

Patients with adrenogenital syndrome more likely to suffer from sexual dysfunction

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Adrenogenital syndrome (AGS) encompasses a group of inherited metabolic disorders caused by a disruption in steroid hormone production in the adrenal cortex. Doctor of internal and sexual medicine Michaela Bayerle-Eder and biochemist Sabina Baumgartner-Parzer from the Division of Endocrinology and Metabolism within MedUni Vienna's Department of Medicine III are studying sexual function and sexual orientation in a group of women with mild and severe forms of AGS. The main findings are as follows: they are much more likely to suffer from sexual dysfunction and problems with gender identification than healthy women. An early diagnosis is therefore extremely important.

This theme is also central to the 6th Scientific Symposium of the Austrian Society for the Promotion of Sexual Medicine and Sexual Health, to be held in Vienna General Hospital on 29—30 November 2019.

Adrenogenital syndrome (AGS) refers to a group of

inherited conditions caused by deficiencies of various enzymes that play a key role in steroid hormone production. By far the most frequent cause is a deficiency in 21 hydroxylase, caused by mutations in the CYP21A2 gene. This leads to a reduction in the production of cortisol and aldosterone and, at the same time, an increase in the production of male hormones. Both boys and girls can suffer from AGS, although they manifest different gender-specific symptoms.

The clinically severe form of "classical AGS" is characterized by masculinization of the external sexual characteristics (enlargement of the clitoris through to formation of a pseudo-penis) in girls (despite internal female genitals) and/or by life-threatening "salt wasting crises" in both genders shortly after birth. In such crises, patients lose salt and water and their blood pressure drops sharply.

"It causes a more extreme growth spurt in childhood, but ultimately with below-average adult height, false puberty with premature development of pubic hair, excessive body hair and acne, a rapidly developing penis in boys and absence of menstruation in girls," explains study author Sabina Baumgartner-Parzer from the Department of Medicine III. In Austria, there is a newborn screening program designed to detect severe forms of AGS to prevent these life-threatening salt wasting crises and/or to be able to give the affected children prompt and adequate treatment, including substitution therapy. Milder forms, known as "non-classical AGS," mostly go undetected, so that patients go for a long time without a diagnosis, due to the later onset and milder nature of the symptoms (hirsutism/excessive body hair, acne, inability to conceive). Parzer-Baumgartner says, "In this situation it is important that a CYP21A2 gene test be performed, so that even mild forms can be reliably diagnosed, as well as asymptomatic genetic carriers of the condition."

Doctor of sexual and internal medicine Michaela

Bayerle-Eder and biochemist Sabina Baumgartner-Parzer are looking at the extent to which prenatal androgenization influences sexual function and gender identity in female AGS patients: "Based on their sexual history and various specific parameters, it was found that AGS patients suffer more from sexual dysfunction and sexual stress than women from the general population. There is an unexpected tendency towards greater restriction of [sexual function](#) and greater psychological stress in patients with non-classical AGS. A large proportion of all volunteers report their gender role as "butch." Women with classical AGS have a higher homosexual preference, so that it can be assumed that prenatal hyperandrogenemia has an influence upon gender role and sexual preference."

The main finding is as follows: particularly patients with non-classical AGS, with less obvious symptoms, suffer more due to late diagnosis, since they have lived longer without any explanation of their "otherness" and have not received any treatment. "For women with signs of masculinization (Note: acne and increased body hair), menstrual problems, inability to conceive and [sexual dysfunction](#), it is therefore important to also consider non-classical AGS when making a diagnosis. Appropriate genetic tests are essential for an [early diagnosis](#) and/or in the context of family planning."

More information: Marie Helene Schernthaler-Reiter et al. Influence of Genotype and Hyperandrogenism on Sexual Function in Women With Congenital Adrenal Hyperplasia, *The Journal of Sexual Medicine* (2019). DOI: [10.1016/j.jsxm.2019.07.009](https://doi.org/10.1016/j.jsxm.2019.07.009)

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