

The case for newborn screening for congenital adrenal hyperplasia in Australia

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TO THE EDITOR: We write to encourage policy debate over newborn screening for congenital adrenal hyperplasia (CAH). Classical CAH is a severe, life-threatening disease affecting about one in 15 000 liveborn infants in Australia.¹ An inexpensive screening test for newborns is available, but this test is not included in the current newborn screening program in Australia.

Three-quarters of children with CAH have the severe salt-wasting type that typically presents with failure to thrive, and progresses to severe hyponatraemic, hyperkalaemic dehydration and shock due to an adrenal crisis within weeks of birth. CAH is the most common cause of ambiguous genitalia in neonates (due to virilisation from adrenal androgens *in utero*); girls with CAH may be incorrectly assigned as boys unless the diagnosis is made without delay.

CAH can be easily detected in neonates before the onset of illness by an established heel-prick newborn screening test that has good specificity and sensitivity, especially when used together with second-tier testing. Screening for CAH has been available for 30 years internationally, and is used in all American states, New Zealand and many countries in Europe, Asia and Latin America. Newborn screening reduces mortality and incorrect sex assignment.² Case reports from Australia³ and overseas⁴ have shown that undiagnosed CAH is a cause of apparent sudden infant death syndrome. These deaths could have been prevented if newborn screening was in place. A pilot study in New South Wales showed that newborn screening for CAH prevented salt-wasting crises and their potential long-term consequences.¹

The cost-effectiveness of newborn screening is difficult to measure, and there is little published evidence on this subject. Although a recent study suggested that CAH screening is not cost-effective,⁵ the only outcome assessed was mortality; other benefits of early diagnosis and intervention — including reduced morbidity and psychological impact — were not assessed. Newborn screening for CAH is not expensive; the cost per test within the laboratory is about \$2, and the incremental cost per

infant is in line with other newborn screening tests.

In a recent survey, the Australasian Paediatric Endocrine Group found that 91% of paediatric endocrinologists considered provision of newborn screening for CAH in Australia to be very important. The Newborn Screening Joint Subcommittee of the Human Genetics Society of Australasia unanimously supports the inclusion of newborn screening for CAH in all Australian states. Two Australian parent and patient advocacy organisations — the CAH Support Group Australia, and Caring and Living as Neighbours — also strongly support the proposal for adding newborn screening for CAH to the current screening program.

Despite clear predicted benefits and agreement among key stakeholders and expert advisers, no state in Australia currently screens for CAH. It is the state governments — guided by the Australian Health Ministers' Advisory Council — who decide on funding for newborn screening tests, and who should be accountable for acting against the weight of expert opinion and systematic evidence.

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1 Gleeson HK, Wiley V, Wilcken B, et al. Two-year pilot study of newborn screening for congenital adrenal hyperplasia in New South Wales compared with nationwide case surveillance in Australia. *J Paediatr Child Health* 2008; 44: 554-559.

2 Working Group on Neonatal Screening of the European Society for Paediatric Endocrinology. Pro-

cedure for neonatal screening for congenital adrenal hyperplasia due to 21-hydroxylase deficiency. *Horm Res* 2001; 55: 201-205.

3 Gozzi TG, Harris NP, McGown IN, et al. Autopsy diagnosis of 21-hydroxylase deficiency CAH in a case of apparent SIDS. *Pediatr Dev Pathol* 2005; 8: 397-401.

4 Strnadova KA, Votava F, Lebl J, et al. Prevalence of congenital adrenal hyperplasia among sudden infant death in the Czech Republic and Austria. *Eur J Pediatr* 2007; 166: 1-4.

5 Yoo BK, Grosse SD. The cost effectiveness of screening newborns for congenital adrenal hyperplasia. *Public Health Genomics* 2009; 12: 67-72. □