

**COMMUNITY CARE AND POPULATION HEALTH
PRINCIPAL COMMITTEE**

STANDING COMMITTEE ON SCREENING

**POPULATION BASED
SCREENING FRAMEWORK**
Updated September 2016

CONTENTS

INTRODUCTION

- Screening
- The screening process
- Screening in Australia
- Emerging issues

AUSTRALIAN CRITERIA FOR THE ASSESSMENT OF POPULATION SCREENING

- Australian criteria for deciding whether a new screening program should be introduced in a defined target population
- Points to consider in using the criteria to assess screening programs

PRINCIPLES FOR THE IMPLEMENTATION AND MANAGEMENT OF SCREENING PROGRAMS

- Key principles
- Points to consider for the implementation and management of screening programs

GLOSSARY

REFERENCES

First endorsed by the Australian Health Ministers' Advisory Council (AHMAC) at its 9 October 2008 meeting. Updates endorsed by the Community Care and Population Health Principal Committee of AHMAC in September 2016.

INTRODUCTION

The Australian Population Based Screening Framework was first developed by the Screening Subcommittee of the Australian Population Health Development Principal Committee and endorsed by the Australian Health Ministers' Advisory Council (AHMAC) in 2008. In 2016, the framework was updated to incorporate new and emerging issues by the Standing Committee on Screening (SCoS) of the Community Care and Population Health Principal Committee (CCPHPC), of AHMAC.

The purpose of the framework is to inform decision makers on the key issues to be considered when assessing potential screening programs in Australia. The framework has been divided into two parts:

- the criteria which should be used to assess whether screening should be offered or a screening program introduced for diseases or conditions.
- the key principles for the implementation and management of screening programs.

The framework is underpinned by the principles of access and equity which are fundamental elements to all population screening programs, and is intended to provide guidance and inform judgement.

SCREENING

The World Health Organization (WHO) defines screening as the presumptive identification of unrecognised disease or defects by means of tests, examinations or other procedures that can be applied rapidly. Screening is intended for all people, in an identified target population, who do not have symptoms of the disease or condition being screened for. The process can identify:

- a pre-disease abnormality;
- early disease; or
- disease risk markers.

The aim of screening for a disease or a risk marker for a disease is to reduce the burden of the disease in the community including incidence of disease, morbidity from the disease or mortality. This is achieved by intervening to reduce individual risk of the disease or detecting the disease earlier on average than is usually the case in the absence of screening and thereby improving disease outcome.

Screening can reduce the risk of developing or dying from a disease, but it does not guarantee that disease will not occur, or if it occurs, that it can be cured. A 'positive' screening test identifies people who are at increased likelihood of having the condition and who require further investigation to determine whether or not they have the disease or condition.

As screening has benefits, costs, and harms, there is an ethical obligation to maximise benefits and minimise harm; and the overall benefits should outweigh any harms that result from screening. When community resources are used to fund screening there should be community consensus that the benefits of screening justify the expense of screening.

Benefits include:

- Reducing the burden of disease on the community and individuals.
- Reducing mortality from the disease.
- Reducing morbidity from the disease.
- Improving disease outcomes.

Harms associated with screening may include:

- **False positives:** when a screening test and assessment delivers a positive result but the individual does not have the disease.
- **False negatives:** when a screening test and assessment delivers a negative result but the individual does have the disease.
- **Over-diagnosis:** is terminology used to explain that some cancers and conditions that are found and treated may not have become life-threatening in an individual's lifetime. It does not refer to error or misdiagnosis.
- **Other physical and psychological harms** that might be experienced as a result of screening or treatment.

In 1968, Wilson and Jungner developed the WHO principles of screening¹. These principles are outlined in the box below. These principles still remain relevant today, when developing criteria for a specific country or screening issue.

WHO Principles of Early Disease Detection

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 whom 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.

¹ Wilson JMG, Jungner G., 1968, *Principles and practice of screening for disease*. Public Health Paper, Number 34. Geneva: WHO.

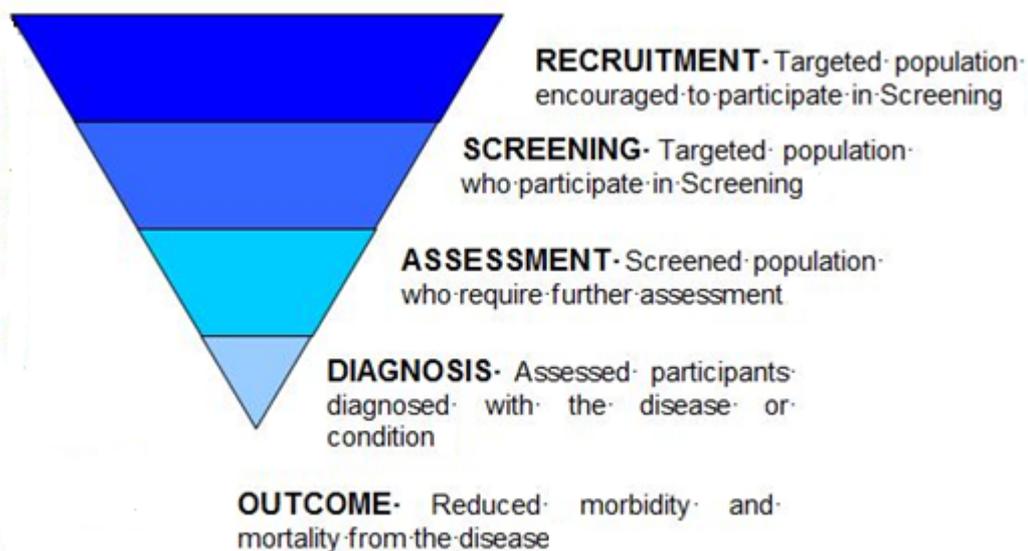
It is important to distinguish between different forms of screening and other health tests available.

- **Population based screening** is where a screening test is offered systematically to all individuals in the defined target group within a framework of agreed policy, protocols, quality management, monitoring, evaluation and review. Population based screening is an organised integrated process where all activities along the screening pathway are planned, coordinated, monitored and evaluated through a quality improvement framework. All of these activities must be resourced adequately to ensure benefits are maximised (e.g. breast cancer screening).
- **Case-finding or opportunistic screening** is where a test is offered to an individual with or without symptoms of the disease when they present to a health care practitioner for reasons unrelated to that disease (e.g. when a GP orders blood tests when a patient presents for a flu shot).
- **Targeted risk screening** is screening of selected high risk groups. This can include genetic screening of people with a strong family history of certain cancers that may have a known genetic cause, or a group with specific exposures through environmental and occupation health, such as asbestos-exposed workers.
- **Routine examinations or planned opportunistic screening** are a well-established pattern of medical examinations extending through life which may provide protection against disease through early intervention (e.g. school based screening).
- **Diagnostic testing** occurs when a test is offered to an individual with symptoms of a disease or medical condition to confirm or exclude the suspected condition.

THE SCREENING PROCESS

The following diagram describes the screening pathway. Underlying the pathway is the principle of quality assurance at each point.

DEFINED TARGET POPULATION



SCREENING IN AUSTRALIA

Tuberculosis was the first disease for which a screening test was identified in the 1940s. In 1949 the first screening program was introduced in Australia to control tuberculosis. The Australian Tuberculosis Campaign provided free diagnostic and treatment services for tuberculosis. Screening for tuberculosis was opportunistic until the 1960s when compulsory community-wide chest X-rays were introduced. In 1976 screening for tuberculosis ceased as illness and death from the disease had been significantly reduced.

Newborn Bloodspot Screening commenced in the 1960s, with all newborns initially being tested for phenylketonuria (PKU). The program has evolved to now screen for more than 25 conditions.

In 1989, an evaluation of both breast cancer screening and cervical screening was undertaken in Australia. Without organised screening programs there was concern that breast and cervical screening was not being conducted in a way which provided optimal benefit to the community. In 1991, BreastScreen Australia and the National Cervical Screening Program were introduced after recommendations from the National Breast Cancer Screening Evaluation and the National Cervical Cancer Screening Evaluation.

The Bowel Cancer Screening Pilot Program was conducted from 2002 until 2004 to test the feasibility, acceptability and cost effectiveness of bowel cancer screening in Australia. The final evaluation report showed that a national bowel cancer screening program would be feasible, acceptable and cost effective. In 2006, the phased introduction of the National Bowel Cancer Screening Program commenced.

In July 2009, the Council of Australian Governments (COAG) endorsed a recommendation that universal neonatal hearing screening would be available in all states and territories by the end of 2010.

In 2016, population-based screening programs available in Australia include:

- The National BreastScreen Australia Program;
- The National Cervical Screening Program;
- The National Bowel Cancer Screening Program;
- Newborn Bloodspot Screening; and
- Newborn Hearing Screening.

A KEY EMERGING ISSUE: GENOMIC SCREENING

A key emerging issue is the potential of genomic testing, technologies and knowledge to impact upon screening. In 2003, the Human Genome Project provided a map of the entire sequence of DNA – the human genome. Genomics will enable the identification of a gene or genes that predispose or increase the risk of an individual or individuals developing certain diseases or conditions, and the identification of multiple genes that may be associated with increased or decreased risk of common diseases, such as cancer or diabetes. These advances in technology have also greatly reduced both the time and cost associated with sequencing genes and genomes, and may have the potential for improving health at the population level.²

In 2008, the WHO discussed the coming of age of genetics in their paper '*Revisiting Wilson and Jungner in the genomic age: a review of screening criteria over the past 40 years*'.³ The paper discussed how genetic screening is being proposed as a vehicle for translating genetic and genomic advances into population health gains. However, they cautioned against the widening gap between technology and the pressure to introduce or expand screening programs before adequate frameworks are in place.

The paper acknowledged the continued relevance of Wilson and Jungner's 1968 screening principles, but highlighted the need to consider how these criteria can be applied to the growing field of genetics and genomics.

While advancements in genetics and genomics provide an opportunity for greater understanding, prevention and treatment of diseases, there are significant harms and risks associated with screening using genetics that have clinical, ethical, legal, social and economic implications.

The Standing Committee on Screening (SCoS) continues to keep a watching brief on the use of genetic and genomic technologies and their potential for application in screening at the population level. The SCoS continues to liaise with other Australian Government committees currently considering genetic and genomic issues.

² Centers for Disease Control and Prevention, Public Health Genomics, Viewed 27 January 2016, from: <http://www.cdc.gov/genomics/gtesting/index.htm>

³ Andermann, A, Blancquaert, I., Beauchamp, S., Dery, V. 2008. *Revisiting Wilson and Jungner in the genomic age: a review of screening criteria over the past 40 years*. Bulletin of the WHO, 86: 241-320.

AUSTRALIAN CRITERIA FOR THE ASSESSMENT OF POPULATION SCREENING

The Australian Population Screening Framework for cancer and other chronic diseases has been adapted from the 1968 WHO criteria but takes into account:

- the need for a strong evidence base in making a decision about the introduction of a screening program including evidence of the safety, reproducibility and accuracy of the screening test and the efficacy of treatment; and
- the requirement that a screening program offers more benefit than harm to the target population.

The decision to introduce a screening program needs to also consider whether the outcomes in the research setting can be reproduced in population screening settings.

This framework is not designed to address targeted testing of high risk groups. In planning the coverage of screening programs, however, steps must be taken to ensure that consideration is given to those at high risk of developing the disease being screened for, and that policies are in place for the appropriate identification and management of individuals in these high risk groups.

AUSTRALIAN CRITERIA FOR DECIDING WHETHER A NEW SCREENING PROGRAM SHOULD BE INTRODUCED IN A DEFINED TARGET POPULATION

When deciding whether a new organised screening program should be introduced in Australia the following criteria should ideally be met:

1. CONDITION

The condition should be an important health problem. The seriousness of the disease relates to issues of cost-effectiveness and ethics of the screening program. As screening programs may cause harm to some, it is important that screening is only undertaken for serious diseases or conditions to justify the potential harms that may occur with screening.

The disease, or risk marker for the disease, being screened for should be well defined.

The epidemiology of the disease in the target population should be known, including the incidence, prevalence and projected trends; and mortality, morbidity and burden of disease by age and sex. The prevalence of the disease should be known as this will affect the positive predictive value of the test and is necessary for interpretation of the test results.

Information on the relationship between risk markers for the disease, an early asymptomatic stage and/or a recognisable latent period should be available. The relationship between the risk marker and the disease should be causal and there should be evidence to show that reducing the risk marker would lead directly to a reduction in probability of developing the disease or in the disease outcome.

CRITERIA TO BE MET

The condition:

- is an important health problem.
- has a recognisable latent or early symptomatic stage.

The natural history of the disease or condition, including, where relevant, the relationship between the risk marker and the disease and the development from latent to declared disease is adequately understood.

2. TEST

The accuracy of the screening test relates to its ability to identify those people who have the disease and to exclude those who do not. The test should be effective in detecting the early stage of a disease.

A screening test should be able to detect most people with the target disease or risk factor (high sensitivity) and be able to exclude most people without the disease or risk factor (high specificity).

If the test is positive it should indicate that the disease is present (high positive predictive value) and if the test is negative for the disease indicate that the disease is not present (high negative predictive value).

The test should be relatively easy to perform and interpret. It should be reliable and give consistent results when used in large populations and should show reproducible results (interobserver variation, intraobserver variation, instrument variation and variation in the biological characteristics being tested should be minimal).

Any harm caused, or that may be caused, by the screening test should be acknowledged, communicated to those undergoing screening and accurately measured. Steps should be taken to minimise or eliminate the harm. The distribution of test values in the target population should be known. A suitable cut-off level for the screening should be defined for what determines both a positive test and a negative test.

There should be consideration of issues that may impact on the test's acceptability to people performing or having the screening test. This could include issues such as convenience, ease of use (if self administered), discomfort, embarrassment, cost and real and perceived risks.

The screening test should be able to be offered in a way that respects peoples' concerns, their right to make choices and their privacy and confidentiality. The test should be able to be delivered consistently regardless of participants' demographic status. It is important there is equity of access to the test regardless of rurality, ethnicity, socio-economic status or disadvantage status.

CRITERIA TO BE MET

The test:

- is highly sensitive.
- is highly specific.
- is validated.
- is safe.
- has a relatively high positive predictive value.
- has a relatively high negative predictive value.
- is acceptable to the target population including important sub groups such as target participants who are from culturally and linguistically diverse backgrounds, Aboriginal and Torres Strait Islander people, people from disadvantaged groups, and people with a disability.

There are established criteria for what constitutes positive and negative test results, where a positive test result means that the person needs further investigations, and a negative test result means the person is rescreened at the usual interval, where applicable.

3. ASSESSMENT

Systems should be in place for safe follow-up for diagnostic assessment of individuals with a positive screening test provided as part of the organised screening program.

This should be provided in a way that provides equity of access to the relevant assessment services regardless of rurality, ethnicity, socio economic status or disadvantage status.

Evidence based guidelines and policies for assessment, diagnosis, intervention and support should be available for people with a positive test result.

CRITERIA TO BE MET

Systems should be in place for evidence based follow up assessment of all people with a positive screening test regardless of rurality, ethnicity, socio economic status or disadvantage status.

4. TREATMENT

There should be evidence that the treatment intervention is effective, will lead to reduction in the burden of the disease or condition and is more effective than treatment at a later stage that would occur without screening. Evidence based best practice guidelines and policies for treatment should exist.

There should be adequate expertise, resources and capacity in the workforce and medical facilities available to provide treatment and support for those diagnosed through screening.

The treatment should be acceptable and accessible to those people who have the disease or condition identified through screening.

CRITERIA TO BE MET

The treatment must be effective, available, easily accessible and acceptable to all patients with the recognised disease or condition.

5. SCREENING PROGRAM⁴

A high level of evidence is essential to make decisions about screening programs as screening is offered to healthy people and has the potential for causing harm that would not have occurred if they had not participated in screening.

A high level of evidence from randomised controlled trials (RCTs) or systematic reviews of RCTs, of the benefit of screening for the disease or condition with a particular screening test and treatment in terms of reduction in burden of disease (morbidity and mortality). The quality of the RCTs should be high.

There should be clear evidence that screening and treating people with early disease (or risk markers) detected through screening, leads to better outcomes than finding and treating disease at a later stage where people present with signs or symptoms of the disease.

An assessment of the benefits and harms of screening should be undertaken. A quantitative analysis of the levels of morbidity and mortality that can be prevented by screening should be undertaken. The benefits can then be set against the financial costs and human costs to the person screened such as anxiety, discomfort, adverse effects, follow-up investigations, over-diagnosis and possible over treatment so that a decision about implementing a screening program can be made.

There needs to be a defined target population that can be identified and invited to participate in screening. The screening program should aim to maximise the benefits to individuals and at a population level.

The program should give more benefit than harm to the target population. Balanced information on the potential benefits and harms of the screening program should be available to the target population to enable them to make an informed decision about participating in screening.

⁴ Adapted from: Strong K, Wald N, Miller A and Alwan A on behalf of the WHO Consultation Group Current concepts in screening for non communicable disease: World Health Organisation Consultation Group Report on methodology of non communicable disease screening, Journal of Medical Screening , 2005 Vol 5 No. 1

The screening program (i.e. test, diagnostic procedures, and intervention) should be clinically, socially and ethically acceptable to both health professionals and consumers.

There should be an agreed policy on the diagnostic investigative assessment of individuals with a positive test result and on the choices available to those individuals.

The screening interval will be determined through evidence of the natural history of the disease from an early latent phase to a more advanced stage.

People who have a positive screening test result undergo assessment and diagnosis to determine whether or not they have the disease or risk marker being screened for. Evidence based guidelines and policies for assessment, diagnosis, intervention and support should be available for people with a positive test result.

Evaluation of a potential screening program must include consideration of whether the proposed program is feasible. Implementation of the screening program should be achievable and consistent in policy and quality nationally. The organisation and coordination of activities across the entire screening pathway are essential elements of a screening program. The infrastructure and systems necessary to manage and implement the screening program to achieve similar outcomes to those achieved in the research setting on which the program is based should exist, or can be developed in a reasonable time frame. This includes:

- invitation and recruitment mechanisms;
- quality improvement systems;
- workforce and facility capacity for screening, diagnosis and treatment;
- education, training and expertise of health professionals;
- monitoring and evaluation; and
- information and support to participants.

If information is unavailable on some of these issues, a pilot could be considered to gain further information.

An economic evaluation should be performed to assess the opportunity cost of the screening program (including testing, diagnosis, treatment, administration, training and quality improvement). The economic evaluation should address the questions of:

1. Allocative efficiency: Is screening worthwhile? (do benefits exceed costs?)
2. Technical efficiency: If screening is deemed worthwhile, what are the most cost effective options for achieving the screening program's objective? These may include other options such as emerging improvements in treatment methods or by funding more resources to increase interventions already in place. The results of the economic evaluation should demonstrate that screening is the most cost effective intervention to reduce the burden of disease.

SCREENING PROGRAM CRITERIA

The Screening Program must:

- Respond to a recognised need.
- Be clinically, socially, legally and ethically acceptable to health professionals, consumers and the Australian public.
- Have a clear definition of the objectives of the program and the expected health benefits.
- Have scientific evidence of screening program effectiveness.
- Identify the target population who stand to benefit from screening.
- Clearly define the screening pathway and interval.
- Ensure availability of the organisation, infrastructure, facilities and workforce needed to deliver the screening program.
- Have measures available that have been demonstrated to be cost effective to encourage high coverage.
- Have adequate facilities available for having tests and interpreting them.
- Have an organised quality control program across the screening pathway to minimise potential risks of screening.
- Have a referral system for management of any abnormalities found and for providing information about normal screening tests.
- Have adequate facilities for follow-up assessment, diagnosis, management and treatment.
- Have evidence based guidelines and policies for assessment, diagnosis and support for people with a positive test result.
- Have adequate resources available to set up and maintain a database of health information collected for the program.
- Integrate education, testing, clinical services and program management.
- Have a database or systems available capable of providing a population register for people screened that can issue invitations for initial screening, recall individuals for repeat screening, follow those with identified abnormalities, correlate with morbidity and mortality results and monitor and evaluate the program and its impact.
- Plan evaluation from the outset and ensure that program data are maintained so that evaluation and monitoring of the program can be performed regularly.
- Be cost-effective.
- Ensure informed choice, confidentiality and respect for autonomy.
- Promote equity and access to screening for the entire target population including important sub groups such as participants who are from culturally and linguistically diverse backgrounds, Aboriginal and Torres Strait Islander people, people from disadvantaged groups, and people with a disability.
- Ensure the overall benefits of screening outweigh the potential harms, including psychological, physical, social, cultural, ethical and legal harms.

6. TREATMENT AND ONGOING MANAGEMENT

There will be policies and procedures in place for onward referral of individuals diagnosed with the risk marker or disease through the screening program. All individuals diagnosed through the screening program will be referred to health professionals with known expertise in the management of the disease or condition to ensure optimal outcomes.

The program will have policies for individuals diagnosed with the disease through the program regarding their status in relation to the screening program in future years.

The program will actively support timely transition from screening program diagnosis to treatment. Psychosocial support for individuals diagnosed through the program will be actively encouraged.

CRITERIA TO BE MET

Treatment and management considerations:

- Ongoing management referral protocols must be established for individuals who have the disease or condition detected through the screening program.
- There needs to be an established policy for the management of individuals who are identified at high risk of developing the disease or condition.

PRINCIPLES FOR THE IMPLEMENTATION AND MANAGEMENT OF SCREENING PROGRAMS

For diseases that meet the criteria provided previously, the following key principles should be considered for implementation and management of a program. Prior to a decision to implement the full program, a pilot may need to be conducted to ensure the program can be adapted to the Australian context.

The following defines the framework which underpins the key principles for implementing and managing a screening program:

- There must be agreement by the Australian and state and territory governments that a population based screening program should be implemented.
- There should be stakeholder agreement and acceptance of the decision to introduce the program.
- A national policy framework should be agreed to which defines the goals and objectives of the program.
- An agreed quality management plan should be in place to ensure ongoing managing of quality and a continuous quality improvement framework.
- Sufficient funding should be agreed and allocated to ensure the screening program is able to achieve its targets and objectives.

KEY PRINCIPLES

1. NATIONAL POLICY AND PROTOCOLS FRAMEWORK

Program policies and protocols must be evidence based, including:

- Develop a detailed national policy framework which includes: the screening age range, screening interval, follow-up tests for those with a positive screening test result, clinical guidelines for treatment and management, ongoing surveillance processes and identification and management of high risk groups.
- Define the screening pathway for the program which is based on the best available evidence. The pathway must be efficient, cost effective and make the best use of resources.
- Screening to diagnosis must be able to be delivered in a timely manner, minimising potential harms of delayed diagnosis and treatment.
- Identify the resources required for the program including funding allocation, workforce and facilities and establish how these resources can be developed or established and used efficiently.
- Define the roles and responsibilities of each level of government.
- Define the governance, organisation and co-ordination of the program at each level of government including the establishment of a register, invitation protocols, follow-up protocols and how quality management processes will be built into the program.

2. PROGRAM PLANNING AND DESIGN

- Identify participation objectives in order to achieve population benefits.
- Identify the timeframes expected to implement the program.
- Develop models for workforce infrastructure and service delivery based on local circumstances and projected needs and demands as the participation rates and target population increase.
- Ensure service delivery model provides equitable access.
- Develop evidence based strategies for recruiting people in the target population (including special sub groups) and ensure ongoing participation.
- Ensure informed consent processes are in place along the screening pathway.
- Develop and provide information and support for participants across all aspects of the screening pathway.
- Agreed participation objectives and plans.
- Program responsibility clearly defined.
- Program organisational structure and governance defined and agreed.
- Extensive consultation – Identify and engage stakeholders at conceptual stage to ensure support for and ownership of the program. Stakeholders could include, but not be limited to: consumers (including disadvantaged groups), expert clinicians in the diagnosis and treatment of the disease, GPs, nurses, community health workers, jurisdictional representatives, epidemiologists, relevant professional college representatives, experts in program management and data collection.
- Obtain consensus on program design.
- Develop processes for the co-ordination of care for people with screen detected abnormalities.
- Ensure there is efficient use of resources.
- Ensure that equity of access and outcome is considered and incorporated into the design of any program.

3. QUALITY MANAGEMENT PLAN

A quality management plan must include evidence based systems and processes for quality management and monitoring, including:

- Develop standards.
- Develop performance measures.
- Develop data dictionary.
- Develop quality assurance processes which are applicable to all elements of the program.
- Develop accreditation processes as required.
- Develop risk management plan.
- Ensure screening program is safe for participants both physically and psychosocially.

- Support ongoing professional development and training to support and sustain the workforce.
- Ensure adequate and realistic funding allocations to achieve objectives – short, medium and long term.
- Ensure equity and consistency of service regardless of regional, rural or remote status.

4. GOVERNANCE AND MANAGEMENT

- Clearly define leadership, advisory and decision making processes.
- Establish management structures at national, state and territory and service level.
- Ability to sustain program and workforce over the life of the program.

5. MONITORING, EVALUATION AND REVIEW

- Develop a formal approach for the ongoing monitoring and evaluation of the screening program.
- Identify appropriate measurable indicators for which data is to be collected to monitor the success of the screening program.
- Develop indicators which enable comparison over years and between international programs if appropriate.
- Develop clear, nationally consistent methods for reporting and collecting data under the indicators.
- Identify reporting milestones (e.g. annually, or related to stages of screening rollout).
- Public reporting should be easily accessible and meaningful for target audiences.
- Ensure monitoring and evaluation are aligned with the quality management plan.
- Identify timeframes or circumstances that would necessitate program review or re-orientation.

GLOSSARY

Assessment:

A follow up test for those people identified with a positive or abnormal screening test. The follow up test determines whether they have the disease or risk marker.

Disease:

A pathological condition of a part, organ, or system of the body resulting from various causes, including infection, genetic defect, or environmental factor, and characterised by an identifiable group of signs or symptoms.

Genes:

Are the basic biological units of heredity. Genes are hereditary units consisting of a sequence of deoxyribonucleic acid (DNA) that occupies a specific location on a chromosome that determines the characteristics of the human body and controls its growth and function. Genes are mainly inherited on chromosomes from a person's parents, but in some circumstances genes can be changed by mutations caused by circumstances such as environmental factors.

Genetics:

The study of heredity that examines single genes and how their function and composition can affect growth and development.

Genomics:

The study of multiple genes and their relationship with one another.

High risk:

Those identified as having significant risk factors for the disease, such as strong family history, identified genetic markers.

Negative predictive value:

The extent to which subjects are free of the disease in those that give a negative screening test result.

Opportunistic case-finding:

The offering of a test for an unsuspected disease when a person presents to a health care practitioner for reasons unrelated to that disease. (modified from Wald, 1994).

Positive predictive value:

The extent to which subjects have the disease in those that give a positive screening test result.

Public health genomics:

Is about effective and responsible translation of genome-based knowledge into public health policy and health services.

Risk marker:

An anatomical, physiological, biochemical or pathological characteristic that indicates a high risk of developing a disease in the future.

Screening:

The presumptive identification of unrecognised disease or defects by means of tests, examinations, or other procedures (modified from WHO, 1968).

Screening pathway:

The screening pathway includes all activities from identification of the target population to diagnosis. It includes invitation, having the test, receiving test results, assessment and diagnosis, as well as monitoring, evaluation and quality improvement activities across the pathway.

Screening test false positive:

An individual for whom the screening test is positive but the individual does not have the disease (or risk marker).

Screening test false negative:

An individual for whom the screening test is negative but the individual does have the disease (or risk marker).

Population-based screening program:

Is where a screening test is offered systematically to all individuals in the defined target group within a framework of agreed policy, protocols, quality management, monitoring, evaluation and review. Population based screening is an organised integrated process where all activities along the screening pathway are planned, coordinated, monitored and evaluated through a quality improvement framework. All of these activities must be resourced adequately to ensure benefits are maximised.

Screening test:

A comparatively simple investigation of anatomy, physiology, biochemistry or pathology that is able to classify people according to their likelihood of having a particular disease or risk marker for a disease.

Sensitivity:

The effectiveness of a test in detecting disease in those that have the disease.

Specificity:

The extent to which a test gives negative results in those that are free of the disease.

Surveillance:

Where people who currently have no symptoms of a particular disease but who are at increased risk of developing the disease due to family history (or other factors), are monitored with a medical test or procedure.

Target population:

The population group that is identified to participate in the screening program.

REFERENCES

Andermann, A, Blancquaert, I., Beauchamp, S., Dery, V. 2008. *Revisiting Wilson and Jungner in the genomic age: a review of screening criteria over the past 40 years*. Bulletin of the WHO, 86: 241-320.

Beaglehole R, Bonita R and Kjellstrom T, 1993, *Basic Epidemiology*. Geneva, WHO.

Centers for Disease Control and Prevention, *Public Health Genomics*, Viewed 27 January 2016, from: <http://www.cdc.gov/genomics/gtesting/index.htm>

Elwood JM, 1990, *Screening programmes in disease control*. In McNeil JJ, King RWF, Jennings GL, Powles JW, eds. *A Textbook of Preventive Medicine*, pp 23-44. Melbourne: Edward Arnold.

Gray JAM, 2004, *New concepts in screening*, British Journal of General Practice, Vol 54, pp292-298.

Holland WW and Stewart S, 2005, *Screening in Disease Prevention*, Oxon, Radcliffe.

National Health Committee, 2003, *Screening to Improve Health in New Zealand*, Wellington

National Health Priority Action Council, *National Service Improvement Framework for Cancer*.

National Screening Committee. 2000. *Second Report of the UK National Screening Committee*. Department of Health, The Scottish Executive, Department of Health and Social Services and Public Safety, Northern Ireland and the National Assembly for Wales. [Second report of the UK National Screening Committee](#).

Strong K, Wald N, Miller A, Alwan A, on behalf of the WHO Consultation Group, 2005, Current concepts in screening for noncommunicable disease: *World Health Organization Consultation Group Report on methodology of noncommunicable disease screening*, Journal of Medical Screening, Vol 12, No 1, p 12-19.

The Cancer Council Australia, *National Cancer Prevention Policy 2004-06*.

United States Preventive Services Task Force, 1996, *Guide to Clinical Preventive Services*, 2nd Edition, Baltimore, Williams and Wilkins.

Wald N, 1994, "Opportunistic screening", Journal of Medical Screening, 1:208

Wilson JMG, Jungner G., 1968, *Principles and practice of screening for disease*. Public Health Paper, Number 34. Geneva: WHO.

World Health Organisation, 2002, *National Cancer Control Programmes: policies and managerial guidelines*, 2nd Edition, Geneva WHO.

World Health Organisation, 2008, *Programs and Projects, Cancer Screening and Early Detection of Cancer*

Bulletin of the World Health Organization, April 2008, *Revisiting Wilson and Jungner in the genomic age: a review of screening criteria over the past 40 years*, Andermann, Blancquaert, Beauchamp & Dery.

ACKNOWLEDGEMENTS

Former Policy Review and New Technologies Working Group of the Former Australian Screening Advisory Committee

Screening Subcommittee of the Australian Population Health Development Principal Committee

Expert Stakeholders

Standing Committee on Screening of the Community Care and Population Health Principal Committee

Australian Government, Department of Health, Screening Policy Section

State and Territory Health Departments