Adrenogenital syndrome AGS

General:

Different pathways of the steroid hormone synthesis can be impaired in AGS. An overload of pre-metabolites results from enzyme blocks (hydroxylases) which leads to overproduction of androgens; synthesis of mineralocorticoids can be affected.

Clinical pictures:

In the congenital form, a common 21-hydroxylase defect is found (95% of the cases, heterozygosity frequency 1:40, with salt-wasting syndrome), 3-hydroxylase defect (rare, with salt-wasting syndrome) or 11-hydroxylase defect (rare, with hypertonia). In boys AGS leads to isosexual perturbations (hypogonadism, pseudopuberty), in girls to intersexual perturbations (female pseudohermaphroditism, virilism, primary amenorrhea, missing development of breasts, hypertrophy of clitoris). Children are tall as adults are small as children(early closure of the epiphyseal plate).

21-Hydroxylase defect (most common):

Increased: ACTH, 17-OH progesterone, testosterone and testosterone/SHBG quotient (women), androstenedione (men), sodium in urine and renin in plasma (salt-wasting syndrome approx. 1/3 of the cases).

Decreased: cortisol in serum, SHBG (women), sodium and aldosterone in serum (salt-wasting syndrome)

Stimulation tests: see ACTH short test, fast increase of 17-OH progesterone, slight increase of cortisol.

A genetic test for 21-hydroxylase is available (also see 21-Hydroxylase).

We recommend a genetic test for 21-hydroxylase :

11-Hydroxylase defect: increased 11-OH corticosterone and cortisol.

17-Hydroxylase defect: decreased testosterone and Estradiol

For complete list of laboratory test offered at Freiburg Medical Laboratory, please visit
http://www.fml-dubai.com/parameter-listings/

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