Psychological impact on children with CAH and their families

The diagnosis of CAH can come as a shock to parents, because their baby now needs life preserving medicines. Since the condition affects the appearance of the genitals in girls, many families feel distressed and confused, and can find it difficult to talk to family and friends.

These feelings of anxiety and distress are completely normal. Specialist psychological support is available and can be arranged through your local hospital to help children and families talk through their worries. Some families may find support groups like CLIMB CAH (www.livingwithcah.com) helpful.

The transition from child to adult services

At some point care will be transferred from children's to adult services. Full and open communication with the young person and their family will facilitate this. Transition Clinics or joint sessions with adult and paediatric services will allow this to happen in the best interest of the young person.

What are the long-term results of CAH?

CAH is a treatable condition where affected individuals can lead normal lives and have a normal life expectancy. General health, sexual function and fertility can be normal in adults with CAH. These may be affected however if the young person has difficulty taking their prescribed treatment.

Information gathering

All the children's hospitals in Scotland collect details on children with CAH as part of a national audit. Your child cannot be identified by anyone from this information and it will be fully discussed with you before we do this. Information may also be collected for international surveys and audits. Consent will be sought from you or your child for this, and you may withhold consent.

Who has reviewed SDSD activity?

NHS Quality Improvement Scotland (QIS)
National Services Division, NHS Scotland (NSD)
Director of Health Information & Technology, GCHB

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This leaflet will be available in other languages (eg Punjabi, Urdu, Polish) or formats (eg larger print) on request from the SDSD office or visit the website www.sdsd.nhsscotland.com.

NHS SCOTLAND

Scottish Differences of Sex Development Network

Information Leaflet

CONGENITAL ADRENAL HYPERPLASIA

What is Congenital Adrenal Hyperplasia (CAH)?

CAH is a condition of the adrenal gland which affects the production of steroid hormones such as cortisol (important for healthy blood sugar, and in times of stress) and aldosterone (important for salt levels in your body, and for blood pressure).

Congenital: As the condition is present from birth it is called congenital.

Adrenal: The adrenal glands sit on top of the kidneys (one on top of each kidney). Our kidneys lie within our flanks.

Hyperplasia: This means enlargement. Without treatment, the adrenals are much bigger than usual.

How common is CAH?

Around 1 in 10,000 children in Scotland are born with CAH.

What causes CAH?

There is a missing enzyme, usually 21 alpha hydroxylase, or 17 beta dehydroxylase, and the adrenals therefore do not make cortisol.

Instead, there is a build up of other hormones which the adrenals make. These are androgens, which can affect the genital appearance in girls.

The amount of cortisol produced by the adrenal gland is controlled by another organ, the pituitary gland, which lies in the base of the skull. Because the adrenal gland has difficulty producing cortisol, the pituitary gland responds by making more adrenocorticotrophic hormone (ACTH) which stimulates the adrenal gland to try and produce more cortisol. Constant stimulation of the adrenal gland in this manner causes it to enlarge (Hyperplasia).

Untreated CAH can result in severe salt loss, usually in the second week of life (but can happen from around 4 days of age). In the past, before treatment was possible, the baby would die.

How can CAH be treated?

CAH is a treatable condition. Treatment involves medical treatment with steroids and salt replacement which will be required in most children.

Medical treatment

The lack of cortisol can be treated using tablets and this will be advised by a paediatric endocrinologist. For infants the tablets are crushed or dissolved. Children are treated with hydrocortisone (works like cortisol) three times daily, and fludrocortisone (works like aldosterone) once daily. It is very likely that your child will require these hormones throughout their life. The risks of salt loss are greatest in infancy, so salt supplements are also given for the first year of life as a liquid.

In addition, because the body needs cortisol to respond to stress, your paediatric endocrinologist will advise how to increase the dose of hydrocortisone if your child is unwell. If your child is unable the take the hydrocortisone because of illness an injection of hydrocortisone may be required, and you will be taught how to do this.

Effective treatment is vital, so newly diagnosed infants may be in hospital for a couple of weeks following diagnosis, to ensure that treatment is effective.

Surgical treatment for genital appearance

In the past, many girl babies had surgery. However, many girls ran into problems in adolescence and adulthood due to these early procedures. Hence it is now recognised that there are advantages to leaving surgery until the child can consent. Some girl babies may even now require early surgery, for example, if they have bladder problems.

Girls with CAH should be assessed by a specialist paediatric surgeon with experience in genital surgery. The aim of surgery is to keep the child healthy, and to help her to make decisions about her own care as she approaches adulthood.

What happens as my child gets older?

The dose of hydrocortisone and fludrocortisone will change as your child grows. Therefore children, especially the younger ones, have to be seen in specialist endocrine clinics several times a year.

It is very important to provide enough treatment to stop the adrenal gland producing extra androgens, as these can cause the body to mature too quickly resulting in early puberty and short height. For these reasons steroid hormone levels need to be monitored, and blood tests are taken at most clinic visits. In addition, in some children finger-prick blood testing is also done at home once or twice a year. However, taking too much hydrocortisone can lead to weight gain or slow growth. Children have their height and weight measured regularly in clinic. Bone maturity is also assessed by X-ray of the wrist occasionally.

The dose of hydrocortisone required to control the CAH may make it difficult for some children to keep their weight down, and this can be a particular problem for girls at adolescence. In some children, other medicines may be used to control puberty or improve growth or weight gain and this will be discussed fully with you.