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LETTERS

UK National Screening Committee Criteria: clarification of two misunderstandings

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We write on behalf of the UK National Screening Committee (UK NSC) in response to your article.[1]

Any comparison of what screening decisions are made by different jurisdictions is very useful. It is particularly helpful to look at the decision-making processes as they are rarely documented in peer-reviewed literature and your paper casts welcome light on these.

Inevitably, such work will lack a detailed insight into some of the subtleties of the process and so will draw rather broad lines. Your representation of the UK NSC is mostly very insightful, but we would like to draw your readers' attention to two aspects that appear in your paper and do not reflect the real-life complexity.

Your paper concludes 'In general, it seems the UK system has higher standards for evidence before starting screening for a condition by requesting evidence from RCTs, which is also evident in the limited amount of conditions screened for compared with the other Western societies. Additionally, assessment seems to steer away from screening, as recommendations in the UK uniquely included a category on developing clinical guidelines instead of focussing on screening.'

First, the assertion that the UK requires RCT level evidence. Manifestly this is not so. The UK recommends screening for nine bloodspot conditions none of which are underpinned by RCT level evidence. In common with other surveyed authorities, the UK NSC uses the adapted Wilson and Jungner criteria as a framework to guide decisions. The criteria set a high threshold for ideal evidence to inform screening decisions, but for some diseases it is unlikely to be possible to produce such evidence. For example, in case of very rare diseases RCTs would be too large, expensive and long to provide useful information. The UK NSC uses the criteria to allow for an understanding of what would be ideal, but also recognises that in some circumstances that will not be available and so different evidence products are used. The UK NSC review and recommendation statements on the bloodspot conditions show the type of evidence that has informed the actual decision (https://legacyscreening.phe.org.uk/msud). It has recently been noted that decision-making relating to bloodspot screening has been lax.[2] So although it is recognised that RCTs may not be possible the UK NSC is also aware that the difficulty of generating evidence does not negate the difficulty of decision-making without it.

Second, somehow by recommending treatment guidelines the statement 'UK NSC seems to steer away from screening' is simply incorrect. A UK NSC criterion addresses the need for clinical management to be optimised prior to screening. This encourages reflection on the additional impact of screening over and above clinical care, for example, cascade testing in siblings or well-managed protocols for early presenting cases. Without these components the viability of screening programmes for rare diseases cannot be accurately evaluated. Neither can they operate effectively when implemented. The work we do on screening programmes is done hand in glove with clinicians caring for these very rare, very sick babies. We are committed not just to making good screening decisions but also to supporting our clinical partners and stakeholders in care for these babies. To achieve this it is essential that screening is not conceived as taking place in a vacuum.

CONFLICT OF INTEREST

The author declares no conflict of interest.

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1 Jansen ME, Metternick-Jones SC, Lister KJ: International differences in the evaluation of conditions for newborn bloodspot screening: a review of scientific literature and policy documents. *Eur J Hum Genet* 2016; **25**: 10–16.
2 Moyer VA, Calonge N, Teutsch SM, Botkin JR: United States Preventive Services Task Force: Expanding newborn screening: process, policy, and priorities. *Hastings Cent Rep* 2008; **38**: 32–39.

Reply to A Mackie

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On behalf of the authors, I write to reply to the Letter to the Editor from Dr Mackie: UK National Screening Committee criteria: clarification of two misunderstandings, published in this issue of EJHG.

We thank Dr Mackie and her team for taking the time to respond to our work and clarifying the review process from the UK perspective. As Dr Mackie's letter highlights, comparisons of screening decisions are useful, yet hard to construct, since decision processes are rarely documented in peer reviewed literature.[1,2] It is only through much needed international discourse around newborn screening that transparency and learnings across the globe can be achieved. In this instance, while efforts were made to engage programs across the globe to support such a discourse, more was needed in terms of reaching out specifically to the UK National Screening Committee (NSC). To this end, we very much welcome Dr Mackie's correspondence to ensure the UK's program is accurately represented.

The UK NSC publishes extensive and insightful reviews and recommendation statements, and their efforts and successes are admirable. Newborn screening programs are always under development, and