Revista Brasileira de Enfermagem
REBEn

EDITORIAL

Newborn blood spot screening: to expand or not to expand?

Farah Seedat[I], Sian Taylor-Phillips[II]

[I] MSc, PhD student, Division of Health Sciences, Warwick Medical School, University of Warwick, Coventry, UK.
[II] PhD , Assistant Professor, Division of Health Sciences, Warwick Medical School, University of Warwick, Coventry, UK.

How to cite this article:
Sedat F, Taylor-Phillips S. Newborn blood spot screening: to expand or not to expand? Rev Bras Enferm. 2015;68(5):491-2. DOI: http://dx.doi.org/10.1590/0034-7167.2015680501i

"The principal constraint on what we can detect in newborn screening is fast becoming what we choose to detect: rather than what is limited for us by technical feasibility or cost"[1].

Technological advances are providing the medical world an opportunity to extend the number of conditions newborn babies are screened for. Tandem mass spectrometry (TMS, also known as MS-MS) is allowing the possibility to screen for a range of disorders on a large scale. TMS is a reliable technique that allows small molecules in samples of serum or whole blood to be measured in a single step. Therefore, from the few drops of blood on a newborn bloodspot, over 50 conditions can be tested. Indeed, expanding the number of conditions a newborn is screened for using TMS is rapid, accurate and cheap. However there are two sides the argument and there are harms that can occur from the expansion, including an increase in false positive cases, overdiagnosis and overtreatment.

In deciding which conditions the newborn blood spot screening should include, is far from straightforward, and careful consideration is required of the benefits and harms before it is recommended for a population. Questions like what is benefit and harm of informing a parent of a child's condition for which there is no treatment or management, or how useful it is to know ones genetic susceptibility to a condition that may only be relevant in the next 15-20 years, are necessary to consider. Warwick Medical School recently examined the ethical implications of expanding the newborn blood spot in a rapid review, and found that there are important issue to consider in deciding how to assess the benefits and harms of expanding the newborn blood spot[1].

A key issue that emerged in this review was the type of evidence required to inform screening decisions for conditions on the blood spot. In particular, scholars have questioned whether randomised control trial (RCT) evidence is necessary to estimate benefits and harms. Those who argue that RCT evidence is necessary, state that it is the least biased and most accurate method to estimate the balance of benefits and harms from screening. On the other hand, advocates of bloodspot expansion point to the fact that majority of conditions that would be considered for blood spot screening are rare. Rare diseases suffer from a lack of evidence due to the lack of funding. Furthermore, conducting an RCT for rare diseases is not feasible, and probably not possible, due to large sample sizes and long follow up required. Therefore they advocate that alternative types of evidence should suffice, although it is not clear what these are.[1] Both arguments pose important concerns on how to make evidence-based decisions and solutions are desperately required to effectively address this gap.

Another issue of debate is the definition of benefit that is to be assessed for newborn screening. On the one hand, it is stressed that benefit should be restricted to the direct mortality and morbidity benefit to the newborn. On the other, advocates for extending screening also extend the benefit beyond the clinical benefit to the newborn, to the various wider benefits to family members and society. An example of societal benefit would be additional research and knowledge that could be gained. A familial benefit would be the reduction in the "diagnostic odyssey" – the long journey of investigations and referrals before a diagnosis is reached. Opponents argue that because many conditions do not have effective treatment, the diagnostic odyssey may simply shift to a 'treatment odyssey' instead. Nevertheless, advocates argue that early detection may still provide supportive and palliative care benefits increasing quality of life. Having a diagnosis may also have psychological benefits for parents to plan the child's future and have realistic health expectations for them. Conversely, screening and

early detection may reduce the diagnosis-free period in which families can bond. Another benefit identified by advocates is the reproductive risk information that families could gain, through which they could benefit from genetic counseling and family planning. However without perfect parental consent, parents who may not want this risk information may receive it [1].

These advantages are clearly not direct benefits to the newborn baby, and little or no clinical benefit makes effective informed consent even more important in a context where informed consent is already difficult. Parents would have to understand information about a huge number of conditions, shortly after birth, where tests are offered as just another postnatal procedure, therefore true informed choice may not be accomplished[1].

Extending the benefit of screening beyond a mortality and morbidity reduction may also have unintended negative consequences such as rising false positive and indeterminate results, thus rising overdiagnosis, overtreatment, and reducing trust in the screening and the health system. Even where there is no overtreatment, in the case of false positive, parents may continue to be over-mindful and over-protective of children even after disease is ruled out. Finally, carrier status can negatively impact a child's psychology, for example lowing self-esteem or causing stigmatisation[1].

This brief note has highlighted some but not all of the ethical complexities that the rapid review found are associated with expanding the newborn blood spot. The points of contention identified from various perspectives in the literature need to be furthered researched so that we can define alternative but robust evidence methods to assess the benefits and harms of screening, as well as agree on the scope of benefit and harm definitions. The harms of screening and the difficulty in obtaining true informed consent need to be carefully considered. Countries must take care to define the type of benefit and harm, and the type of evidence that they will consider at the outset of evaluating which conditions to include on the newborn bloodspot. While the consequences of these conditions are devastating, we must tread cautiously before veering down the path of screening. In our intention to do well with technological advances, we do not want to unintentionally unleash several, but serious, harms to the population.

REFERENCES

1. Taylor-Phillips S, Boardman F, Seedat F, et al. The Ethical, Social and Legal Issues with Expanding the Newborn Blood Spot Test. Conventry (UK): Warwick Medical School; 2014.