National Screening Advisory Committee (NSAC) National Screening Unit (NSU)		
Minutes Wednesday 27 July 2022		
Venue	Mercy Centre, Wellington and by video conference	
Start Time	10:00	
NSAC members present	Dr Carol Atmore Dr Karen Bartholomew Sheila Beckers Professor Barry Borman Stephanie Chapman Gerardine Clifford-Lidstone Pania Coote (Chair) Professor Mark Elwood John Forman Dr Gary Jackson Dr Kate Neas Dr Pat Tuohy	
Other attendees	**NSU** Dr Rebekah Roos Dr Dougal Thorburn	**Auckland Starship Children's Hospital** Dr Gina O'Grady
Apologies	Professor John McMillan Dr Jane O'Hallahan (Deputy Chair) Dr Nina Scott	

Item	Subject and summary
1.	**Welcome, apologies and introductions** The Chair opened with a karakia timatanga and welcomed attendees. Apologies were received from Prof John McMillan, Dr Jane O'Hallahan and Dr Nina Scott. A round of introductions was given. **Declaration of conflicts of interest** Conflict of interest register was tabled. **Meeting minutes and discussion arising** • The October 2021 meeting minutes were circulated to NSAC members in November 2021 and subsequently published on the NSU website. *Breast cancer screening* The November 2021 minutes included NSAC's consideration of age-extension for women aged 70-74 years. NSAC concluded the evidence for screening women aged 70-74 years should be framed favourably. This was incorrect because concerns were raised. At this July meeting, NSAC members reiterated their concerns around the potential impact of age extension on equity, particularly the risk of widening the equity gap for Māori and Pacific women; and the demands on the BSA programme as it attempts to catch up screening volumes in the face of COVID pandemic impacts.
2.	**Record of NSAC decisions tabulated** A table of NSAC decisions was provided to attendees. NSAC members noted that it has been six years since non-invasive prenatal screening (NIPS) was favourably reviewed by NSAC and advanced for consultation on its addition to the antenatal programme as a contingency test. NSAC members requested that the NSAC work programme also be circulated via email. **Action:** Dr Karen Bartholomew requested that the following items be considered for NSAC's future work programme: newborn screening for Butyrylcholinesterase as a biomarker for SIDS, oral cancer screening and antenatal infectious disease screening. **Action:** Dr Pat Tuohy asked if NSAC could have an update on the vision and hearing component of the Well Child review from the Child and Community Health team at the next meeting in November.
3.	**Spinal Muscular Atrophy (SMA) Screening** Dr Gina O'Grady (paediatric neurologist, Starship Children's Health) presented a review of the evidence for adding SMA screening to New Zealand's Newborn Metabolic Screening Programme. **SMA is the most common genetic cause of infant mortality** SMA is a rare progressive neuromuscular disease caused by the homozygous deletion of the SMN1 gene, leading to an SMN protein deficiency. This causes loss of motor neurons in the spinal cord, resulting in muscle atrophy, weakness, and paralysis. Life expectancy is less than two years in the most severe form of SMA (type 1) accounting for approximately 60% of cases. Most children with type 2 survive into adulthood. Types 3 and 4 do not usually affect life expectancy. Disease severity is modified by the SMN2 gene – the more copies of SMN2 a person has, the milder their SMA phenotype (SMN2 genes also produce the mRNA transcriptase required to make normal SMN protein, but in smaller amounts).

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	The recent advent of disease modifying treatments has provided impetus for some countries to introduce newborn screening for SMA, so that they can identify affected children in the pre-symptomatic stage. There is good evidence of better treatment outcomes with early intervention.
	
	Preliminary assessment of SMA newborn screening against NZ's screening criteria:
	
	Suitable candidate for screening?
	
	• Important health problem – most common genetic cause of death in infancy, high morbidity and mortality
	• Asymptomatic stage and natural history are relatively well understood
	• There is a recognised disease marker: exon 7 deletion in SMN1 gene
	• All ethnicities are affected, however numbers are very small.
	
	Suitable test?
	
	• Screening for SMA on dried blood spot card
	• Well established diagnostic genetic test detects exon 7 deletion in SMN1 gene (both alleles)
	• Analysis can be paired with existing PCR testing for SCID
	• Low incidence of false positives and false negatives
	• Two tier screening – SMN1 deletion and SMN2 copy number testing
	• Paediatric neuromuscular specialist review and clinical diagnosis also required.
	
	However, there is some complexity of SMN2 copy number and relationship with phenotype and disease progression.
	
	Effective and accessible treatment or intervention?
	
	• Novel therapies show promise in improving motor function especially for pre-symptomatic individuals
	• Three approved and accepted treatments:
	  • Nusinersen (Spinraza) – intrathecal splice modifying agent
	  • Zolgensma – single dose gene therapy
	  • Risdiplam (Everysdi) – oral splice modifying agent.
	• Treatments are expensive (either one-off cost of gene therapy or annual costs of disease-modifying treatments).
	
	SMA therapies are not currently publicly funded in New Zealand (nusinersen and risdiplan are registered with Medsafe but remain on Pharmac's "options for investment" list).
	
	The long-term outcomes of new treatments are unclear, as are long-term adverse events.
	
	High quality evidence that screening is effective?
	
	There is good international evidence that screening is effective. However, there are small numbers and short-term follow up only.
	
	Do benefits outweigh harms?
	
	Nusinersen and gene therapy show significant improvements in the short-term for pre-symptomatic cases, less so in early symptomatic cases. As noted above the long-term outcomes of new treatments are unclear, as are long-term adverse events.
	
	Health system capable of supporting the screening pathway?
	
	Facilities for the diagnosis and treatment of SMA are available in New Zealand. There is sufficient expertise (although somewhat limited to Auckland – geographical access is difficult). Multi-disciplinary teams required for ongoing supportive care even with novel therapies. However, the small number of specialists involved improves interdisciplinary working.

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	Consideration of social and ethical issues? There is limited New Zealand data (small numbers), but available incidence and prevalence data suggests that SMA affects all ethnicities – no marked ethnic differences. Newborn screening for SMA in the absence of a publicly funded novel treatment creates an ethical issue – other countries that have introduced SMA newborn screening have done so as publicly funded disease-modifying treatments have become available. Ethical concerns remain even for countries with publicly funded treatment eg, clinically challenging nature of treatment (nusinersen requires repeated spinal injections). Also, uncertainties around how patients with mild forms of SMA will respond to treatment (but most countries initially only fund treatment for those with 1-3 copies of SMN2). In the absence of publicly funded treatment, a number of families with an infant affected by SMA move overseas, mostly to Australia, to access treatment. Māori and Pacific patients are less likely to move overseas. *Consideration of cost-benefit?* Costs of novel treatments for SMA are high. The cost of implementation and ongoing screening for SMA in New Zealand is likely to be marginal compared with the actual cost of treatment. Pharmac's cost utility analysis for nusinersen concluded that: ..*while nusinersen would provide substantive incremental benefits of current treatment… the very high cost of treatment resulted in cost-effectiveness for all models to be considerably lower than what Pharmac would historically consider to be good value.* However, Pharmac's actual cost-effectiveness figures are not available. SMA screening meets most of the New Zealand criteria for screening, however, evidence gaps remain, particularly with respect to cost-effectiveness. **Overseas recommendations** • UK National Screening Committee does not currently support a national newborn screening programme for SMA (next SMA review expected during 2022) • USA added SMA to its recommended uniform screening panel in 2018. As of June 2021, 34 State newborn screening programmes were fully implemented and eight States were progressing implementation • Australia – NSW and ACT pilot programme is now a permanent programme. Department of Health has encouraged other states to implement screening. • Other countries with newborn screening programmes include: Taiwan, Canada, Japan, Germany, Italy. **Conclusion** The approval of novel SMA treatments internationally has changed the landscape in terms of expectations of improved outcomes. However, these treatments are enormously expensive and clinically challenging. Pharmac's Advisory Committees have recommended the disease modifying drugs nusinersen and risdiplam for treatment of SMA. However, the drugs are currently unfunded and remain on the "options for investment" list. Pharmac's 2020 assessment of nusinersen anticipates that newborn screening would be required if this therapy was funded, enabling diagnosis before symptoms develop to improve treatment effectiveness. (In addition to Dr O'Grady's presentation to NSAC members, a full memo on Screening for SMA was provided to NSAC members prior to the meeting for consideration).

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	NSAC discussion included: • Whether there is a possibility of over-treatment for people who may not develop symptoms until adulthood. Dr O'Grady advised that less than one percent of people would fall into this category • Gene therapy as a treatment could be more cost-effective as it is a one-off, compared to the other treatments that are ongoing. Dr O'Grady advised that gene-therapy tends to only be used for the more severe type 1 SMA • It was noted that pre-conception testing for the SMA carrier state in parents is currently a recommendation of RANZCOG. Pre-conception testing could reduce the number of babies being born with SMA. However not all population groups access pre-conception testing • It was also noted that SMA is a more clinically significant condition than Down Syndrome (in terms of prognosis) and Down Syndrome is currently part of antenatal screening • What genetic information is revealed from a positive screen? Dr O'Grady explained that because the test only looks for Exon 7 in the SMA gene it does not identify carrier status • Is SMA screening any different in kind from PKU screening? Dr O'Grady pointed out that conditions currently included in newborn blood spot screening (including PKU) are all genetic conditions so SMA is no different in this respect • Is there enough capacity for genetic counselling associated with screening for SMA? Dr O'Grady noted that even without newborn screening for SMA, affected children would still be identified through clinical presentation, therefore SMA screening is unlikely to significantly increase the workload of genetic counsellors, only the timing of genetic counselling would shift • Pharmac's decision to fund SMA treatments is likely to assume that pre-symptomatic newborn screening is available nationally to maximise the benefit of early treatment • There could be some limited benefits to screening even without treatment eg, giving families information for subsequent pregnancies and reducing the 'diagnostic odyssey' families describe when they face delays in diagnosis. However, screening for SMA in the absence of publicly funded treatments creates a significant ethical issue. **NSAC conclusions regarding SMA screening:** • NSAC considers there to be an optimal and synergistic profile for the implementation of a SMA newborn screening programme and treatment options. Noting that it is a rare condition, NSAC considers SMA to have met most of the screening criteria, with further work on cost-effectiveness needed. • NSAC recommends the progression of development of a screening pathway, with implementation dependent upon treatment availability. In addition, any programme will need to monitor the long-term outcomes of treatment. • NSAC recommends that further work on the cost-effectiveness of SMA screening in the New Zealand setting also be carried out. **Next steps:** The NSU will: • further consider newborn screening for SMA against the National Health Committee screening criteria • consider the implications for ADHB laboratory services regarding potential addition of SMA screening to the Newborn Metabolic Screening Programme, including timeframes necessary for them to plan and implement such screening, and the estimated costs of screening using the same assay platform as SCID • maintain contact with Pharmac regarding developments around public funding for SMA treatments • maintain a watching brief of international newborn SMA screening developments.

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4.	**Māori Monitoring and Equity Group** Pania Coote (Chair) gave an update on the work being undertaken by the Māori Monitoring and Equity Group (MMEG). • MMEG has been focused on the BreastScreen Aoteoroa review. • A Breast Screening Advisory Group has been established which includes two MMEG members. • MMEG recently reviewed the breast screening workforce, acknowledging that there are workforce shortages in the screening sector and that there is the potential to tap into the covid workforce. Further discussion was carried over into item 6 below.
5.	**Health Structure Reforms** Stephanie Chapman (Group Manager, National Screening Unit) gave a brief update on the Health Structure reforms in relation to NSU: • NSU is now part of the National Public Health Service and Te Whatu Ora (Health New Zealand). • NSU has been moved in its entirety and has not been split up into different functions. • NSU now sits in the same organisation as some of the screening providers. • There are advantages and opportunities associated with being joined up with the rest of the health service. • NSU continues to focus on the large projects which need to be delivered as well as progressing our Te Tiriti and Equity Strategy.
6.	**NSU Te Tiriti o Waitangi and Equity Strategy Plan** Dr Dougal Thorburn (NSU, Public Health Physician) gave an update to NSAC members on progress of the NSU's Te Tiriti o Waitangi and Equity Strategic Plan. Dougal had previously sought advice from NSAC at the May 2021 NSAC meeting. Prior to his presentation, NSAC members introduced themselves to Dr Thorburn and indicated what they would like to learn from his presentation. NSAC members expressed an interest in discussing: • iwi partnerships and how to manage them (locally and nationally) • relationship with Te Aka Whai Ora • how health system reforms will influence working with Māori • how does NSAC relate to operational issues of NSU? • should NSAC focus on existing or new screening programmes • integration of cancer screening programmes with cancer services • what does a Treaty partnership look like? • making sure that changes to health system results in improved outcomes – don't want to see things stay the same • how will primary care fit into the new system and the need for connected systems. Dr Thorburn identified the key frameworks which should guide the work of NSU, including He Korowai Oranga and Whakamaua: Māori Health Action Plan 2020-2025. The activities of NSU should also align with the Principles of Te Tiriti o Waitangi: Partnership, Options, Tino-Rangatiratanga, Active Protection and Equity. NSU also want to align their work with the Principles of Māori Data Sovereignty and Māori Research Ethics including Te Ara Tika and with the NSU Quality Principles (Principle 3: National screening programmes will achieve equitable access and outcomes for all population groups).

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	Dr Thorburn gave an update on the NSU's Te Tiriti and Equity journey, which included a series of workshops followed by a whole day wānanga in August 2021. At the wānanga, NSU staff agreed on a NSU vision to: • *provide high quality, equitable and mana-enhancing national screening programmes for people and their whanau.* They also agreed on five commitments (goals) around each of the five principles of Te Tiriti o Waitangi. In order to meet these goals NSU will need to develop an operating model which will in turn need to consider governance and leadership of the NSU. MMEG was contracted to lead a project to develop a governance model for NSU that aligns with Te Tiriti o Waitangi. MMEG and NSU held a hui on 9th June 2022 where Gabrielle Baker (Baker Consulting) presented the project findings. • A wide range of stakeholders were consulted as part of the project including National Kaitiaki Group (NSU), Hei Āhuru Mōwai, Mana Wahine, Māori Women's Welfare League, Data groups (eg, Te Mana Raraunga), Tāngata whaikaha rōpū and a selection of Māori Health Providers. • The project found very few existing co-governance models. Those that exist tend to be from the community and voluntary sectors. • The two-house model has been used in the health sector and recognises the spaces held by Tangata Whenua and Tangata Tiriti and the shared space between them. • Three options for governance were presented to MMEG, ranging from improving the existing system, increasing power sharing and through to large-scale change. • MMEG and NSU members present at the hui expressed their preference for a significant change in governance arrangements in order to meet the NSU's Te Tiriti and Equity goals. ***NSAC discussion included:*** • Te Whatu Ora and Te Aka Whai Ora are using a double-hulled waka approach (waka hourua) to deliver improved health outcomes for Māori • the challenge of liaising with iwi locally to deliver national screening programmes • at a national level NSU can work with Māori organisations that are also national level eg, Hei Āhuru Mōwai • localities have iwi partnership boards that could guide/influence screening locally • a whole system approach to screening pathways is needed that is connected to primary/community care providers. GPs have a responsibility to make sure screening programmes are successful. NSAC has a role in asking what can be done in screening to improve Māori health outcomes in relation to both existing screening programmes and potential future screening programmes. NSAC members thanked Dr Thorburn for his presentation and acknowledged the significant work he has carried out in this area. **Action:** the final report *Te Tiriti o Waitangi, (Co-) Governance and the National Screening Unit* by Baker Consulting will be emailed to NSAC members.
7.	**Programme Updates** Stephanie Chapman gave a brief update on the current priorities for NSU and progress being made in the following areas: • Cervical screening – low cost and free screening for priority groups is available in some settings eg, through support to screening services. Priority groups include Māori and Pacific and unscreened or under-screened women.

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	• Bowel screening – national roll-out was completed in May 2020. The government has also announced a lower age range (from 50 years) for Māori and Pacific from 2023. Evaluative implementation of the lowered age will be carried out in Tairawhiti and Waikato • The NSU has restarted work to consider the benefits, implementation options and costs of introducing non-invasive prenatal screening (NIPS) into the publicly funded screening pathway for Down syndrome and other conditions. A refresh of the cost-effectiveness analysis done in 2015/16 is currently being undertaken • The national screening solution (IT system) for bowel screening will also be used for cervical screening and eventually, breast screening.
8.	**Other business** **Covid recovery plan** • The NSU's covid recovery roadmap and activities was shared with NSAC members. The roadmap aims to promote screening sector recovery and catch up on screening coverage backlog, with a specific focus on equity. The roadmap outlines activities to support recovery in BreastScreen Aoteoroa and the National Cervical Screening Program. **The goals for the BreastScreen Aotearoa Recovery Plan are:** • Return to pre COVID-19 breast screening volumes, adjusted for population growth. • Screening coverage target is reached or exceeded for all ethnicities. • Programme target for rescreening period reached or exceeded for all ethnicities. • Equity gap closes and does not return to pre-COVID-19 levels. **The NCSP aims to achieve a phased recovery with the following goals:** • Phase one (July 2022 to June 2023): Plan for and return to pre-COVID-19 coverage levels for all regions prioritising equity. The NCSP will use the interim recovery plan to achieve this level of coverage to ensure the programme is in good stead to implement HPV primary screening for July 2023. • Phase two (July 2023 onwards): Implementation of the HPV Primary Screening programme. The programme aims to achieve coverage targets and equity in cervical screening within five years. Dr Karen Bartholomew noted that for BSA, goals one and two did not align with each other. Stephanie Chapman responded that equity comes first in the recovery roadmap and that the aim is to reach the coverage target for all ethnicities, or to meet pre-covid levels, whichever is higher. **Action:** NSAC requested an update on the covid recovery at the next NSAC meeting.
	Meeting dates: Next meeting: Wednesday 23 November 2022 Meeting closed at 15:45hrs.