MedicalXpress

Experts recommend universal screening of newborns for congenital adrenal hyperplasia

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Today, The Endocrine Society released a new clinical practice guideline on the diagnosis and treatment of congenital adrenal hyperplasia (CAH). The guideline features a series of evidence-based clinical recommendations developed by an expert task force.

The guideline, published in the September 2010 issue of the *Journal of Clinical Endocrinology & Metabolism* (*JCEM*), a publication of The Endocrine Society, is endorsed by the American Academy of Pediatrics, Pediatric Endocrine Society, the European Society for Paediatric Endocrinology, the European Society of Endocrinology, the Society for Pediatric Urology, the Androgen Excess and PCOS Society, and the CARES Foundation.

CAH is a genetic disorder of the adrenal glands that affects about one in 10,000 to 20,000 newborns, both male and female. The adrenal glands make the steroid hormones cortisol, aldosterone and androgens. In individuals with CAH, the adrenal glands produce an imbalance of these hormones which can result in ambiguous genitalia in newborn females, infertility and the development of masculine features such as development of pubic hair, rapid growth in both girls and boys before the expected age of puberty.

"If CAH is not recognized and treated, both girls and boys undergo rapid postnatal growth and early sexual development or, in more severe cases, neonatal salt loss and death," said Phyllis Speiser, MD, of Cohen Children's Medical Center of New York and Hofstra University School

of Medicine, and chair of the task force that developed the guideline. "We recommend that every newborn be screened for CAH and that positive results be followed up with confirmatory tests."

Other recommendations from the guideline include:

- Prenatal treatment of CAH should continue to be regarded as experimental. Such therapies should be pursued through protocols approved by Institutional Review Boards at centers capable of collecting outcomes data on a large number of patients so that risks and benefits of this treatment can be defined more precisely;
- Diagnosis should rest on clinical and hormone data while genotyping should be reserved for equivocal cases and genetic counseling;
- Regarding treatment, glucocorticoid dosage should be minimized to avoid iatrogenic Cushing's Syndrome. Mineralcorticoids and, in infants, supplemental sodium are recommended in classic CAH patients;
- Clinicians should avoid the routine use of experimental therapies to promote growth and delay puberty, and patients should avoid adrenalectomy;
- Early single-stage genital repair should be considered for severely virilized girls and should be performed only by surgeons experienced in this type of procedure;
- Clinicians should consider patients' quality of life, consulting mental health professionals as appropriate;

- At the transition to adulthood, clinicians should monitor for potential complications of CAH; and

- Clinicians should exercise judicious use of medication during pregnancy and in symptomatic patients with nonclassic CAH.

"People with classic CAH should have a team of health care providers, including specialists in pediatric endocrinology, pediatric urologic surgery (for girls), psychology and genetics," said Speiser. "Other than having to take daily medication, people with classic CAH can have a normal life."

More information: The fact sheet can be found online at: www.hormone.org/Resources/uplo … bilingual-081310.pdf

Provided by The Endocrine Society