



dsd-LIFE ABSTRACT FOR THE GENERAL PUBLIC

Title

Testicular function in adult male patients with congenital adrenal hyperplasia

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What did we want to know?

Current knowledge on testicular function in males with congenital adrenal hyperplasia (CAH) is scarce. We wanted to know the gonadal function of the adult male patient with CAH.

What did we do?

Males with CAH provided blood samples to assess hormone levels, semen samples to assess fertility, and underwent an ultrasound to examine the testicles. As CAH is a rare disease, this study was performed in 6 European countries, including 14 academic hospitals.

What were the main results?

Serum testosterone concentrations were below the reference range in 14 of 69 men, of which 50% also had abnormal levels of hormones stimulating the testicles. In males with normal testosterone concentrations, only 15% had abnormal gonadotropin levels. Evaluation of sperm quality revealed decreased sperm concentrations (15/39), motility (13/37) and abnormal morphology (4/28). Evaluation of the testicles by ultrasound showed testicular adrenal rest tumors (TART) in 39/80 males, with a higher prevalence in men with the most severe form of CAH (14/18) and in men with increased 17-hydroxyprogesterone (20/35) or androstenedione (12/18) serum concentrations. Forty-three children were fathered by 26/113 participants.

What does this mean?

Men with CAH have a high risk of impaired gonadal function: e.g. developing aberrant hormone concentrations, spermatogenic abnormalities or presence of testicular adrenal rest tumors. Regular assessment of endocrine gonadal function and imaging for TART development are recommended, in addition to measures for fertility protection.

Reference:

Engels M, Gehrman K, Falhammar H, Webb EA, Nordenström A, Sweep FC, Span PN, van Herwaarden AE, Rohayem J, Richter-Unruh A, Bouvattier C, Köhler B, Kortmann BB, Arlt W, Roeleveld N, Reisch N, Stikkelbroeck NMML, Claahsen-van der Grinten HL; dsd-LIFE group. Gonadal function in adult male patients with congenital adrenal hyperplasia. *Eur J Endocrinol.* 2018 Mar;178(3):285-294. doi: 10.1530/EJE-17-0862. Epub 2018 Jan 16.