Does detection of this condition provide families with actionable information that assists them in making informed choices about reproduction in the future?
 - 2.4 What emotional or social benefits does early detection provide?
 - 2.5 What harms may arise from screening for the condition in the newborn period?
3. The natural history of the condition, including development from latent to declared disease, should be adequately understood.
 - 3.1 What information is known on the natural history of the condition in Australia or comparable international populations?
 - 3.2 When would the condition usually be detected clinically?
 - 3.3 Explore the current knowledge of penetrance of the condition. Are there known benign or milder late-onset forms?

The screening test

4. There should be a suitable test protocol to identify the presence of the condition.
 - 4.1 What test protocols could be used to identify the presence of the condition? Is there consensus on the most appropriate test protocol?
 - 4.2 When considering the test protocol, what is the clinical and analytic validity based on a consideration of:
 - sensitivity
 - specificity
 - false positive rate
 - false negative rate
 - positive predictive value
 - negative predictive value?
 - 4.3 Is the test protocol simple and reliable?
 - 4.4 Can the test protocol be performed on the available dried bloodspot?
 - 4.5 Can the test be multiplexed within existing newborn bloodspot screening panels?
 - 4.6 What is the cost of the test protocol?
 - 4.7 Will genetic testing be used as part of the test protocol? If genetic testing is needed:
 - Will this be by common mutations or sequencing?
 - Which mutations would be tested?
 - What is the penetrance of the mutations?
 - Are there variants of unknown significance?
5. The test protocol should, on balance, be socially and ethically acceptable to health professionals and the public.
 - 5.1 Can the test protocol detect other conditions of clinical or unknown significance and/or carriers and, if so, what are the implications?
 - 5.2 What are the potential benefits and harms associated with the preferred test protocol(s)?

The intervention

6. Health care services for diagnosis and management should be available so that these services can be offered if there is an abnormal screening result.
 - 6.1 What health care services are currently involved in the diagnosis and ongoing management of the condition?
 - 6.2 What impact would screening for the condition have on the health care services that would be required to support diagnosis and management following an abnormal screening result?
 - 6.3 Is diagnostic testing readily available and reliable?
 - 6.4 Do current health care services have capacity to support the diagnosis and ongoing management of the condition?
 - 6.5 Are current health care services of sufficient quality to support the diagnosis and ongoing management of this condition?
 - 6.6 Is there equitable access to these health care services for families, including those from rural and remote areas?
7. There should be an accepted intervention for those diagnosed with the condition.
 - 7.1 What accepted intervention(s) is (are) available for newborns that receive an early diagnosis through screening?
 - 7.2 How well is the intervention and treatment pathway understood? Is there agreement on when intervention is required?
 - 7.3 How effective is the intervention? Does it alleviate the symptoms of the condition or slow or halt its progression? What influence does the intervention have on quality and length of life?
 - 7.4 How urgent is the intervention? Does the intervention need to be initiated before symptoms of the condition present?
 - 7.5 Is the intervention readily available and accessible?
 - 7.6 What are the potential harms associated with the intervention, and to what extent can these harms be mitigated or managed?
 - 7.7 What is the cost of the intervention? What costs will be incurred for the diagnosis, management and treatment of conditions, including the costs for false positives?
 - 7.8 Is there equitable access to the intervention for families, including those from rural and remote areas?

Additional considerations

8. The benefit of screening a condition must be weighed against its impact on the program as a whole.
 - 8.1 Can screening for this condition be achieved within the current screening pathway?
 - 8.2 Is the addition of this condition likely to require ethical considerations that may warrant a separate consent process?
 - 8.3 Would it be likely that screening for the condition would impact negatively upon other elements of the program? For example, could it be anticipated that participation rates might fall?
 - 8.4 Are there any additional costs, such as the purchasing of new technology or training, which are associated with screening for this condition?
 - 8.5 What is the economic impact of excluding/including the condition? Do benefits exceed costs? Is it cost-effective to screen? It may be necessary for a detailed economic evaluation to consider these questions and other relevant economic issues.
9. What other information relevant to decision making should be considered that has not been captured elsewhere?

Glossary

Please note that the terms contained in this glossary have been defined as they pertain to this policy framework and newborn bloodspot screening. As such, many of the terms may be applicable only in this context.

Abnormal result	A result of the screening test that indicates that the newborn is at increased risk of having a tested condition.
Accessioning	The receipt, register and coding of the dried bloodspot, which is done when it is received by the laboratory for testing.
Accountability	The obligation of an individual or organisation to give account of, acknowledge and accept responsibility for decisions, actions and policies and to disclose the results of these in a transparent way.
Adverse event	Any unintentional harm or suffering that arises as a result of participating in screening that is unrelated to an underlying condition of the individual being tested.
Analytic validity	How reliably the screening test is able to measure the marker analyte used to identify those at risk of a condition.
Assay	A laboratory procedure that is used to identify and measure a marker analyte within the dried bloodspot sample.
Benign	Having no harmful effects.
Bloodspot card	The piece of filter paper on which the bloodspot sample is collected and identifying information about the baby being screened is entered.
Bloodspot sample	The blood that is taken from the heel prick of a newborn.
Bloodspot sampling	The process of taking blood from a newborn by pricking their heel, placing one or more spots of that blood on a piece of filter paper and then allowing it to air dry. This creates the dried bloodspot.
Burden of disease	The impact of a condition on an individual, a population and/or a health system, which can be measured by indicators such as financial cost, morbidity, mortality and years of healthy life lost due to having the condition.
Clinical guidelines	Statements and recommendations about health care for a specific condition and/or clinical circumstance, which are intended to help health care providers and patients make decisions that will optimise patient care. Clinical guidelines are informed by a systemic review of evidence and can relate to diagnosis, treatment and/or management of a condition.
Clinical significance	Where the test result can lead to important changes in the clinical status of the person tested, such as giving them interventions they might not have received without the test results.
Clinical validity	How accurately the screening test predicts the presence, absence or risk of a condition. It is based primarily on the test's sensitivity and specificity.

Common mutations	Alterations in the DNA sequence, such that the genetic message carried by a gene(s) differs from what is found in most people. A mutation is considered common if it occurs in more than 1 per cent of the population.
Condition	A disease, disorder, syndrome, illness, abnormality or health problem that affects a person's health status, wellbeing or quality of life. There may be different forms of a condition, which can have varying impacts. To be included in newborn bloodspot screening programs, a condition or forms of a condition must be serious and lead to significant morbidity or mortality.
Consent	The decision by an individual to undergo a medical intervention. Consent to participate in newborn bloodspot screening is provided by the family on behalf of a baby.
Consumers	Members of the public, including families that consent to or refuse screening, as well as those individuals and families with no involvement with the programs.
Continuous quality improvement	A constant process of reflection and readjustment of performance that involves planning, implementing, reviewing and improving safety and quality.
Criteria	Rules, principles or standards that are used as reasons for making a judgement or decision.
Data dictionary	A document that defines the data elements that should be collected within a given database. A data dictionary can describe the fields that should be collected within the database, their specifications, and how the elements combine to enable assessment against an indicator.
Diagnosis	The confirmation of the presence of a condition, which includes evaluating observed signs and symptoms.
Diagnostic test/testing	A test that confirms whether a person does or does not have a condition.
Dried bloodspot	Dried spots of blood from a heel-prick of a newborn. In the context of this framework, it is also taken to include any relevant information on the bloodspot card.
Eligible population	The group of people who meet the criteria for being able to participate in a screening program.
Equity	Fair access to newborn bloodspot screening programs and to the health outcomes that result from participating in these programs.
Evaluation	A comprehensive program-wide assessment of performance in response to a specific issue or set of circumstances.
Evidence-informed	Guided by the best evidence available, including information from research and experience.
Facilities	See <i>Health care facilities</i> .
False negative	A test result that incorrectly indicates a person does not have a condition when they do have that condition.

False positive	A test result that incorrectly indicates a person has a condition when they do not have that condition.
Family	The adult(s) with legal responsibility for the newborn screened through newborn bloodspot screening. This includes parents, guardians, carers and care-givers (where appropriate).
Family-centred	Being respectful of, and responsive to, the values and preferences of families that are offered screening and participate in the program. It also involves including families in the planning and delivery of health systems and services, including program and policy development, quality improvement, patient safety initiatives and health care design.
Follow-up care	The steps that are taken after an abnormal result from the newborn bloodspot screening test to enable the diagnosis, treatment and/or management of the condition as well as provide support to those who may be suffering from physical, psychological or social harm resulting from screening.
Gene	The basic physical and functional unit of heredity contained within the DNA code. Genes code for molecules that influence the expression of a person's characteristics. A faulty gene or set of genes can potentially result in a person having a condition.
Genetic testing	The study of a person's DNA in order to identify any differences that may indicate the genetic cause of, or susceptibility to, a condition.
Governance	The way in which people and processes are organised to support governments and decision makers to steer a program.
Governance structure	See <i>Governance</i> .
Harm	An undesired outcome or disadvantage—in this case, from participating in a screening program. Harms might include a one-off significant event or smaller events which occur over time. They can include physical, psychological and social harms as well as outcomes such as over-diagnosis and over-treatment. Examples include injury from the heel-prick test, stress and anxiety and any feelings of stigmatisation resulting from being diagnosed with a condition.
Health care agencies	Organisations involved in health care. For newborn bloodspot screening, this includes the places where bloodspot samples are taken, the laboratories and the places where specialist care is provided. It also includes the health departments that support the programs.
Health care facilities	Premises that provide health care services. This includes hospitals, outpatient clinics and specialist centres such as birthing centres.
Health care provider	A qualified person or organisation that provides health care services. Examples include primary care (e.g. general practitioners), nursing and midwifery care, and follow-up care from doctors and counsellors.
Health care services	Services provided by Australia's health care system, in both public and private sectors. This includes the health professionals, health care facilities and equipment involved in delivering the services.

Health outcome	The change in the health status of a person, group of people or population, which occurs because of undergoing an intervention for a condition.
Health professional	Anyone working within the health care system, including health care providers, laboratory staff, scientists, managers, decision makers and policy makers.
Horizon scanning	A process of collecting and analysing information to detect emerging, continuing or potential threats, opportunities, problems, challenges, trends and other likely future developments that are relevant to an organisation.
Incidence	The occurrence of new cases of a condition in a population within a specified time period (e.g. one year).
Indicator	A measure of performance that enables an assessment of whether a program's aim and objectives are being achieved.
Information system	The people, information technology and processes that collect and analyse data and information.
Informed consent	A voluntary decision to accept or decline a screening test, which is underpinned by sufficient information, including benefits and risks, to support an appropriately informed decision.
Intervention	An action that is designed to change the outcome or course of a condition, such as a treatment or option to help manage the symptoms.
Jurisdiction	A geographic area with a set of laws which are different from neighbouring areas. In Australia, the Commonwealth and each state and territory are separate jurisdictions.
Laboratory staff	Refers to the personnel who work in and manage the laboratories that test the dried bloodspot, including scientists, managers, administrative staff and others.
Management (of a condition)	The coordinated provision of interventions for the ongoing care and treatment of a person after their diagnosis with a condition.
Marker analyte	A chemical compound that is measured in the blood sample and is used to indicate whether a baby is at an increased risk of a condition. Different conditions have different marker analytes.
Maternity service provider	An organisation or health care provider that delivers care during pregnancy, labour, birth and/or after birth. Examples include hospitals, general practitioners, birthing centres and community and independent midwives.
Missed cases	People who are eligible for newborn bloodspot screening who do not receive an abnormal screening result but who are diagnosed at a later stage with a condition for which they were screened. Missed cases can arise when eligible people are not offered screening, do not participate in screening (e.g. by refusing to participate) or are screened but the condition is not detected.
Monitoring	The systematic collection, assessment and reporting of program data in order to consider the program's performance in line with its objectives.

Morbidity	The state of being diseased, unhealthy or not physically or psychologically well as a result of having a condition.
Mortality	Deaths within a population that are caused by or associated with a condition.
Multidisciplinary	Combining or involving a range of academic disciplines, fields of expertise or professional specialisations (e.g. medical specialties), which are usually complementary in terms of skills, qualifications and experience.
Multiplex(ed) testing/screening	The use of an assay to measure the presence of more than one marker analyte in the same cycle of the assay. It is distinguished from procedures that measure one marker analyte at a time.
Natural history of a condition	The course a condition takes in a person over time in the absence of intervention.
Negative predictive value	The chance that a person with a negative test result truly does not have the condition. This is calculated as: $\text{True negatives} / (\text{true negatives} + \text{false negatives}) \times 100.$
Newborn bloodspot screening	This includes all processes associated with the taking a bloodspot sample from newborn babies and testing it to determine if those babies are at increased risk of a number of rare but serious conditions. It is also known as the Guthrie test and the 'heel-prick test'.
Newborn bloodspot screening program	The policies, processes, people and facilities that are involved in providing newborn bloodspot screening. It involves the efforts and interactions of all health professionals along the screening pathway, including health care providers such as clinicians, midwives and nurses; laboratory staff; and decision makers, policy makers and managers.
Normal result	A result of the screening test which does not suggest that the newborn is at increased risk of a condition.
Nucleotide	The basic building block of nucleic acids such as DNA.
Open culture	A 'culture' is a general sense of how things are done in an organisation. An open culture describes an organisational environment that is safe and just and in which people feel supported and are encouraged to identify and report events.
Open disclosure approach	A way of communicating openly with screening program participants, staff and others following an adverse event. The discussion should include an expression of regret for any harm that occurred as a result of the adverse event and an explanation of what happened, why it happened, what the consequences are and what is being done to prevent it from happening again.
Over-diagnosis	The diagnosis of a condition that will never cause symptoms or death for the person diagnosed. Thus over-diagnosis can result in people being treated for a condition when treatment is not necessary.
Participation	The action by families and newborns of taking part in newborn bloodspot screening.

Penetrance	A measure of the proportion of individuals in a population who carry a specific gene that also express the associated trait.
People-centred approach	See <i>Family-centred care</i> .
Policy	A statement that defines principles or rules for guiding decisions and actions.
Population based screening (program)	An organised approach for testing all people in a target population for a condition(s), generally before symptoms appear.
Positive predictive value	The chance that a person with a positive test result truly has the condition. This is calculated as: True positives/(true positives + false positives) x 100.
Primary purpose (of a dried bloodspot)	The main reason that the dried bloodspot sample is taken—that is, to do a blood test that aims to detect an increased risk for a number of conditions.
Private sector	A part of the economy where organisations such as hospitals are privately owned and not owned or operated by the government.
Private setting	See <i>Private sector</i> .
Procedure	The process or method to be used to put a policy into action. It outlines who will do what and where, when and why they will do it.
Process	A series of actions taken to achieve a particular result.
Protocol	A set of rules for explaining the correct conduct and procedures.
Public sector	A part of the economy where organisations such as hospitals are owned and operated by the government.
Public setting	See <i>Public sector</i> .
Quality	How well a health care service is performing to produce a desired outcome. There are a range of dimensions that can be used to assess quality, including whether the health care service is appropriate, acceptable, effective, efficient, responsive, continuous, sustainable, accessible, equitable, capable and safe.
Recruitment	The process of finding and attracting people to participate in the screening program.
Reporting	The ways in which data that have been collected against indicators, or for evaluation or review purposes, are communicated to the broad range of stakeholders involved in newborn bloodspot screening.
Review	A smaller one-off investigation or consideration of a specific program area or issue.
Sampling	See <i>Bloodspot sampling</i> .
Screening	The process of identifying people in a defined population who are at increased risk of a condition but do not yet have signs or symptoms of that condition. The aim is to identify conditions early to enable early intervention and management so that morbidity and mortality are reduced.
Screening panel	The conditions screened for by a newborn bloodspot screening program.

Screening pathway	The journey that a family will follow when they participate in a screening program.
Screening program	See <i>Newborn bloodspot screening program and Population based screening (program)</i> .
Screening test	A test that indicates whether a person has an increased chance of having a condition even if they have no symptoms of the condition.
Secondary use (of a dried bloodspot)	The ways in which bloodspots could be used after the newborn screening test has been completed. Examples include research, forensic/police investigations and coroner's investigations.
Sensitivity	A statistic that measures the performance of a test. It equals the percentage of people who are correctly identified as having the condition tested for. This is also called the true positive rate.
Sequencing	A laboratory technique that is used to determine the precise order of nucleotides in DNA.
Serious incidents or harms	Any adverse impact sustained through screening or related follow-up that has significant consequences for newborns, parents, staff or organisations involved in newborn bloodspot screening and which warrants a comprehensive response. These may be one-off significant events or smaller events that occur over time. What constitutes a serious incident or harm should be detailed in local policies with strategies for how they can be managed.
Specificity	A statistic that measures the performance of a test. It equals the percentage of people who are correctly identified as not having the condition for which they were tested. This is also called the true negative rate.
Symptom	A physical or mental feeling that indicates the existence of a condition.
Tandem mass spectrometry	An analytical technique used in newborn bloodspot screening laboratories that allows the identification of multiple conditions in the one process by separating and measuring substances according to their weight and chemical composition. Many marker analytes can be determined in the one procedure, allowing identification of those babies at risk of the conditions included in the screening panel.
Target population	The group of people to whom screening is aimed and offered, informed by scientific evidence to determine who will most benefit from participating in screening.
Terms of reference	A statement of the scope and limitations of an activity or a group of people working together to achieve a shared goal—for example, on a project, committee, working group or program.
Test protocol	A formal process that typically outlines requirements and activities to be completed by the newborn bloodspot screening laboratory in relation to the screening test for a condition.

Transparency	Operating in a way that is open and honest and makes it easy for others to see what actions and decisions are occurring. This involves intentionally disclosing information that is relevant, accessible, timely and accurate.
Unknown significance	Where it is unclear or it cannot be determined whether the test result is clinically significant.
Unsuitable sample	A bloodspot sample that cannot be used by the laboratory for testing because the quality of the bloodspot has been compromised. This can occur from events such as incorrect timing of bloodspot sample collection, incorrect method of collection and/or mishandling of the bloodspot sample card post-collection.

Appendix A: Policy development

CONTEXT: WHY A POLICY FRAMEWORK

In 2013, Australian health ministers received a request from a professional group to expand newborn bloodspot screening to include congenital adrenal hyperplasia (CAH). Health ministers forwarded this request to the Standing Committee on Screening (SCoS). In responding to the health ministers' request, SCoS recognised the success of newborn bloodspot screening programs but also noted that there were no national policies supporting the programs and no decision-making process to enable assessment of CAH. In response to this, SCoS established the Newborn Bloodspot Screening Working Group to draft a policy framework to address the policy gap, facilitate decision making and support the future success of newborn bloodspot screening programs in Australia.

ABOUT THE NEWBORN BLOODSPOT SCREENING WORKING GROUP

The Newborn Bloodspot Screening Working Group was established in March 2014 to draft the policy framework on behalf of SCoS. It was responsible for developing a project and communication plan and drafting the policy framework with relevant input from stakeholders. The working group was a multidisciplinary group comprising individuals with expertise that spanned the newborn bloodspot screening pathway. The membership of the working group is described below. The working group met 14 times over the 18 months in which the policy framework was drafted. The working group was disbanded in early 2016.

Member	Representing / area of expertise
Clinical Associate Professor Craig White	Chair, independent health policy expert
Ms Samantha Chandler	Consumer
Ms Rebecca Doherty	Jurisdictional screening policy expert
Dr Jan Fizzell	Standing Committee on Screening representative
Dr Janice Fletcher	Newborn screening program management expert
Ms Karla Lister	Jurisdictional screening policy expert and Project Manager
Dr Jim McGill	Royal Australasian College of Physicians representative
Ms Marie Mutton	Australian College of Midwives representative
Dr Ainsley Newson	Senior Lecturer in Bioethics
Mr Ricky Price	Newborn screening scientist
Dr Bernie Towler	Commonwealth Government representative
Ms Gail Ward	Standing Committee on Screening representative
Professor Bridget Wilcken	Human Genetics Society of Australasia representative
Clinical Associate Professor Veronica Wiley	International newborn screening expert
Secretariat	
Ms Selina Metternick-Jones and Ms Faye Bowman	Screening Policy Section, Department of Health, WA

ABOUT THE POLICY DEVELOPMENT PROCESS

The policy development process followed an iterative approach. First, the working group agreed on the core components of a high-quality and effective screening program, taking guidance from the Australian *Population Based Screening Framework* and other sources of information. These components were then used as a foundation for the policy framework, arriving at the key topics: background; program overview; program implementation; quality and safety; and decision making.

Having developed an outline of the document, the content for each policy area was then drafted. In the later stages of policy development, the content was collated into one document and reviewed in full by working group members on numerous occasions. The content was informed by multiple sources of information, including relevant international and national policy documents and guidance; academic literature; local program information; and stakeholder input.

COMMUNICATION AND ENGAGEMENT

A guiding principle of the working group was to work openly and engage early and often with its stakeholder groups. In line with this, the working group released communiques after each meeting, which were included on the project website and also shared with stakeholder networks.

The working group also held two stakeholder workshops and a national survey. Through these engagement approaches, approximately 450 individuals provided input into the policy framework, particularly relating to the:

- needs and expectations of stakeholder groups from a high-quality and effective newborn bloodspot screening program
- benefits of newborn bloodspot screening
- potential harms and risks
- decision-making criteria
- high-level policies within the policy framework.

The input provided by these processes was used to guide the development of the policy framework.

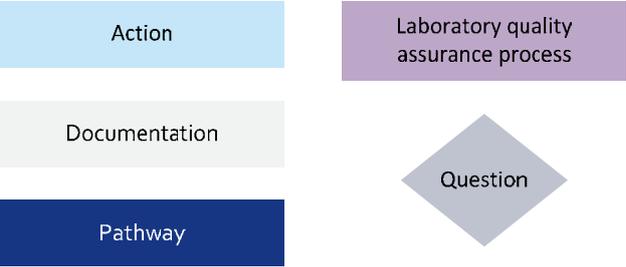


Appendix B: National screening pathway

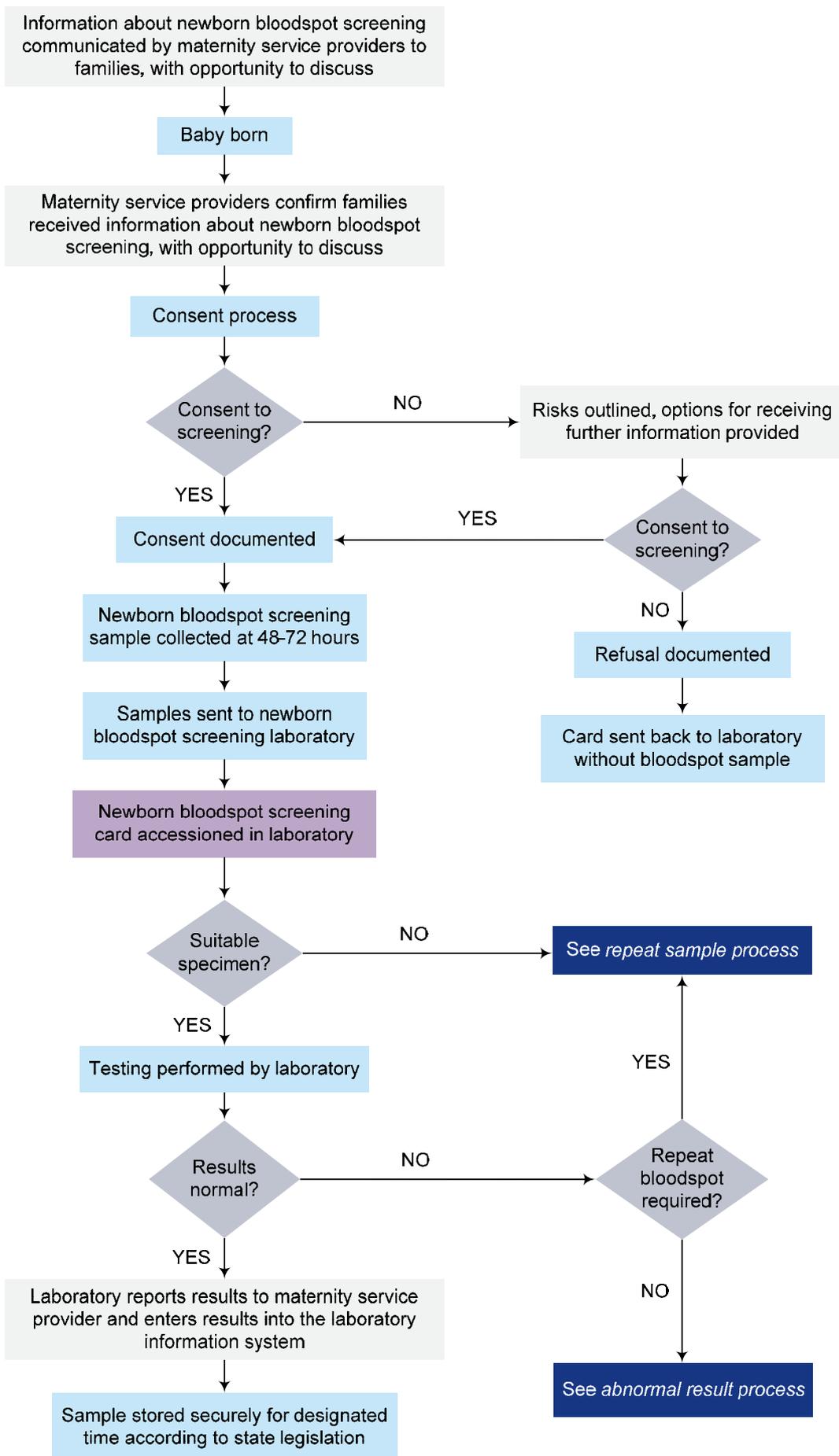
The screening pathway is presented as a flow chart on the following pages, and outlines the steps followed:

- I. from when a family enters the program to the point of a normal or abnormal result
- II. for an abnormal result
- III. for a repeat sample.

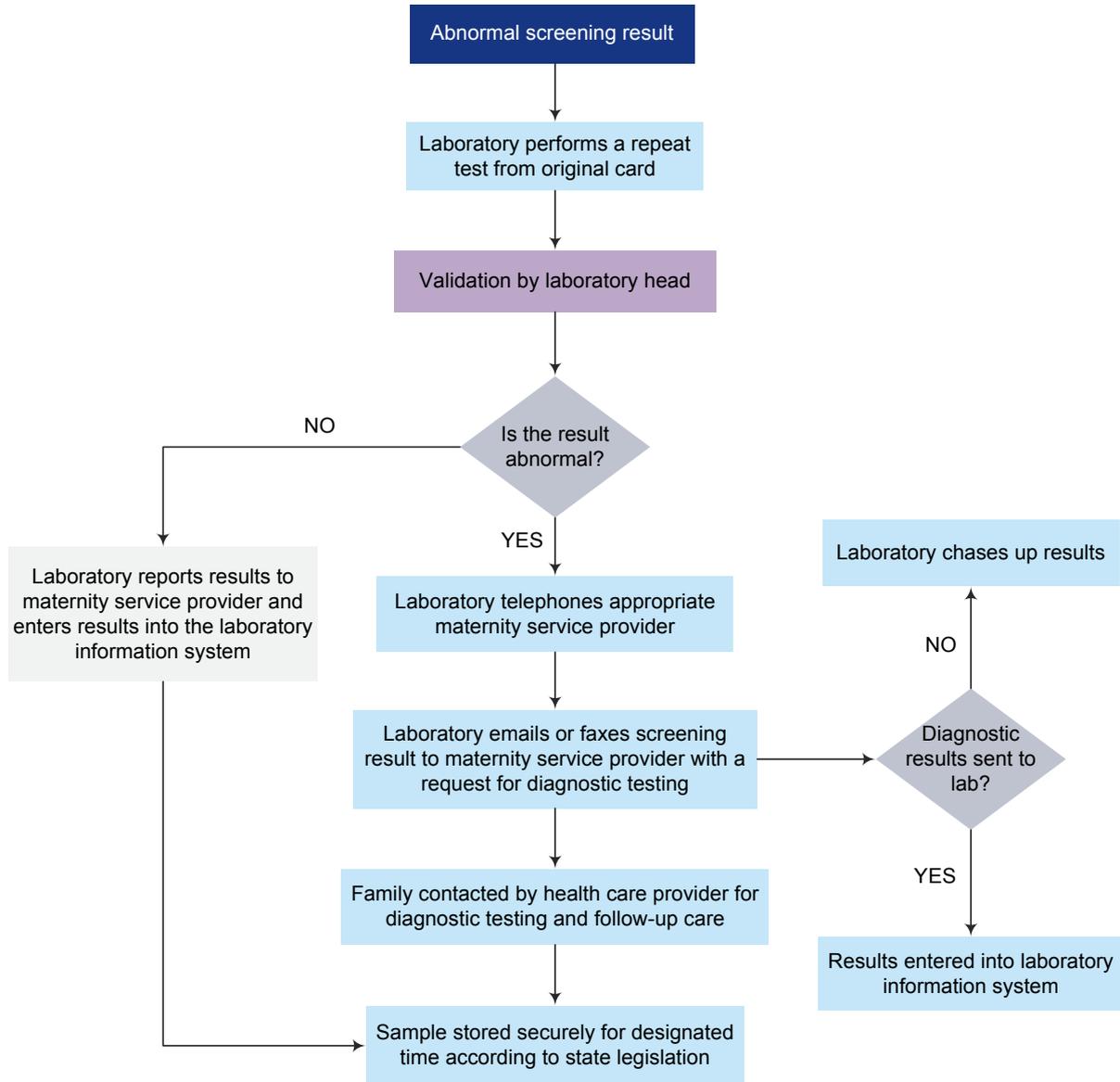
Key



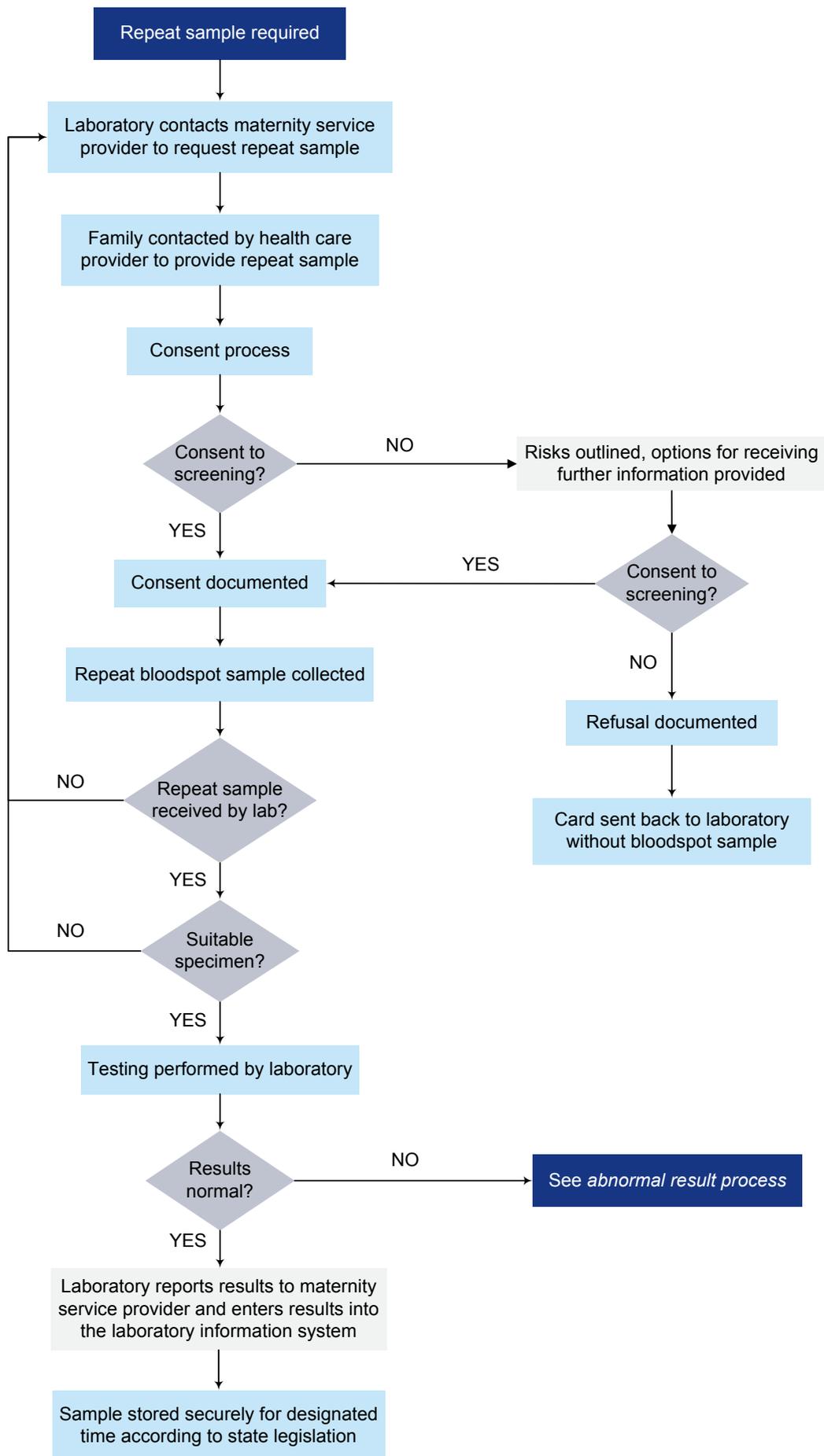
PART I: OVERALL SCREENING PATHWAY



PART II: ABNORMAL RESULT PROCESS



PART III: REPEAT SAMPLE PROCESS



Appendix C: Nomination form (addition)

Nomination form requesting assessment of a condition for *addition to* newborn bloodspot screening

Please submit to the Newborn Bloodspot Screening Program Management Committee via
SCoS@health.gov.au

Date received:

(to be completed by secretariat)

Questions	Response
Name of nominator(s)	
Organisation(s) (if applicable)	
Contact details (address, phone, email)	
Role(s) (for example, clinician, researcher, parent, advocate etc.)	
Condition nominated for assessment (specifying form(s), if applicable)	
OMIM* or other names for the condition	

*Online Mendelian Inheritance in Man: <http://www.omim.org/>

Instructions for completion

- Please complete as many of the 'response' sections within this form as possible, citing relevant references within the text by number, then list and attach all references at section 6.
- It is recommended that a nominee who is not from a newborn bloodspot screening program seeks the advice and guidance of their jurisdiction's newborn bloodspot screening program regarding the required documentation and evidence in order to make a submission for the addition or removal of a condition.
- When the nomination form is complete, it should be submitted to the Newborn Bloodspot Screening Program Management Committee.

1. THE CONDITION

The condition should be a serious health problem that leads to significant morbidity or mortality. There should be a benefit to conducting screening in the newborn period; and the natural history of the condition, including development from latent to declared disease, should be adequately understood.

Guiding questions	Response
What is the incidence of the condition in Australia? Is this determined clinically or through screening studies in other countries?	
What is the burden of disease associated with the condition, including morbidity and mortality? What is the spectrum of disease—in particular, are there mild or late-onset forms?	
At what age would the condition usually be detected clinically?	
What are the benefits of early diagnosis and intervention/treatment? (Consider such benefits as early intervention, prevention of symptoms, reduction of disease severity, provision of a definitive diagnosis, emotional and social benefits and provision of information that would assist families with reproductive decision making.)	
What are the possible harms of screening and/or early diagnosis?	

2. THE TEST

There should be a suitable test protocol to identify the presence of the condition, and the test protocol should be socially and ethically acceptable to health professionals and the public.

Guiding Questions	Nominator's response
Describe a detailed methodology for the test (for example, tandem mass spectrometry, immunoassay, molecular), including any second-tier testing required. Provide reference to a published methodology and describe any modifications required.	
Can the test be performed on the same dried bloodspot specimen that is used currently? If not, what additional sample would be required?	
For the proposed testing protocol, comment on the:	
clinical and analytic validity	
sensitivity	
specificity	
false positive rate	
false negative rate	
positive predictive value	
negative predictive value	
Can the test be multiplexed?	
What other conditions may be detected (clinical or of unknown significance)?	
What would be the cost of the test?	
If DNA analysis is required, would testing include common mutations, a panel or full sequencing?	
What are the potential harms associated with the test protocol?	

3. THE INTERVENTION

There should be an accepted intervention for patients with recognised disease, and facilities for diagnosis and management should be available so that these services can be offered if there is a positive screening result.

Guiding Questions	Nominator's response
What diagnostic testing is necessary? Is it available and reliable? What is its associated cost?	
What is the established intervention/treatment for this condition?	
Do all patients require an intervention or treatment upon diagnosis? If not, can those who require treatment be distinguished from those who do not?	
How effective is the intervention/treatment? (Does it alleviate symptoms, slow/halt progression?)	
What are the impacts on quality of life?	
How urgent is the intervention/treatment? Must it be initiated before symptoms present?	
What are the potential harms of the	
intervention/treatment?	
What is the cost of the intervention/treatment?	
What facilities are required to deliver the intervention/treatment? Do current health care facilities in each state and territory have capacity, and are they of sufficient quality, to support the intervention/treatment? Is there equitable access to the intervention/treatment?	

4. COST-EFFECTIVENESS

Guiding Questions	Nominator's response
Provide any available evidence for the cost-effectiveness of screening for this condition, either from Australia or internationally.	

5. ANY OTHER COMMENTS

6. REFERENCES

Please list and attach relevant references

Appendix D: Nomination form (removal)

Nomination form requesting assessment of a condition for *removal from newborn bloodspot screening*

Please submit to the Newborn Bloodspot Screening Program Management Committee via
SCoS@health.gov.au

Date received: _____ (to be completed by secretariat)

Questions	Response
Name of nominator(s)	
Organisation(s) (if applicable)	
Contact details (address, phone, email)	
Role(s) (for example, clinician, researcher, parent, advocate etc.)	
Condition nominated for assessment (specifying form(s) if applicable)	
Screening method	
OMIM* or other names for the condition	

*Online Mendelian Inheritance in Man: <http://www.omim.org/>

Instructions for completion

- Please complete as many of the 'response' sections within this form as possible, citing relevant references within the text by number, then list and attach all references at the end of the form.
- It is recommended that a nominee who is not from a newborn bloodspot screening program seeks the advice and guidance of their jurisdiction's newborn bloodspot screening program regarding the required documentation and evidence in order to make a submission for the addition or removal of a condition.
- When the nomination form is complete, it should be submitted to the Newborn Bloodspot Screening Program Management Committee.

Guiding Questions	Nominator's response
When was screening initiated for this condition and why?	
What is the rationale for proposing to remove the condition from screening? Provide relevant information drawing on current screening experience and a review of literature to support removal.	
What is the incidence in Australia? Is this determined clinically or through screening studies in Australia or other countries?	
What positive impacts would removing this condition have on the program (for example, in terms of the impact on families, on the laboratory, on maternity service providers etc.)?	
What would be the clinical implications of removing the condition from screening? Include reference to the burden of disease associated with the condition, including morbidity and mortality, and the spectrum of disease.	
Are there other risks of removing this condition from screening (for example, impact on the ability to detect other conditions; impact on the family, including future reproductive risk; community concern etc.)?	
Is the condition screened internationally?	
Would removal of this condition from screening have any other implications for the quality of the program?	
Are there any alternatives to removal (for example, alterations to cut-offs, further follow-up testing etc.)?	
For the current testing protocol, comment on the:	
clinical and analytic validity	
sensitivity	
specificity	
false positive rate	
false negative rate	
positive predictive value	

negative predictive value	
Is the test multiplexed?	
Does testing identify other conditions (clinical or of unknown significance)?	
What would be the cost implications of removing the test?	

ANY OTHER COMMENTS

REFERENCES

Please list and attach relevant references

Appendix E: Screening principles

WORLD HEALTH ORGANIZATION SCREENING PRINCIPLES

Condition

- The condition should be an important health problem.
- There should be a recognisable latent or early symptomatic stage.
- The natural history of the condition, including development from latent to declared disease, should be adequately understood.

Test

- There should be a suitable test or examination.
- The test should be acceptable to the population.

Treatment

- There should be an accepted treatment for patients with recognised disease.

Screening Program

- There should be an agreed policy on who to treat as patients.
- Facilities for diagnosis and treatment should be available.
- The cost of case findings (including diagnosis and treatment of patients diagnosed) should be economically balanced in relation to possible expenditure on medical care as a whole.
- Case findings should be a continuing process and not a 'once and for all' project.

The above screening principles were developed by Wilson and Jungner in 1968 and subsequently endorsed by the World Health Organization to guide programs aimed at the early diagnosis of disease. These criteria are considered to be the gold standard and have been used worldwide to assess conditions for screening. In line with this, they have informed Australia's *Population Based Screening Framework*.

It is widely accepted that, in the context of rare diseases and newborn bloodspot screening, these principles are a starting point but require further consideration [50, 51]. This is because these principles focus on common conditions, whereas the conditions screened in newborn bloodspot screening programs are rare. In addition, the above principles do not provide sufficient detail to consider in full the additional social and ethical issues relating to newborn bloodspot screening compared with screening a population of consenting adults. Therefore, the above principles are commonly amended to suit newborn bloodspot screening and the local context in which programs operate [52–54].

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