Managing congenital adrenal hyperplasia requires shared decisions among patients, families, and healthcare professionals

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The Endocrine Society today issued a Clinical Practice Guideline that offers best practices for healthcare providers on how to promptly diagnose, treat, and manage patients with congenital adrenal hyperplasia (CAH), an inherited endocrine disorder, throughout their entire lives.

The guideline, titled "Congenital Adrenal Hyperplasia Due to Steroid 21-Hydroxylase Deficiency: An Endocrine Society Clinical Practice Guideline," was published online and will appear in the October 2018 print issue of The *Journal of Clinical Endocrinology & Metabolism* (*JCEM*), a publication of the Endocrine Society. This is an update of the Society's 2010 Guideline, to reflect newer published data and prospects of advances in diagnosis and treatments. The guideline emphasizes shared decision making among CAH patients, their families, and healthcare professionals when it comes to the medical, surgical, and psychological management of the disorder.

Congenital adrenal hyperplasia (CAH) is an inherited genetic disorder in which the adrenal glands, which make essential hormones for body functions, do not function properly. Classic CAH, which is common enough that it is screened shortly after birth in many countries, may cause life-threatening episodes of shock due to salt-wasting and dehydration. Female infants are usually diagnosed at birth because they have ambiguous genitalia (external sex organs that resemble male genitals). However, they still have normal internal female organs (ovaries

and uterus). A male infant with classic CAH usually appears normal at birth, although he may show signs of early puberty.

Non-classic CAH is a milder and more common form of the disorder that may not appear until childhood or adulthood. Symptoms can include early pubic hair growth and acne, masculine characteristics, and infertility. With proper care, people with either type of CAH can live long and healthy lives.

"The management of CAH requires a multi-disciplinary team of experienced healthcare personnel who integrate the endocrine, genetic, gyneco-urologic, reproductive, and mental health aspects of care," said Phyllis W. Speiser, M.D., of the Cohen Children's Medical Center of New York, Northwell Health, and the Zucker Hofstra School of Medicine in New York. Speiser chaired the writing committee that developed the guideline. "Our new guideline stresses the importance of shared decision making between healthcare professionals, patients, and their families when it comes to treatment and the need for ongoing care."

Recommendations from the guideline include:

- All newborn screening programs should incorporate screening for CAH, and infants with positive screens should be referred to pediatric endocrinologists.
- Prenatal therapy for CAH should be avoided (except as part of ethically-approved protocols) due to incompletely defined postnatal risks.
- Healthcare professionals should inform all parents of pediatric patients with CAH (particularly girls with ambiguous genitalia) about surgical options, including delaying surgery until the child is older.
- All surgical decisions for minors should be the prerogative of

families (i.e., parents with assent from older children) in joint decision making with experienced surgical consultants.

- Adolescents with CAH should start the transition to adult care several years prior to dismissal from pediatric endocrinology to ensure continuation of care throughout their entire life.
- Growing individuals with classic CAH should receive maintenance therapy with hydrocortisone and should avoid chronic use of more potent or long-acting glucocorticoids, which can have adverse side effects.
- Patients with CAH (and parents of minors) should seek mental health treatment to address any CAH-related psychosocial problems.

Provided by The Endocrine Society