

Relevance of Neonatal Pulse Oximetry Screening for Critical Congenital Heart Disease

English summary

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SUMMARY

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Health problem

Critical congenital heart disease refers to heart defects resulting from malformation of one or more parts of the heart during the early stages of embryonic development. Critical congenital heart disease results in poor blood flow or decreased blood oxygen levels. This causes the body's organs and tissues to be hypo-oxygenated, which can lead to organ damage and life-threatening complications. Medical attention is needed from the moment of diagnosis. Surgical or catheter-based intervention is required during the first month of life, and lifetime cardiology follow-up is necessary thereafter. The etiology of these defects is multifactorial, most likely linked to environmental factors and sometimes associated with genetic variants. Critical congenital heart disease has a prevalence of 1 to 3 cases per 1,000 live births and accounts for approximately one fourth of all congenital heart disease.

The identification, treatment and clinical outcomes of patients with critical congenital heart disease have improved considerably in the past few decades. Prenatal diagnosis and neonatal screening are two early detection strategies that permit management planning involving the family and healthcare personnel. Despite the use of second-trimester ultrasound for prenatal diagnosis and of the physical examination as detection techniques, a varying proportion of newborns with critical congenital heart disease are not identified before discharge from the hospital or birthing centre. These newborns will then be urgently admitted with an acute condition and often in a state of heart failure, with an increased risk of neurological impairment. Severe neurological impairment reportedly occurs in 5 to 10% of critical congenital heart disease patients, with milder neurological problems occurring in a further 25%, mainly in those with a late diagnosis.

Mandate

The Ministère de la Santé et des Services sociaux (MSSS) asked the Institut national d'excellence en santé et en services sociaux (INESSS) to assess the relevance of neonatal screening for critical congenital heart disease using the pulse oximetry test.

Methodology

To fulfil this mandate, the scientific literature was systematically searched for data on the performance, efficacy and safety of the pulse oximetry test for detecting critical congenital heart disease in newborns. Contextual and experiential data were collected from perinatal professionals, a patient association, and parents or caregivers of children with heart disease. The organizational issues related to the use of the pulse oximetry test (timing of the test, site and algorithm) and the optimal implementation approaches (new

program, existing program or preventive clinical practice) in the Québec context were discussed with experts on an advisory committee and with members of a monitoring committee consisting of representatives from the various organizations and associations concerned. The entirety of the data (scientific, contextual and experiential) was submitted to the Comité d'excellence clinique en dépistage des maladies chroniques for deliberation on the final recommendation.

Overview of the situation in Québec

Based on a questionnaire administered as part of the present work, about three quarters of Québec's birthing hospitals and almost one third of its birthing centres are currently using pulse oximetry to screen for critical congenital heart disease. This test is apparently performed in almost all cases by a nurse in the hospital setting and always by the midwife in birthing centres¹.

Screening test performance

A certain consistency is observed in the results of the two most recent meta-analyses, which were conducted about six years apart, with regards to performance of the screening test. The authors of the more recent analysis conclude that, given the large size of their review as well as their precise estimates of sensitivity and specificity, further research into the accuracy of this screening method is not necessary. They consider the pulse oximetry test to be highly specific and moderately sensitive.

A very high degree of heterogeneity has been observed for all the performance parameters examined in the studies published subsequent to the two retained systematic reviews. This heterogeneity appears to be mainly due to the diversity of the conditions included among the primary targets of study. Also, the exclusion of newborns who had previously received a prenatal diagnosis from most studies tends to underestimate the performance of pulse oximetry, which would have most likely detected such cases. Lastly, the referral rate to specialized services and the false-positive rate should be interpreted with caution because in many cases, pulse oximetry identifies serious, noncardiac conditions, and the benefit of identifying these is not taken into consideration. The pulse oximetry test is considered a screening test that, if added to prenatal ultrasound and the physical examination, could potentially improve the overall detection sensitivity for critical congenital heart disease.

Efficacy of neonatal screening

In an observational ecologic study, American states that have implemented routine neonatal screening for critical congenital heart disease have achieved a significant (33%) reduction in deaths for cases diagnosed prenatally and postnatally, compared to states that have not implemented mandatory neonatal screening policy. The results of this study, which used population-based birth and death registries, have not been replicated

¹ In this report, the term "birthing centre" also includes home births assisted by a midwife. Midwife teams have a regional mandate, and although a woman may choose to give birth at home, she must first register with a birthing centre to obtain the services of a midwife.

to date and should be interpreted with caution, since the methods, the timing of the diagnosis, and practice changes over time cannot be taken into account with this design. American data from a cardiothoracic surgery database showed a significant decrease in the number of deaths at 1 year, but some sub-analyses tended to indicate that the pulse oximetry test was not responsible for this decrease, which appears to be due instead to prenatal diagnosis. A retrospective cohort study at three tertiary centres in the United Kingdom was unable to show that pulse oximetry testing confers a benefit in terms of 1-year postnatal mortality. Two of these three studies also attempted to identify links between early postnatal diagnosis and reduced mortality but were unable to do so. Lastly, mortality was assessed in these studies at different time points and according to different approaches, all of which presented various types of biases and limitations.

Safety of neonatal screening

Neonatal pulse oximetry screening for critical congenital heart disease seems to engender a risk of false negatives and varying types of false positives. Other, noncardiac hypoxemic conditions identified, even serious ones, will generally be classified as false-positive results in performance analyses of pulse oximetry screening for critical congenital heart disease, even though various experts have recognized that their identification is clinically beneficial. There is a consensus regarding the need for psychological support for parents following a positive screening result, and even more so when a diagnosis of critical congenital heart disease is confirmed.

Ethical issues and social acceptability

According to the literature, screening for critical congenital heart disease using the pulse oximetry test is generally considered acceptable by parents of children born in a variety of settings, including in the context of early postpartum discharge, as well as by the general public or citizens. According to the literature and the consultations held for the purpose of the present work, the overall perception of the pulse oximetry test by medical personnel and health professionals is positive and detecting heart and lung problems is seen as beneficial. In addition, the medical personnel who encounter positive test results were in favour of making the test routine, provided there are sufficient human resources. The issue of consent raised in the literature regarding pulse oximetry screening for critical congenital heart disease was also discussed among the experts and professionals on the different committees consulted.

Organizational aspects

As required by the MSSS mandate, a careful analysis was performed to determine the optimal approach for implementing a screening process for critical congenital heart disease using pulse oximetry, if deemed relevant. The advantages and drawbacks of a structured screening program were compared with those of a "preventive clinical practice" approach. Given the significant organizational requirements associated with structured screening programs and the fact that a significant proportion of birthing hospitals have already integrated the use of pulse oximetry into their clinical process, the

experts and stakeholders consulted indicated that they favoured a more flexible preventive clinical practice approach as a standard of care for newborns.

In this regard, the literature on the organizational aspects of pulse oximetry screening shows that it is essential to adapt the protocols according to the birth setting and that interpreting the algorithm requires training for the personnel who perform the test. Based on the experiential and contextual data collected, major changes would not have to be made to the organization of Québec's healthcare system in order to manage and follow pulse oximetry-positive cases because the continuum of care for pediatric cardiology services is well established. The experts agree that the ideal time for testing is at about 24 hours of life to avoid a significant number of false positives, but also to prevent many infants from becoming symptomatic before the test is performed. The test should be carried out before early discharge and allow for a certain amount of flexibility so that all newborns can be tested within a reasonable amount of time, including those in neonatal intensive care units.

Economic aspects

About 20 economic evaluations of pulse oximetry, that are essentially cost and cost-effectiveness studies from various countries, have been published since the early 2000s. Very few of these considered patient quality of life in the cost-effectiveness analysis or the potential benefits of screening for major illnesses other than critical congenital heart disease. The exactness of the assumptions, particularly regarding test duration and the use of disposable or reusable probes, can have a major impact on the cost of a population-based pulse oximetry screening program, which varies from CAD\$3.47 to CAD\$35 per newborn, based on the retained studies. The stakeholders consulted believed that the cost of pulse oximetry would not be an obstacle to expanding current practice. Despite the heterogeneity of the results, the economic literature shows that the systematic implementation of pulse oximetry screening, compared to current practice, would be considered "cost-effective" according to commonly accepted efficiency thresholds.

The integration of the entirety of the scientific, contextual and experiential data permits the following findings to be stated:

- Critical congenital heart disease can result in organ damage and life-threatening complications;
- Critical congenital heart disease requires medical attention at diagnosis, surgical or catheter-based intervention as soon as possible during the first month of life, and lifelong cardiology follow-up;
- The prognosis varies according to the critical cardiopathy present, but neurodevelopmental impairment is the most important comorbidity and is, in part, associated with late diagnosis;

- Some newborns with critical congenital heart disease would not be identified before discharge from the hospital or birthing centre, despite the use of second-trimester ultrasound and the physical examination as detection techniques;
- Adding pulse oximetry testing to prenatal ultrasound and the physical examination could potentially improve the early detection of certain types of critical congenital heart disease and identify newborns with other severe hypoxemic conditions;
- Despite indirect evidence from one study, it is difficult to conclude that pulse oximetry is effective in reducing mortality in infants with critical congenital heart disease, but experts appear to be unanimous about its potential for reducing morbidity through prompt postnatal management of cases of critical congenital heart disease and of any other severe hypoxemic condition;
- Pulse oximetry screening for critical congenital heart disease generally seems to be considered acceptable by parents, the general public and medical personnel, and by the various health professionals involved;
- According to the economic literature, the systematic implementation of pulse oximetry screening, compared to current practice, would not engender an inordinate incremental cost for society;
- No major changes would have to be made to the organization of care and services for managing and following pulse oximetry-positive cases in Québec, because the facilities that offer mother-child services have professional teams on site to assess the condition of the newborn, and the continuum of care for referral services to pediatric cardiology is well established;
- A balance between identifying newborns with non-stabilized transient circulation and the risk of infants becoming symptomatic before the pulse oximetry test is performed should be considered when deciding on the optimal moment to perform the test;
- In the experts' opinion, there is clinical benefit associated with identifying non-cardiac hypoxemic conditions using the pulse oximetry test.

In light of these findings:

INESSS recommends that the practice of pulse oximetry as basic care for newborns be expanded and standardized in all of Québec's birthing hospitals and birthing centres, in order to screen for critical congenital heart disease and any other hypoxemic condition.

This main recommendation is accompanied by a series of additional considerations.

Clinical and professional aspects:

- Similar to assessing vital signs to verify the health of an asymptomatic newborn, pulse oximetry should be considered as an additional tool for the early management of a newborn who might have a serious condition.

- As part of good clinical practice standards, the use of pulse oximetry would be implicitly included in the consent to care signed by the parents, but the health professional would have to inform them, as for any other procedure, of what is about to be done.
- Under no circumstances should pulse oximetry replace existing standards of care, such as the physical examination.
- The pulse oximetry protocol should be based on that of the Canadian Paediatric Society (Appendix F), while allowing a certain amount of flexibility so that all Québec newborns can be tested within a reasonable amount of time, including those in intensive care units.
- The professionals involved in this practice should record the pulse oximetry test result and the subsequent course of action in the newborn's chart, in accordance with the regulatory standards to which they are required to adhere in their practice.

Aspects regarding the organization of care and services and support of care quality:

- Given that a large proportion of centres have already put the recommended practice in place, efforts to optimize the implementation of pulse oximetry should be aimed at engaging those birthing hospitals and birthing centres that have not yet done so.
- A list of minimum specifications required for pediatric pulse oximeters could be made available to staff. To reduce costs, the use of reusable probes should be encouraged, depending on the setting and availability of such probes.
- Human and material resources for implementing this good clinical practice standard in birthing hospitals and birthing centres must be sufficient to ensure the quality of care for newborns.
- The various professional orders and associations concerned should facilitate the dissemination of this recommendation and support its integration into their members' practice. An online training tool could be developed to support professionals in interpreting pulse oximetry test results. The academic training of future professionals who perform this test could also be strengthened to take the present recommendations into account.
- To ensure the quality of the procedure, the identification of congenital cardiopathy and other hypoxemic conditions of the newborn by pulse oximetry could be the subject of an evaluation project by the medical quality assurance committees, or by the departments or the birthing centres concerned. Such a project could provide a portrait of the practice as well as an assessment of its impact on neonatal care.

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