

CORRESPONDENCE

Neonatal Screening for Metabolic and Endocrine Disorders

by Prof. Dr. med. Erik Harms, Prof. Dr. med. Dr. rer. nat. Bernhard Olgemöller
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Comment on the Positive Predictive Value

I enjoyed reading this informative and well structured article. Everyone involved in neonatal screening—in hospital and clinical practice and in the laboratory—would be well advised to have this review article and its practical tables ready to hand.

I do not think, however, that the positive predictive value reported by the authors on page 16 for the immunochemical 17-OH progesterone screening (3%), can be deduced from the authors' data on the specificity (adrenogenital syndrome [AGS] 99.39%, page 16) and the confirmed cases (AGS: 216) or the incidence (1:12 771) in *Table 1* on page 14; and neither can it be concluded from the cited bibliographical source (2).

The positive predictive value for AGS screening can therefore be assumed to be in the region of 1.3%, which means that about 80 (rather than 30) neonates would have to be examined in order to identify one baby with the syndrome.

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REFERENCES

1. Harms E, Olgemöller B: Neonatal screening for metabolic and endocrine disorders. *Dtsch Arztebl Int* 2011; 108(1–2): 11–22.
2. Nationale Screeningreports der Deutschen Gesellschaft für das Neugeborenencreening (DGNS): <http://www.screening-dgns.de/screeningregister-1.htm>

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In Reply:

We thank Dr Wolter for his comment because he is absolutely right. Based on both the numbers of adrenogenital syndrome (AGS) in *Table 1* and the laboratory results on the average specificity for 17-OH progesterone screening in 2005–2008 (page 16), the result is indeed a much higher number of false positive specimens and therefore a lower predictive value of the screening for AGS ($12\,771 \times (100 - \text{specificity}) = 12\,771 \times 0.61 = 78$).

In none of the testing procedures used for neonatal screening did the data on specificity among the screening laboratories vary as widely as for 17-OH progesterone screening. This may in part be due to the fact that at present there are no consistent cut-off values used. The main reason for the wide range, however, is the varying proportion of samples of preterm babies that individual laboratories receive from perinatal centers. In premature babies, 17-OH progesterone is often raised without AGS (one possible cause for error, *Box 2/B*, page 18).

The positive predictive value for AGS screening of 3%, which was reported on page 16, relates to mature babies born at term (after 37 complete weeks of gestation). We should have made that clearer in the text.

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Conflict of interest statement

The authors of both contributions state that no conflict of interest exists.