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EDITORIAL

Neonatal screening for critical congenital heart disease: a rigorous exercise still short of a conclusion

Rastreio neonatal de cardiopatias congénitas críticas: um exercício rigoroso ainda aquém de uma conclusão

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Critical congenital heart disease (CCHD) remains an important cause of neonatal morbidity and mortality, and a proportion of cases still escape both prenatal ultrasound and the routine newborn examination, presenting only after discharge. Pulse oximetry (POx) screening was proposed to close this gap: it is simple and non-invasive, and over the past fifteen years it has been adopted in many countries as a complement to clinical examination. Yet the yield of any screening test depends on the population in which it is applied.

Ruivo et al. report a five-year experience (2017-2022) of neonatal screening for CCHD by POx at a tertiary hospital in Portugal.[1] Of 11,800 newborns admitted to the postnatal ward, 94% (n = 11,094) were screened; 4.8% failed the initial screening, and 60 infants were ultimately referred to Paediatric Cardiology. No case of CCHD was identified, whereas six infants were diagnosed with non-critical congenital heart disease and two with non-cardiac conditions – a false-positive rate of 0.5% and a positive predictive value of zero for CCHD. Analysing and publishing data that point towards a low benefit of a protocol widely acknowledged as a key instrument for neonatal screening globally requires considerable courage and merits substantial acknowledgment. The authors conclude that, although the program was feasible and well implemented in terms of adherence, its diagnostic yield in this particular setting is questionable, possibly due to the hospital's advanced prenatal diagnostic capacity. A closer look at the six "non-critical" diagnoses warrants caution before even this more modest yield is taken at face value. The two cases of aortic arch narrowing are explicitly described as stable, with no echocardiographic progression on follow-up – raising the question of whether these truly represent structural heart disease requiring intervention, or incidental findings of uncertain clinical significance. Similarly, the two atrial septal defects reported are not characterised by size, haemodynamic relevance, expected or final evolution, information without which it is difficult to determine how many of these six cases represent true disease, as opposed to normal variants or self-limited findings that would have resolved without ever being detected.

The primary strength of this study lies less in its quantitative data than in the underlying perspective. Rather than uncritically endorsing an international recommendation,[2] the authors ask a question rarely posed about screening programs already in place: is this method, in fact, adding value in this specific hospital or even in our country? This question warrants consideration against the history of POx screening in Portugal, which has never undergone a formal national validation process. The first description of its implementation in a Portuguese maternity unit dates to 2013-2014, at the

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Centro Hospitalar Universitário de Coimbra.[3] Its subsequent spread – including to the hospital in the present study, from 2017 onward – occurred hospital by hospital, by local decision, following international recommendations rather than any national mandate or decision. To date, POx screening for CCHD has never been the subject of a dedicated cost-effectiveness analysis at the national level. Assessing the local or national validity of a practice that was adopted progressively, rather than assuming it as given, is precisely the exercise this editorial advocates should become the standard whenever screening programs, regardless of how well-intentioned, are evaluated.

The main weakness of the study is, just as clearly, the absence of a cost-effectiveness analysis. The program consumed substantial resources – 530 failed initial screenings, 396 repeat evaluations, 60 referrals, and 56 echocardiograms – without detecting a single case of CCHD over five years. The study provides no estimate of what this represented in nursing time, echocardiography or other direct and indirect costs, nor any estimate of the quality-adjusted life-years potentially gained through the non-critical diagnoses that were made. The international literature shows this relationship to be far from straightforward: in the setting of home birth and early discharge in the Netherlands, the estimated incremental cost-effectiveness ratio was €139,000 per additional case of CCHD detected;[4] the seminal United Kingdom evaluation by Ewer et al. concluded that the cost-effectiveness ratio of pulse oximetry plus clinical examination compared with examination alone is £24,900 per diagnosis in a population with antenatal screening for CHDs.[5] Both conclude that screening was likely to be cost-effective given acceptable thresholds at the time, but present no definitive conclusions. Without such an analysis, the present study cannot fully substantiate the conclusion it appears to be moving toward.

The next steps seem clear to us. On one hand, a systematic review of the evidence on POx screening in settings with high prenatal diagnostic coverage is needed, clearly distinguishing what is known about diagnostic effectiveness from what is known about economic efficiency – the review by Grosse et al. on the costs and cost-effectiveness of POx screening is a useful starting point, precisely because it systematises highly heterogeneous estimates from the United Kingdom, the United States, and China.[6] On the other hand, and more importantly in the national context, there is a need to understand where this screening genuinely adds value in Portugal. Data from the National Register of Congenital Anomalies shows that the rates of prenatal diagnosis vary significantly across different regions and by level of maternity hospital differentiation[7] – strongly suggesting that the added value of POx is unlikely to be uniform between a Lisbon tertiary hospital and a maternity unit in a region without easy access to fetal echocardiography. A national multicentre study, stratified by hospital level of differentiation and by prenatal diagnosis coverage, would finally allow the question this work leaves open to be answered: in which hospitals does universal POx screening make sense, and in which might it be dispensable or replaced by reinforced clinical surveillance. This would be, in our view, the first example of rigorous scrutiny of a neonatal screening policy in Portugal – an analysis that this study clearly emphasizes as necessary.

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