

## CASE STUDY: Lizzie – Adult Onset CAH

I've had irregular periods from the start and saw my GP in my teenage years who put me on the pill to regulate them but I got lazy and came off it while at college (as I didn't need it for actual birth control purposes.) When I came off the pill, my periods stopped completely and my skin was really bad (greasy with acne). I went back to the GP and after further tests, I was sent to see an endocrinologist who did an ACTH stimulation test and I was diagnosed with late onset congenital adrenal hyperplasia (LOCAH).

I also had an ultrasound scan, which indicated that I had polycystic ovaries. I was told that as I had a mild form of CAH, I did not need steroid medication so was put back on the pill and also spironolactone (which I was told was a diuretic and an anti-androgen). Thankfully this combination of treatment has kept my periods regular and cleared up my skin!

## CASE STUDY: Jo – Adult Onset CAH

After years spent thinking I had PCOS (Polycystic Ovarian Syndrome), I have just been diagnosed with Non-Classical CAH. I started taking 2.5mg of prednisolone 3 weeks ago and luckily I seem to be responding well to it. I no longer fall asleep at 8pm every night or wake up with no energy and all my minor allergies seem to have disappeared.

My main concern and the reason why I had further investigation is that I've been trying for a baby for over a year. At 18 I was diagnosed with PCOS, but I have regular periods, no acne and a scan showed no cysts on my ovaries, so they started to doubt the PCOS diagnosis and ran further tests until I received the diagnosis of NCCAH. Now I know what the problem is and am on treatment to regulate my hormones, I am hoping to improve my chances of becoming pregnant!

Information produced by the Congenital Adrenal Hyperplasia Support Group

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*"I can't tell you how much reading about the experiences of other parents who have children with NCCAH via your newsletters and on your website, has uplifted my family – thank you so much!"*

*"The support of the CAH group and the flow of information especially during the CAH conferences have been invaluable to us"*

Any further information can be obtained from:

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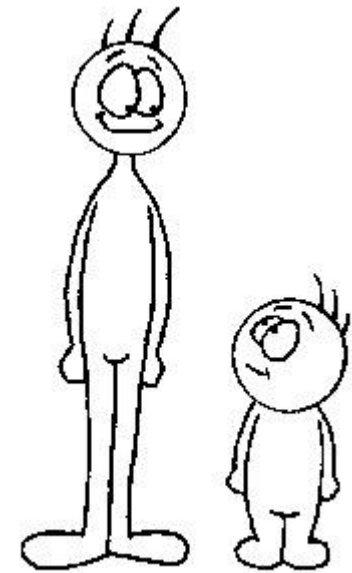
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**Non-Classical/Late-Onset  
Congenital Adrenal  
Hyperplasia**



The mildest form of CAH (often referred to as Non-Classical or Late-Onset CAH) is not immediately life threatening. This variation of the condition can cause a range of symptoms, some noticeable in early childhood and others not until adulthood. It affects children, women and men at any age.

In children, symptoms of non-classical CAH are:

1. Rapid early growth
2. Premature development of pubic hair
3. Body odour
4. Oily skin and hair
5. Severe acne
6. Mood swings

Females are born with normal genitals but may also have the following symptoms:

1. First menstrual period at early age
2. Irregular periods
3. Thinning hair (male pattern baldness)
4. Excessive hair growth (coarse facial hair, which can develop over neck, breasts and body, especially up the front of the abdomen)
5. Voice may become deeper
6. Symptoms of Polycystic Ovarian Syndrome (PCOS) may have been diagnosed
7. Infertility

Males also are of normal appearance at birth and the condition often goes unrecognised, as the symptoms can be quite subtle:

1. Short Stature
2. Early beard growth
3. Enlarged penis compared to testes size
4. Low sperm count/infertility

Often treatment with steroids is not necessary in women with Late-Onset CAH. Instead, giving oestrogen as in the oral contraceptive can regulate testosterone from the ovary. In fact, the treatment of Late-Onset CAH is usually the same as for the Polycystic Ovary Syndrome because the two conditions are so similar.

## How is Adult Onset CAH Diagnosed?

The doctor will measure androgens (male hormones) in the blood and note that it is far higher than expected.

Then the blood will be tested for the partly processed hormones. In practice the commonest fault in over 95% of patients occurs in an enzyme called 21 hydroxylase. The partly processed backlog hormone in this variety of CAH is called 17 hydroxyprogesterone (17-OHP) and this is the hormone the doctor will measure. If it is high then it is very likely the patient has CAH.

Sometimes to confirm the diagnosis, another test called a short synacthen test is performed. This involves an injection (intravenous or intramuscular) of synthetic ACTH (Adrenocorticotrophic Hormone) with measurements of both cortisol and 17-OHP. The doctor will then compare circulating concentrations of 17-OHP before and 60 minutes after ACTH administration. Stimulated values are invariably grossly elevated (>35 nmol/L) in patients with non-classic forms of CAH.

Sequencing of the CYP21A1 gene to confirm the clinical and biochemical diagnosis is a useful adjunct to hormonal measurements.

In terms of diagnosing cortisol deficiency, a peak cortisol results of >550nmol/L is a normal result; anything less than this should involve a discussion with your endocrinologist over hydrocortisone replacement therapy (taken either daily or at times of an inter-current illness). Evidence suggests that at least 30% of adult patients have some degree of cortisol deficiency and may be prone to stress-induced adrenal insufficiency.

Safety warnings will also need to be in place such as sick day rules for increasing daily dose, use of steroid /emergency cards and ID emblems as well as education with emergency hydrocortisone kits.

## CASE STUDY: Matthew—Late-Onset CAH

Our elder son Matthew was diagnosed as having CAH at the age of 7. His consultant said this condition was mild and that contributed to his late presentation. After a series of tests and diagnosis we were deeply shocked to learn that he had a bone age of 14 years. Although tall (4" 11") for a 7 year old, we were told it was unlikely he will reach his full genetic height potential.

Matthew has always eaten well and grown at a terrific speed. We wrongly presumed that his tall stature was inherited from us. Looking back, there were signs of abnormality earlier but being first time parents we perceived them as unusual but not unnatural. Apart from rapid growth, he did have a strong body odour from the age of 5 and greasy skin with acne but it wasn't until he started to grow pubic hair nearly 2 years later that we sought medical help.

Matthew is responding well to treatment and his growth has slowed down considerably since starting on Hydrocortisone. Last year, due to the advancement of puberty he was put on Prostag injections and his bone age has remained stable. We are delighted that the condition does not appear to have affected Matthew greatly and he is developing into a confident and happy boy and is a serious football enthusiast!

## CASE STUDY: Freya—Late-Onset CAH

Our daughter Freya was diagnosed with salt wasting CAH at the age of 4. Unlike many parents we were relieved to discover that Freya had a condition that could be treated as opposed to the big "C" as initially we had been concerned she had a tumour. It also answered many hospital visits that were left inconclusive and the jigsaw all started to fit.

Freya is full of life and at the tender age of 6 takes everything in her stride, including her tablets 4 times a day and hospital visits. We are learning as we go and that's the whole family, a few traumas' along the way and many questions from all angles i.e. her siblings, family members and friends but so far so good!