Newborn screening may miss adrenal-gland disorder

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Review of Minnesota records finds 22 percent error rate.

(HealthDay) -- Routine newborn screening failed to identify about one-fifth of infants with an adrenal gland disorder called congenital adrenal hyperplasia, a new study has found.

This genetic disorder is characterized by a deficiency of the hormones aldosterone and cortisol, and overproduction of male sex hormones (androgens). The effects can range from mild to severe.

In the study, researchers looked at data on more than 800,000 newborns who underwent routine screening in Minnesota between 1999 and 2010. Of those newborns, 52 with congenital adrenal hyperplasia were identified and 15 -- 22 percent -- with the disorder were missed.

The study found that a negative screening result does not definitively rule out the condition, the researchers said.

"Screening programs should educate clinicians about false-negative results so any patient for whom there is clinical concern for [congenital adrenal hyperplasia] can receive immediate diagnostic testing, particularly females with ambiguous genitalia," said Dr. Kyriakie Sarafoglou, of the University of Minnesota Amplatz Children's Hospital in Minneapolis.

The study appears June 13 in the Journal of the American Medical Association.

More information: The U.S. National Institute of Child Health and Human Development has more about congenital adrenal hyperplasia.