National Screening Advisory Committee

National Screening Advisory Committee (NSAC)
Note of the meeting held on 20 October 2023

Members present:

- Dr Suzanne Crowe (Chair)
- Dr Ellen Crushell
- Professor Andrew Green
- Ms Dolores Keane
- Ms Jillian van Turnhout
- Dr James O'Mahony
- Dr Velma Harkins
- Dr Alissa Connors
- Dr Irene Regan
- Ms Hilary McGouran
- Dr Mary Codd
- Professor Sheelagh McGuinness
- Ms Lora Ruth Wogu
- Dr Susan Kent
- Dr Aoife Doyle

Secretariat:

- Inese Freimane, Population Health Screening Unit
- Luke Crealy, Population Health Screening Unit
- James Scully, Population Health Screening Unit

Department of Health:

- Joanne Uí Chrualaoich, Population Health Screening Unit

Health Information and Quality Authority (HIQA):

- Dr Máirín Ryan, Deputy Chief Executive and Director of Health Technology Assessment (HTA)
- Dr Susan Spillane, Head of Assessment, Health Technology Assessment (HTA)
- Dr Helen O'Donnell, Senior Health Technology Assessment (HTA) Analyst in attendance for agenda item 4

Member Apologies:

- Professor Ciaran O'Neill
- Dr Deirdre Murray
- Dr Paul Kavanagh
- Dr Jennifer McSharry
- Professor Michael Rigby

1. Welcome & Introductions

The Chair welcomed everyone to the meeting and invited attendees to introduce themselves. The meeting marked the first for Dr Suzanne Crowe as the Chair of NSAC, following her appointment by Minister Donnelly in July 2023.

The Chair noted that this was the last meeting for several members, including Dr Mary Codd, Prof. Sheelagh McGuinness, Ms Jillian van Turnhout, and Dr Ciaran O'Neill who were concluding their terms on the Committee. The Chair was joined by the other NSAC members present in expressing their gratitude to each of the outgoing members for their time and contributions to the work of the Committee.

Conflict of Interest

The Chair reminded members of the need to declare any potential conflicts of interest. There were no new conflicts of interests declared.

Minutes of the 25 May 2023 meeting

The minutes of the 25 May 2023 meeting were circulated by the Secretariat to Committee members prior to the meeting. The Chair sought any matters of accuracy on the draft document. None were identified and the minutes were approved.

Matters arising

N/A

2. Revised Standing Orders, Terms of Reference and NSAC Criteria

The Chair highlighted to members that at the meeting on 25 May 2023, the Committee endorsed the revised Standing Orders, excluding the section regarding observers.

The Chair noted that by the end of 2023 the Committee will have undergone a significant refresh of its membership with four new members appointed in January 2023, a new Chair appointed during the summer and a number of members reaching the end of their terms on the Committee. It was suggested that these changes provide the Committee with an opportunity to reconsider the role of observers. Therefore, the Chair decided to pause the attendance of observers at Committee meetings until a review of Standing Orders is complete. Members voiced their support for this approach and noted that to fully utilise the role of observers, their purpose and role needs to be clear. It was noted that the current NSAC observer was advised of this situation and will be updated as necessary.

Decision:

The Committee decided that it would review its list of stakeholders and identify stakeholders who may be invited to attend future NSAC meetings on a rolling basis. The Committee decided that the text in Section 8 of the Standing Orders needs to be updated to provide a clear description of the role and purpose of observers who attend NSAC meetings.

Action: The Secretariat to prepare a list of stakeholders for the observer role for the Committee to review. The Secretariat to re-write Section 8 (observers) of the Standing Orders to outline the role of observers.

NSAC Criteria and Terms of Reference

In light of the updated European Council Recommendation on cancer screening, the Committee was asked to decide whether it may wish to consider expanding the remit to include additional methods, such as targeted or risk-based screening. It is also suggested that any advice issued is reviewed routinely every three years, or beforehand, if significant new evidence becomes available.

Decision: The Committee agreed in principle that expansion of NSAC's remit to include additional methods, such as targeted or risk-based screening should be seen as a natural progression for the Committee. However, this matter would require further consideration at the next meeting.

Action: Secretariat to draft a summary paper on common approaches used in international screening programmes and contextual factors, including the 2022 EU Cancer Screening Recommendation, NSAC Bill and NSAC founding principles.

3. Updated European Council Recommendation on cancer screening

The Secretariat updated the Committee on the recent engagement by the Department of Health with its European colleagues on the updated European Council Recommendation on cancer screening and the pilots that are underway in Ireland. Officials from the Department of Health attended two European-level meetings where updates were provided on the processes surrounding the Recommendation.

The Committee was informed that arising out of the updated Recommendation, efforts are now underway to establish a 4-year Joint Action on cancer screening, titled *EUCanScreen*, funded under the European Commission's *EU4Health* programme. The Joint Actions are commonly used in a European context to coordinate an operational action by the member states within the framework of the common foreign and security policy. In this case, 31 countries (all EU member states plus Norway, Ukraine, Moldova and Iceland) will be examining topics ranging from the sustainability of cancer screening programmes to the engagement/participation of minority or vulnerable groups in screening.

Subject to approval of the application by the Health and Digital Executive Agency (HaDEA), the Department of Health, in partnership with the National Screening Service, Royal College of Surgeons Ireland and the International Agency for Research on Cancer (IARC), will be involved across a wide range of tasks in the project. It is hoped that Ireland will be able to gain valuable experience through the Joint Action and share learnings of Ireland's recent progress in cancer screening.

An update was also provided on three EU-led pilot projects currently underway on lung, gastric and prostate cancer screening. These pilots are in the early stages and the Secretariat is in regular contact with the Irish-based principal investigators involved.

The Committee welcomed the update on the European Council Recommendation and the pilot projects. It was requested that the Department of Health continues to engage with European and national colleagues, providing updates to the Committee as necessary.

A draft concept note was presented to the Committee by the Secretariat, outlining plans to convene a cancer screening subgroup to assist in the consideration of the implementation of the updated Recommendation in Ireland. This note was developed following previous discussions at the May meeting, including the need for dialogue with key stakeholders and collaboration with European colleagues.

Attendees generally reacted positively to the contents of the paper, with some indicating their interest in joining and contributing to such a subgroup. Some members asked for clarification around the reporting structure back to NSAC, which the Department is considering.

The Secretariat will revise the paper, based on feedback provided by Committee members and circulate in advance of the December meeting.

4. Expansion of the Newborn Bloodspot Screening (NBS) Programme

a) Presentation of the Health Technology Assessment (HTA) of the addition of Spinal Muscular Atrophy (SMA) to the National Newborn Bloodspot Screening Programme (NBS).

Dr Helen O'Donnell, Senior Health Technology Assessment (HTA) Analyst in HIQA, provided a detailed presentation on the Health Technology Assessment (HTA) for the addition of Spinal Muscular Atrophy (SMA) to the National Newborn Bloodspot Screening Programme (NBS) in Ireland.

Following the presentation, the Committee thanked Dr Helen O'Donnel and the team in HIQA for the comprehensive HTA report. The Chair then invited members to discuss which resulted in the unanimous decision by NSAC members present to make a recommendation to the Minister for Health for the addition of Spinal Muscular Atrophy (SMA) to the NBS Programme.

In making this recommendation and in line with the HIQA HTA report, the Committee raised several important factors that would need to be carefully considered in advance of the SMA being added to the NBS Programme:

- The Committee noted the severity variance between the SMA subtypes. Screening and associated early treatment are likely to have more significant impacts in terms of outcomes where it detects SMA subtype 1 as compared to subtypes 3 and 4. However, while the tests available identify the number of genetic deletions of the SM1 gene and copies of the SMN2 gene, this does not perfectly correlate with the disease severity. As such, the Committee recommended screening for all SMA subtypes, rather than limiting it examining a specific number of copies.
- The Committee considered it important to highlight that as a rare disease, SMA has associated challenges when it comes to data availability. Data limitations specifically noted in HIQA's HTA included uncertainty around associated drug treatment prices and the small sample sizes used in clinical studies. As a result, it was not possible to conduct a full cost-effectiveness analysis in this case.

- The Committee suggested that in advance of SMA being added to the NBS Programme, clear treatment pathways must be designed and implemented. This was deemed necessary to ensure clarity for patients, parents, and clinical staff. This would likely involve the creation of 'watchful waiting' structures for patients diagnosed with less severe SMA subtypes who often remain asymptomatic until adulthood.

- In its previous recommendation regarding Severe Combined Immunodeficiency (SCID), the Committee noted that the required equipment for that condition is not currently used in Ireland and that significant planning is required to procure and implement the test. Given that screening for SMA involves the same PCR-based technology as screening SCID, similar operational challenges exist, and consideration must be given to the timing of these arrangements, given the planned move of the National Newborn Screening Laboratory to the new children's hospital.

- The Committee noted further expansion of the NBS Programme would require additional budget or a reallocation of resources from other parts of the Health Service.

Decision: The Committee decided to recommend to the Minister for Health that Spinal Muscular Atrophy (SMA) be added to the National Newborn Bloodspot Screening Programme based on the evidence provided in the Health Technology Assessment (HTA) carried out by HIQA.

Action – Chair to write a letter to the Minister for Health recommending the addition of Spinal Muscular Atrophy to the National Newborn Bloodspot Screening Programme.

5. HIQA Work Programme

Dr Máirín Ryan, HIQA, provided an update in relation to the HIQA work programme. While HIQA resources have been focused on completing the Health Technology Assessment (HTA) on Spinal Muscular Atrophy (SMA), attention will now move to progressing other pieces of work.

The review of evidence for an age range expansion in the BowelScreen programme is now underway and work has begun on the requested evidence review for Developmental Dysplasia of the Hip. It was stated that a draft review on this will be presented to the Committee at its December meeting.

Lastly, a HTA to evaluate evidence for a screening programme for Abdominal Aortic Aneurysm (AAA) is due to commence shortly. HIQA committed to providing further updates on their Work Programme at the next NSAC meeting.

6. NSAC Annual Call 2021/2022

Joanne Uí Chrualaoich, Department of Health, presented the Committee with updated information on two conditions, Familial Hypercholesterolemia (FH) and Genetic Hemochromatosis (GH).

Submissions to add both conditions were received as part of the Annual Calls in 2021 and 2022 and have been the focus of discussions at previous NSAC meetings. The Committee was asked to decide on whether these conditions would be suitable for an evidence review by HIQA and if they should be added to HIQA's work programme.

Familial Hypercholesterolemia

The Commmitee received a paper prior to the meeting with information that was collated by the Secretariat. The paper covered several areas, including:

- Impact of not treating FH appropriately
- Types of screening available
- Current Irish/international practice
- Recent developments
- Known limitations and gaps

It was noted that Familial Hypercholesterolemia (FH) screening can be used to identify individuals at high risk of premature cardiovascular disease and that evidence suggests that cascade and population screening methods, along with advancements in genetic testing and risk assessment tools, are leading to improving FH diagnosis and management. However, challenges, including underdiagnosis and the cost of testing, must be addressed to ensure that FH screening reaches its full potential in preventing cardiovascular events.

The Committee's discussions centred around the levels of premature cardiac disease. It was noted that there are existing affordable treatments available for FH. Therefore, screening for FH could potentially have a cost saving effect following implementation. Based on the evidence presented, the Committee agreed to recommend FH for an evidence review based on the information available to the Committee.

Decision: The Committee decided that Familial Hypercholesterolemia (FH) is suitable for an evidence review by HIQA that it should be added to the HIQA work programme.

Action: The Chair to write to HIQA to request that an evidence review of Familial Hypercholesterolemia (FH) is added to the HIQA Work Programme. The Secretariat to update the status of FH on the NSAC work programme and inform those who submitted proposals on FH through the Annual Calls of the Committee's decision.

Genetic Hemochromatosis (GH)

A submission proposing the introduction of population-based screening for Genetic Hemochromatosis (GH) was received through the 2022 Annual Call. The submission was discussed at the 25 May 2023 meeting, and the Secretariat committed to compiling more information for the Committee to consider.

This paper produced by the Secretariat and was sent to Committee members in advance of the meeting. It provided a summary overview of screening for GH, its burden, routes to diagnosis, existing guidelines, treatment options, and the economic costs associated with treatment.

The Committee discussed the content of the paper, and several concerns were noted regarding the suitability of GH for screening. While there were examples of targeted screening in certain countries referenced, such as in the USA, there is currently no evidence of population-based screening taking place internationally.

Another concern raised was that testing only shows genetic predispositions for GH and not the condition itself. As there are high rates of people in Ireland with a genetic predisposition to GH, there would be a risk that population-based screening could result in large numbers of people being identified despite not being at risk of developing the condition.

Additionally, it was highlighted that GH as a condition progresses at a slow rate, with established and effective treatment pathways already available.

Based on the information available, the Committee felt that there was limited evidence to support a recommendation for population-based GH screening to be subject to a full evidence review by HIQA. Instead, it was suggested that more work should be done across the health system to raise awareness amongst the public of the symptoms and risks of Genetic Hemochromatosis.

Decision: The Committee decided not to recommend GH for inclusion in the HIQA work programme. Further consideration of this proposal is deferred, pending significant new evidence emerging.

Action: The Secretariat to update the status of GH on the NSAC work programme and inform those who submitted proposals on GH through the 2022 Annual Call.

7. Communications & Engagement Update

The Secretariat provided an update on recent communications and engagement since the May 2023 meeting. This included the launch of an Expression of Interest process for the Chair of the Committee. A press release announcing the appointment of the new Chair, Dr Crowe was issued on 20 October 2023.

The Secretariat also launched an Expression of Interest for five NSAC vacancies in the areas of Epidemiology, Ethics, Public Voice, Health Economics and Obstetrics. The advertisements for these roles were hosted on both the Stateboards and NSAC websites. The vacancies are being promoted via social media while also featuring in Press Releases for appointment of the new Chair.

The Secretariat noted that they are developing plans for Annual Call 2023, which is expected to be launched by the end of the year.

8. Administrative

Ethics Framework

The Request for Quotes process to procure an expert to develop an ethics framework for the Committee closed on 15 September. The Department of Health expects to evaluate responses received and conclude the procurement process before the next NSAC meeting.

The Chair thanked attendees and closed the meeting.

The next meeting of the Committee is scheduled for 8 December 2023.

National Screening Advisory Committee

National Screening Advisory Committee (NSAC)
Chair's Actions
Following the 20 October 2023 meeting
Notification of Chair's action on behalf of the NSAC

Action No.	Chair's action	Status
1	Chair to write to the Minister for Health to recommend the addition of Spinal Muscular Atrophy (SMA) to the National Newborn Bloodspot Screening Programme.	Complete
2	Chair to write to HIQA to request that an evidence review of the condition Familial Hypercholesterolemia (FH) is added to the HIQA Work Programme	Complete

I confirm that I have taken the Chair's actions recorded above.

Suzanne Crowe

Dr Suzanne Crowe
Chair, National Screening Advisory Committee

Date: 08 December 2023