Canada's Drug Agency (CDA-AMC) External Consultation: Developing a Process for Horizon Scanning for Emerging Rare Newborn Screening Conditions

Comments from the Institute for Quality and Efficiency in Health Care (IQWiG)

28 November 2025

1. Do you agree with the proposed eligibility criteria for identifying conditions to include in the emerging rare conditions list for newborn screening, as described in Table 1 in the discussion paper? (https://www.cda-amc.ca/news/consultation-discussion-paper-developing-process-horizon-scanning-emerging-rare-newborn)

☐ Yes ☒ Yes, in part ☐ No

Please provide your reasoning and suggestions for how to enhance the criteria. We are interested in hearing your perspective on the scope of the criteria, whether they are too narrow (i.e., may lead to the missed identification of emerging conditions), or too broad (i.e., may lead to diffuse results that may not be applicable for the intended purpose).

Although market approval of a new drug does not necessarily imply an added benefit, it is generally a good concept to use the market approval of new first-in-class drugs for identification of possible new screening conditions. The criteria in Table 1 should nevertheless be refined, as they appear overly broad.

Eligibility criterion 1 addresses the rareness, but not the seriousness of a disease. In our view, seriousness is much more important and should be included in the criterion (i.e., column #1). It appears inadequate to subsume seriousness under "definition" in column #2, since both terms cover two completely different aspects of a disease.

Eligibility criterion 2 encompasses the availability of "an early intervention". Screening for a disease, however, will only be beneficial, if the intervention is clearly more effective in asymptomatic than in symptomatic stages of the disease. Otherwise, it would be sufficient to wait until children develop symptoms and treat them. Thus, the definition in column #2 should be rephrased as follows: "Early intervention is defined as an effective treatment that requires to be initiated before symptoms develop." And it is not necessary here to focus on the early life phase, because this is already included in criterion 1.

2. Do you agree with the proposed process to identify emerging rare conditions to be monitored, as described in Figure 1 in the discussion paper?

☐ Yes ☒ Yes, in part ☐ No

Please provide your reasoning and suggestions for how to enhance the process so it meets the objective of creating an inclusive, broad and relevant list of emerging rare conditions for monitoring.

The sequence of steps described in Figure 1 should be reordered. The key selection criterion for a possible new screening program is the availability of an effective early intervention. Therefore, this step should be the first, rather than the second one. If the process starts with the identification of "potentially eligible conditions" and any rare disease of the newborn qualifies, the likely result would be a list of hundreds of rare diseases. It appears more efficient to monitor the availability of new drugs.

We recommend that regulators, such as Canada's Drug Agency, use their influence during the licensing process of new drugs to allow early identification of newly treatable rare conditions. If drug manufacturers are required to describe in detail how patients were identified for their clinical studies on the new substance, this would make it easier not only to identify such conditions, but also to learn more about the diagnostic options.

3.1 Are you aware of any additional emerging rare conditions not identified on the proposed sample list of emerging rare conditions that could meet the eligibility criteria? If so, please provide the name of the condition and any additional information supporting your reasoning on its potential eligibility.

We are not aware of any additional rare condition that could be included in Canada's newborn screening program. IQWiG is currently assessing screening for metachromatic leukodystrophy (MLD), but CDA is already working on this topic, too.

3.2 Are you aware of any therapies in the pipeline related to the sample list of emerging conditions, or the condition you recommended in question 3.1? If so, please describe the therapy and provide any additional information on its developmental stage and if it has received market approval in any country, if the information is available.

No, we are not aware of any such new therapy. We recommend to screen the lists of applications submitted for marketing authorisation of medicines in Europe. These lists are published by EMA (European Medicines Agency) on a monthly basis (https://www.ema.europa.eu/en/medicines/medicines-human-use-under-evaluation).

4. Do you agree with the process for monitoring and maintaining the emerging rare conditions list over time?

☒ Yes ☐ Yes, in part ☐ No

Please provide your reasoning and suggestions for how to enhance the process of updating the list to provide timely and relevant information.

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5. Recognizing the potential disruption from emerging genomic sequencing technologies on newborn screening, do you have recommendations on how future horizon scanning activities could incorporate genomic sequencing into the process? In particular, in the context of genomic sequencing, we are interested in your thoughts on how to address the potential for an increased volume of conditions for future monitoring and the appropriateness and relevance of the eligibility criteria for the list.

Genome sequencing of newborns is linked to many difficulties, including medical, legal, ethical, social, economic, and organizational ones. For example, screening will also detect mild variants, non-pathogenic variants, and variants of uncertain significance (VUS) of known or yet unknown polygenic conditions. We believe that interdisciplinary approaches and sensitive policy frameworks are essential in this context, as outlined for example by Chetta and colleagues (https://pubmed.ncbi.nlm.nih.gov/40863452/). It still appears very likely that whole-genome sequencing of newborns will gain a role in routine healthcare within the next 10 or 20 years.