Screening early in life for untreatable conditions

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Executive summary

Health Council of the Netherlands

The blood spot (‘heel prick’) test is used to screen newborns for rare but serious health conditions. Early detection and treatment can significantly improve their health and prevent severe disability or even death. There are now calls to consider screening early in life also for untreatable conditions. The State Secretary for Health, Welfare and Sport (VWS) has requested advice from the Health Council about the desirability of an early-life screening programme for untreatable conditions.
The Council’s Perinatal Screening Committee has taken up this request.

A fundamental principle of newborn screening is that screening should lead to significant improvements in the child’s health. However, early-life screening for untreatable conditions could benefit children and their parents in other ways. First of all, screening reduces the typically long and uncertain period between first symptoms and diagnosis. Reducing the diagnostic delay for rare conditions could alleviate the negative effects that untreatable conditions have on the welfare of children and their parents. Early diagnosis allows for parents to prepare, both practically and emotionally, for life with a child with a serious untreatable condition. Screening can also allow for parents to access services that most parents consider helpful for their children and help them optimize family life decisions. Access to therapeutic interventions also holds promise of physical benefits. Early initiation of supportive treatment, preventive measures or experimental medical interventions could help reduce the risk of physical harm. Another benefit is that screening provides parents and family members information about reproductive risks. A societal benefit of screening is that it increases knowledge about rare conditions.

There are also disadvantages associated with screening for untreatable conditions. Disadvantages are that screening for conditions with a wide variety of phenotypic expression gives children and their parents very little certainty about the severity of symptoms, the age of onset or prognosis. Another disadvantage is that a positive test result for a condition that will manifest later in life deprives children and their parents of the opportunity to spend the initial years of the child’s life relatively carefree. Depriving the child of these ‘golden years’ could be viewed as potentially harmful.
A positive test result could also violate the child’s right to an open future, as important life decisions following the disclosure of personal health information are made by others.
In addition, there are the usual disadvantages of screening, such as the negative consequences of false-positive and false-negative results and incidental findings. These can lead to

unwarranted distress and follow-up diagnostic testing, or conversely convey a false sense of reassurance. A disadvantage for society at large is that screening places a considerable burden on all parties involved in the logistics, information provision and follow-up of screening.

The committee does not perceive any principle objections against screening for untreatable conditions. Given the wide spectrum and diverse characteristics of untreatable conditions, however, the ratio between the benefits and risks of screening will have to be assessed for each condition separately. To this end, the committee established a framework to assess the benefits and risks on a case-by-case basis. Here, the driving principle is that screening should benefit the child.

Based on this framework, the committee has currently been unable to identify any untreatable conditions that could be considered for screening. The majority of untreatable conditions are inappropriate for screening due to either large phenotypic variation, limited knowledge about the specific conditions or lack of a reliable screening test. The committee also anticipates practical difficulties in obtaining the scientific evidence required to substantiate the claim of psychosocial benefits for the child. This lack of evidence is the reason why at this moment no nationwide screening programme for untreatable conditions exists in other countries.

The committee acknowledges that a long quest for the right diagnosis can have detrimental effects on the psychosocial well-being of families. However, what is at stake is whether a nationwide screening programme for untreatable conditions early in life is the solution that is best suited to address this problem.

At this moment in time, the committee is not convinced of the utility and proportionality of such a screening programme. Alternative strategies may be better suited to reducing the negative effects of diagnostic delay for a larger number of families than would be the case with screening for a handful of rare conditions. The committee therefore recommends investing in strategies that are more effective and that are not associated with the disadvantages of nationwide screening. Current improvements in terms of low-threshold referrals and broad diagnostic testing in cases of developmental disability will play an important role.

Over recent years, centres of expertise have been established in academic medical centres in the Netherlands for children with rare conditions and developmental disabilities. These efforts have already led to progress in tackling the issue of diagnostic delay. The committee views that diagnostic delay could be reduced even further through efforts to enhance access to specialty centres for broad follow-up diagnostic testing and by stimulating multidisciplinary collaboration between healthcare providers.

A key aim is that parents know how to quickly find their way to a specialty centre for low-threshold broad diagnostic testing as soon as a child begins to lag in development. Parents can be spared a great deal of suffering if the

concerns they have about their child are acknowledged.

With respect to the goal of providing families with information about reproductive risks, the committee considers preconception screening for carrier status and perhaps prenatal screening as the preferred approaches. Preconception screening for carrier status is currently being explored by the minister. Once developments in drug research and innovation lead to the availability of a therapeutic intervention for a specific condition, it is important that assessing the condition's eligibility for newborn blood spot screening is initiated as quickly as possible.

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The Health Council receives most requests for advice from the Ministers of Health, Welfare and Sport, Infrastructure and Water Management, Social Affairs and Employment, and Agriculture, Nature and Food Quality. The Council can publish advisory reports on its own initiative. It usually does this in order to ask attention for developments or trends that are thought to be relevant to government policy.
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