



Congenital Adrenal Hyperplasia
www.cahisus.co.uk


Great Ormond Street Hospital for Children
NHS Foundation Trust

DEPARTMENT OF ENDOCRINOLOGY



Christmas Newsletter~ December 2013

The support group had its twice yearly meeting at the Institute of Child Health on Saturday 9 November 2013. The programme was a very full one and there was so much general talking about subjects that we did overrun a little!! Just as well we did though because the weather outside was awful but quite a number of people managed to make the Lord Mayors Fireworks which was spectacular.

Fundraising

Big thanks to Nick and Linda Woodcraft and all who supported him as Nick raised £2300.00. Nick completed a 10 km mud run in March, the HSV Spring Triathlon in May, a 1K swim between the piers in Brighton in June and in September Perranporth Surf Challenge Triathlon 2013. Absolutely brilliant stuff and well done Nick on all these achievements which have really helped us with our fund.

In addition a big thanks to Bernie Mcgregor who bravely shaved off her hair to raise funds for CAH research.

Sports Bands

Our sport bands have proved to be popular and Paul has done an excellent job in raising funds for us in selling these. Many thanks to Paul for doing this and of course to his family who come along and help with these at the meetings.

Launch of Adrenal Insufficiency Card

As you will know we have been working hard on the emergency protocols. At the last meeting we launched our More at 4 campaign which is about giving additional hydrocortisone at 4 am during illness.

The 'More at 4' campaign says that at 4 am in illness an extra double/triple dose of hydrocortisone should be given based on double/triple the morning dose. The usual morning dose should also be doubled/tripled. This morning dose must also always be given as the 4 am dose is an additional dose and not a replacement dose. This additional dose must be given in all illnesses whether receiving medication orally or IV if in hospital.

This additional dose at 4 am is really important in order to avoid life threatening hypoglycaemia (low blood glucose levels) that can occur during episodes of illness. For more on this please look at our leaflet 'Extra Hydrocortisone Dose at 4 am in Illness' on <http://www.cahisus.co.uk/leaflets.htm>

To support this further we have redesigned the Adrenal Insufficiency Card. Many thanks to Amanda Copper who has done a lot of the art work and design for us and you can now see the card on our website and download it. We still have a number of cards available at clinic so you can pick them up there as well but the website is probably the easiest way to get hold of one straight away. www.cahisus.co.uk

The cards contain all the information that you need for urgent contacts and advice, along with details of what to do when unwell. Everyone needing glucocorticoid should have one of these and we strongly urge families to get a few of these cards, so that they can be given to carers of their children, placed in emergency kits and to the children themselves in their school bags or pockets.

This does NOT replace Medic Alerts but should be used in conjunction with the Medic Alert, which have the wording 'Adrenal Insufficiency' and should be worn at all times.





Congenital Adrenal Hyperplasia
www.cahisus.co.uk



Further Work on Emergency Care

Kathy Geertsma and Peter Hindmarsh met with Professor John Wass at the Royal College of Physicians to start to look with the College at creating uniform standards for the management of emergency situations in people with Adrenal Insufficiency. We have spearheaded this campaign and a lot of the work that we have done in creating a solid knowledge base in our leaflets has been well recognised by Professor Wass at the College.

We are going to follow this further early in 2014 with further meetings at the College of Physicians to get some work done to ensure uniform standards across the Ambulance Services and in Accident and Emergency situations. As many of you know Accident and Emergency departments are staffed largely by adult physicians who bring in paediatricians whenever a child is in casualty. We do think it is important however, that we target these consultants in Accident and Emergency Medicine in order that we ensure that uniform protocols are in place for dealing not only with children but also for adults with adrenal insufficiency. We will be working on this in 2014 and will keep you updated in our meeting and through the newsletters on how we are developing this idea.

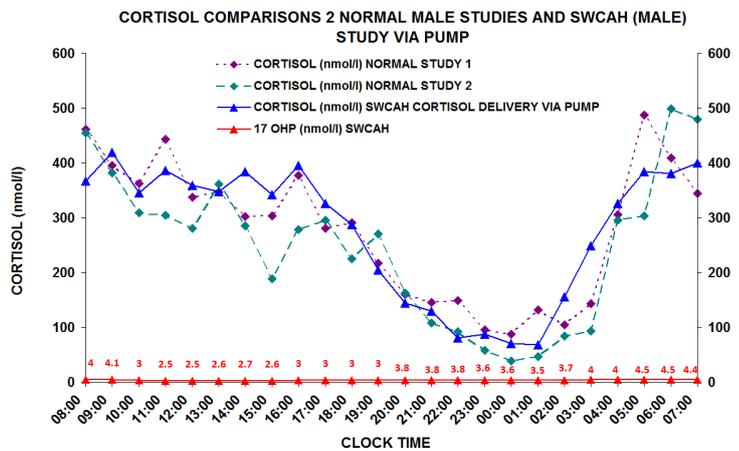
Profiles and Delivery of Hydrocortisone

At Great Ormond Street Hospital and University College London Hospitals we are very proud of the profile studies that we undertake. Despite receiving considerable amount of criticism from several people we have been able to establish the value of these profiles in a number of presentations that have been given at international meetings.

Our work shows that these profiles reduce the amount of hydrocortisone that is needed by many individuals and also indicate that often a

switch to a 4 times per day dosing schedule is required. The profiles have also demonstrated how important it is to try to get as close as is possible to the normal 24 hour circadian rhythm of cortisol.

As a quick taster we show a profile of two males without congenital adrenal hyperplasia along with a male who has salt wasting congenital adrenal hyperplasia and has received replacement hydrocortisone via a pump.



The exciting thing about this study is how we can achieve the circadian rhythm using this form of treatment. The challenge is to now try and see if we can mimic this as closely as possible with oral therapy and certainly our initial work with 4 times per day therapy would suggest that we are getting closer and closer to this ideal.

Getting doses right

At the meeting information was presented on the importance of the clearance of cortisol from the blood stream. When we take hydrocortisone it appears in the blood as cortisol but its handling in the blood stream is also important for getting good control of congenital adrenal hyperplasia. The way in which cortisol is removed from the blood stream can be measured and is known as the clearance of cortisol.





Congenital Adrenal Hyperplasia
www.cahisus.co.uk



Cortisol clearance differs in individuals and the overall message is that people with fast clearances will need more frequent doses of hydrocortisone.

A study on 40 patients receiving hydrocortisone the clearance, also known as the half-life in the circulation, was anything between 40 and 220 minutes. This means that whenever we are thinking of replacement therapy it is not only the peaks and troughs of cortisol that are important but also how long it last in the blood.

Although we can start treatment with fixed doses per body size once this has been established then fine-tuning is needed because the clearance is so variable in people. This means that some people will only need 3 times per day dosing and others 4 or more per day.

Our Story – Alice and Richard Jackman

Our thanks to Alice and her son Richard Jackman, who told us about Richard's journey with Asperger's and Cushing's disease. Cushing's is when the body makes too much cortisol and in Richards's case this was the result of a pituitary tumour. Having the tumour removed left Richard with panhypopituitarism - resulting in Adrenal Insufficiency and needing lifesaving hydrocortisone replacement therapy. Richard and Alice centred their story on the difficulties Richard had faced with employment following his many health problems.

Disability and Discrimination

One of the areas that attracted considerable interest and discussion at the meeting was the whole question of disability and discrimination, particularly with respect to congenital adrenal hyperplasia.

We asked Catherine Casserley from the Cloisters Chambers to present the legal view

of disability discrimination law in employment and education. We have a full set of the slides available on our website but Catherine pointed out a number of important areas.

The first is the definition of disability in which a person has a disability if that person has a physical or mental impairment and the impairment has a substantial long term adverse effect on the person's ability to carry out normal day to day activities.

The key for those with congenital adrenal hyperplasia and adrenal insufficiency is to consider would the individual be disabled if they did not receive the treatment. The straight answer of course is 'yes' because the condition is life-threatening. As such CAH and Adrenal Insufficiency fulfil all the criteria for definition of disability.

Catherine then went on to look at the 'Equality Act 2010' which states clearly that it is unlawful for an employer to discriminate against an applicant or employee in certain situations and this largely returns to the whole question of disability. She presented all different types of discrimination which are nicely outlined in the slides and these are really important for our older children and young adults to consider when they come to employment and work.

With respect to schools she made the point that it is unlawful to discriminate against admission as a pupil with a disability and the terms in which a person is offered admission to a school.

Schools must not discriminate against a pupil in the way the education is provided and the way that a pupil is afforded access to a benefit facility or service, e.g. school trips.

The school must make reasonable adjustments in terms of accommodating the pupil within the school environment.





Congenital Adrenal Hyperplasia
www.cahisus.co.uk



What is reasonable is also a big question and is largely tested by answering the question to what extent taking any particular step will be effective in overcoming the substantial disadvantage that the disabled pupil faces.

All these issues have been explored in the slide show that Catherine very kindly provided and which we now have available for you on our website 'Articles' page.

An Innovative Project (Full details to be announced at a later date)

Thanks to Anne and Chris who have been working really hard on their project that they are sure will be very beneficial to all.

Developments at University College London Hospitals (UCLH)

Two linked developments are taking place at UCLH.

The first is that Professor Hindmarsh has been appointed as Clinical Lead for Endocrinology at UCLH.

The second is part of the work that he will be undertaking there is to create the day case, inpatient and outpatient facilities that will allow adrenal patients to be seen onwards from 19 years to 25 years of age.

We have talked about this quite a lot at various meetings now thanks to the hard work Kathy has put in on behalf of our group, UCLH is keen to explore and get this service in place.

There has been top level support in the hospital for this right from the Chief Executive and Chairman so it is going to happen!! There is quite a lot of design work to do and recruitment to be made but the main thing is that we have the top level support and that will ensure success.

We will keep you up-to-date on this because this is going to be a really important development for young people.

Next Meeting

We are going to hold our usual Spring meeting towards the end of May.

I would like to take this opportunity to thank all our members and indeed all who came to our last meeting - it was fantastic to see such a good turnout and the feedback has been so rewarding.

Thanks too, to Angela and Chris for running the pump display and also big thanks to Jacqui Mayes, who is a Nutritional Therapist, for making herself available at meetings to help members with any dietary advice.

Wishing you all a very Merry Christmas and a very happy, healthy New Year

With best wishes

Professor Peter Hindmarsh

*Merry
Christmas*

