Cystinosis Foundation UK News
Forever Growing

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## Editor's Note

***Hello everyone**, This issue contains information on research which the Cystinosis Foundation UK is funding. We also have an article by Dr Dalton on the how, why and when patients need to take their Cystagon prior to their blood tests. On a lighter note a teacher from my daughters school has also sent in an article about what its like to teach a child with Cystinosis. Last but not least the foundation wishes to thank Kate Timmins who is helping to look after the Cystinosis website along with Matt Blackham. They both work really hard to keep the website upto date.—so thanks to both of them both!*

***Satinder***

## Exciting Results from Sunderland

The prodrug research sponsored by the Foundation at Sunderland University passed a highly significant milestone recently.

To recap, these prodrugs are chemically modified cysteamine (the active ingredient in Cystagon) designed to be transported round the body and be smuggled into cells where they will release cysteamine on the spot where it is needed. If successful this approach would lead to many fewer side effects and much lower doses because little would be lost through excretion.

Having proved that they are transported into normal kidney cells and release cysteamine inside them, it has now been shown that this also occurs with cystinotic cells *where their ability to remove cystine was shown to be at least as good as cysteamine*. These results, combined with the very low cell toxicity that they showed, are extremely encouraging.

**Prof Roz Anderson**

However there is still more work to be done before we have a usable medicine. Sunderland have made nine variants of their ideas but so far only one has been tested and shown to work in cystinotic cells. All should be evaluated to identify the best candidate. Then there are a number of standard and detailed toxicity tests that will be required before the best candidate can go forward to clinical trials. To reach that position the Trustees have recently *made an award of over £93,000* to fund the work for three more years.

If you would like to read about the technical details of the work done so far, visit our website and see the report by Professor Roz Anderson the leader of the project.

***John Terry***

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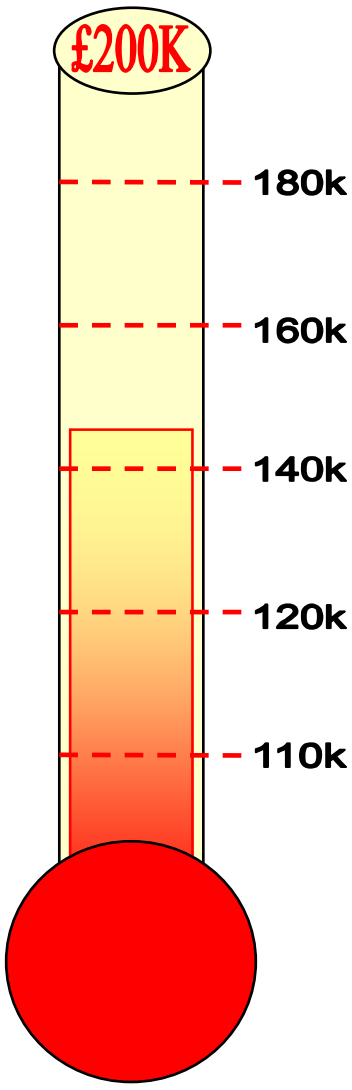


# A National Cohort Study of Adult Cystinosis Patients

*Below is a description of a study, led by Professor John Feehaly and Dr William van't Hoff, that the Trustees have agreed to fund. It will cost £23,443 over 12 months. Their efforts and our investment will be wasted without the support and collaboration of as many as possible of our adult patients. When you are approached please remember this. The outcomes from the research will be not only to your benefit but also to all the younger patients coming along.* ***Jonathan Terry***

People with cystinosis may develop problems affecting ***many*** parts of the body as ***well*** as the kidneys. The commonest problems are sensitivity to light, underactive thyroid, and reduced growth. Fortunately, treatment such as kidney transplantation and cysteamine therapy means that people with cystinosis are now living longer. One consequence is that new, previously unrecognised features of the disorder are being recognised. These ***can*** include ***diabetes and muscle-wastage***.

**Dr Van't Hoff**

Most patients with cystinosis are seen in paediatric clinics, but as they get older they transfer to adult clinics. Most move to the care of adult renal physicians, while a few are followed in specialised metabolic clinics. Most patients transfer to local renal units according to individual circumstances, which means that adults with cystinosis are seen in many different units, with little opportunity for specialisation and continuity of follow up.

**Development of Study Group**

We have started a UK Cystinosis Study Group with the aims of improving the understanding of the effects of cystinosis in adults, improving the transition of patients with cystinosis from paediatric to adult care, improving management and outcomes in adults with cystinosis, and co-ordinating research.

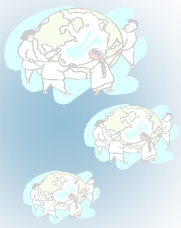
The group includes adult and paediatric kidney specialists from different parts of the country as well as patients (Table).

| Person | Representing | Organisation |
|---|---|---|
| Dr William van't Hoff | Paediatric nephrology | Great Ormond Street |
| Prof John Feehally | UK Renal Association | Leicester |
| Dr Peter Andrews | London & South | St Helier Hospital |
| Dr Graham Lipkin | Midlands & Wales | Birmingham |
| Dr John Sayer | North & Scotland | Newcastle |
| Mr Jonathan Terry | Patient | Cystinosis ***Foundation UK*** |
| Ms Anne-Claire Panniset | Patient | Cystinosis ***Foundation Support*** |

We plan to collect information about medical problems from as many adults with cystinosis as possible by questionnaires, clinical assessment, and review of clinical notes.

We plan to see each participant again every 5 years, so that we can continue to learn how cystinosis ***affects patients*** as they get older. We also hope that this work will help us to develop new models of care, in other words organising healthcare for adults with cystinosis to enable them to see a cystinosis expert without too much travelling.

The Cystinosis Foundation has been helping from the beginning to develop this project, and has generously offered to provide some funds to help us employ a researcher so that we can move the project forward as quickly as possible. The proposed research will be reviewed by a nationally-approved Research Ethics Committee soon. If approved, invitations for assessment will be sent out to adult patients in early 2009. We will keep members of the Cystinosis Foundation updated at regular intervals. In the meantime, we welcome your comments: contact the **Cystinosis Foundation *UK*** ***(advice@cystinosis.org.uk )*** ***Dr W Van't Hoff***



# The Importance of Measuring White Cell (Leucocyte)

The importance of white cell cystine measurements in the diagnosis and clinical management of patients with cystinosis cannot be over emphasised. The measurement is particularly valuable in determining how well Cystagon treatment is working, but there are problems, specifically in relation to the time of blood sampling time, that can lead to misleading results. The purpose of this "essay" is to discuss some critical points that, hopefully, will lead to be a better understanding of the white cell cystine test.

**Diagnosis-** Increased white cell cystine concentrations are only observed in individuals with cystinosis. Despite improved genetic diagnosis, the measurement of white cell cystine remains the easiest and quickest test to confirm or rule out a clinical diagnosis of cystinosis. A blood sample for a diagnostic test can be taken at any time, and under virtually any circumstance, provided the white cells are correctly prepared within 24h of sampling. Our experience, and that of the majority of other laboratories offering a diagnostic service for cystinosis, is that an elevated white cell cystine concentration (>1 □mol ½ cystine/g protein) is diagnostic of the disease.

**Benefits of Cystagon-**Although critical for diagnosis, the real importance of white cell cystine measurement in cystinosis, is as a measure of how well the Cystagon treatment is working. Before treatment with cysteamine/Cystagon became available, the long term outlook for patients with cystinosis was extremely poor. However, since the introduction of cysteamine/Cystagon therapy, though some problems persist, life can be fun and fulfilling. Growth can be significantly improved and end stage renal failure can be delayed for many years. Consequently, it is vital that your doctor can accurately check how well your prescribed dose of Cystagon is working.

**How to check how well Cystagon is working -** Checking how well a drug is working is very important in deciding the dose required and how often it needs to be given. If you have high blood pressure and you are prescribed a drug to lower blood pressure, then it is easy to check how well the drug is working, simply by measuring blood pressure. In cystinosis, the pioneering studies on cysteamine/Cystagon demonstrated that reductions in average white cell cystine concentrations were associated with better growth and slower progression of kidney disease. Consequently, the check on how well Cystagon is working in any patient, is the measurement of their white cell cystine level.

**How constant is your white cell cystine level? -** If appropriate decisions about dose/frequeny of Cystagon are to be made it is important that your clinician has confidence in the white cell cystine results. In this respect, it is very important to realise that your white cell cystine level is not constant. There will be some variation both during the day and from day to day, depending on your mealtimes, diet, growth, and any infection. However, these changes, except perhaps for infection, tend to be small compared with the lowering effect of Cystagon.

**How does Cystagon affect your white cell cystine level? -** Following a dose of Cystagon, it is absorbed in the gut and appears in the blood. Some of the Cystagon will get into cells and start to reduce cystine levels. The biggest fall in white cell cystine level is usually 1-2h after taking the dose of Cystagon. After this time, the level will slowly increase back towards the pre-dose level. This means that a blood sample should never be taken for white cell cystine level <4h after a dose of Cystagon, as the level could be very misleading.

**If the white cell cystine level is changing how can measuring it be of value to the doctor? -** BE CONSISTENT. This applies to clinic staff and patients. Try to make sure that your blood *cont ->*

samples for white cell cystine measurement are always taken at around the same time after a dose of Cystagon. In addition, it is vital that the time of your last dose, the actual dose, and the time the blood sample was taken are accurately recorded.

**What is the best time to have my blood taken for white cell cystine levels? -** The best time is just before your next scheduled dose of Cystagon. This will tell your doctor the highest level that your white cell cystine will be on your current dose/frequency prescription; most of the time the level will be lower.

**Examples:** ***Frequency of Cystagon dosing, 4 times a day.*** **-** If you take your morning dose at 07:00 try to have your blood sample taken at 13:00. If you know that normally you will not have your blood taken until 15:00 then take your dose at 09:00.

***Frequency of Cystagon dosing, 3 times a day.*** **-** If you take your morning dose at 07:00 try to have your blood sample taken at 15:00.

**What should I do if my clinic appointment is usually early or mid-morning? -** This is not a problem. The best thing to do is to delay your first dose of the day until after your blood sample has been taken. This may mean that instead of 6h between dose and blood sampling the time may be extended to 10h or more. Provided this interval is the same, within an hour or two, every time you have a white cell cystine blood test, it will allow your doctor to manage your Cystagon dose/frequency with confidence.

**The important question -** All the advice suggests that, on Cystagon treatment, the target white cell cystine concentration should be <1 μmol ½ cystine/g protein. I am taking a high dose of Cystagon, 3 times a day, but my blood samples for white cell cystine levels are usually taken 12h after my last dose and the results are usually around 1.2 to 1.3 μmol ½ cystine/g protein.

**Is my Cystagon dose/frequency effective? -** Yes. The result tells your doctor that when you are taking your Cystagon dose every 8h your white cell cystine level is <1 μmol ½ cystine/g protein for most, if not all of the time.

**The important points to remember:**

**TAKE THE PRESCRIPTION.** If there any problems with the dose of Cystagon or maintaining the frequency, then discuss them with your doctor or nurse specialist so that the best dose and frequency for you can be organised.

**BLOOD SAMPLING FOR WHITE CELL CYSTINE LEVELS - BE CONSISTENT.** Try to organise that the blood is always taken at around the same time after a dose of Cystagon, preferably just before your next scheduled dose.

It is vital that the time of your last dose, the actual dose, and the time the blood sample was taken are accurately recorded.

With a bit of understanding, effort, and discussion between patient and doctor or nurse specialist, every patient with cystinosis will get the maximum benefits of Cystagon treatment.

***Dr Neil Dalton***

# Teaching A Child with Cystinosis

When I was first told that I was going to have a child with cystinosis in my class, I have to admit I was clueless. It was a condition I had never heard of and although it sounded like something 'medical', I really had no idea what to expect. On learning more about the severity of the condition, my ignorance grew steadily into shock and then, finally (possibly the day after...) into panic. As the plethora of medicines and tablets were arranged before me, fear began to take over. I had had no previous medical training or experience of any kind. What would I do if something went wrong? Shouldn't this child have a 24 hour nurse or something? But the previous class teacher reassured me that everything 'would be fine'. And of course everything was fine.

**Avneet Seehra & Mrs Dale**

In school, there were lots of people who had worked with Avneet before and their knowledge and expertise were so useful to me, especially during the first few weeks of term. Some had been trained in how to properly administer the medicines and everyone was so calm I began to think that maybe it wasn't going to be such a big deal after all. I was very grateful to have that experience on site – some of my queries were so trivial I wouldn't have wanted to trouble Avneet's parents by asking them.

Information about cystinosis was displayed in the medical room, staffroom, office and my classroom so that any other adults coming into school would have easy access. The timings and clear dosage information, which had been helpfully provided by Avneet's family, were invaluable on the days when the adults who had responsibility for administering the medicines were unavailable.

Avneet's parents were so helpful and approachable and the support they gave me at the start of the year was wonderful. I was provided with information that was thorough, but not overwhelming. Laminated cards with important information were provided and we have constantly referred to them all year. Whenever changes were made to Avneet's dosage, these cards were updated and replaced in school.

As well as making sure I was well informed, Avneet's parents also made themselves fully available to us. If Avneet was ill, or there was a problem with her medicine, we could always ring to find out what we should do. I can't tell you how much that helped to relieve my fear that I would do something wrong that would make Avneet ill. I know it is not always possible for parents to be at home all day, and even if they are they have better things to do that wait for a phone call from an over-anxious teacher! But just to know we had a mobile contact, and that it was fine for us to call and check something... anything... was a great relief.

Having that open dialogue with Avneet's parents has had the most impact on my ability to deal with the cystinosis and get on with the teaching! I cannot stress enough how important this has been. Just a quick 2 or 3 minute chat in the morning or on the playground in an afternoon has been brilliant – now sometimes it's just a 'thumbs up' if everything has been ok. We also have a home school book that we use to write anecdotes of things that had happened either at home or school. Although we don't write in it everyday, it is another way to make sure we communicate fully.

Cont->

Forever Growing

We would like to know what you think of the newsletter and we would be grateful if you could share any ideas with us on how to improve it.

This newsletter is a way for you to share information and voice your opinions so please write in with articles.
You can contact me either by post or email

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***Inspirations...***

***When the world say's,***
***"Give up"***
***Hope whispers,***
***"Try it one more time!"***
***Author Unknown***

Although I feel that we manage Avneet's condition very well in school, I would be lying if I said it has had no impact on her learning. Unfortunately, the many unavoidable medical appointments and sometimes prolonged absences have meant that Avneet has gaps in her learning. However, the strong relationship we have with her parents has meant that we talk in depth about what exactly Avneet has missed and what can be done at home to fill those gaps. We share resources between home and school where necessary and have informal meetings about Avneet's next steps in between parent's evenings.

Coming to the end of the school year, we are all delighted with the progress that Avneet has made. She really has learned a lot this year, as have I. I have thoroughly enjoyed teaching her, as much as I have enjoyed building such a strong relationship friendship with her parents.

Cystinosis is frightening to an inexperienced teacher, but we are easily reassured. It does have a massive impact on children's learning, but there are ways to minimise it - with good communication, preparation and lots of team work. My advice to any parents of children with cystinosis is just to talk to their teachers and let their teachers talk to you. Be honest, be open and don't laugh at their silly questions and worries!

*Mrs Dale Ridgeway School*

## Believe in Yourself

There may be days when you get up in the morning and
thing aren't the way you had hoped they would be,
That's when you have to tell yourself that things can get better.
There are times when people disappoint you and let you down,
But those are the times you must remind yourself
to trust your own judgements and opinions,
to keep your life focused on believing in yourself.
There will be challenges to face and changes to make in your life,
and its up to you to accept them.

Constantly keep yourself headed in the right direction for you.
It may not be easy at times, but in those times of
Struggle you will find a stronger sense of who you are.
So when the day s come that are filled
with frustration and unexpected responsibilities,
Remember to believe in yourself
and all you want your life to be,
because the challenges and changes
will only help you to find the goals
that you know are meant to come true to you.

Keep believing in yourself. ***(Author Unknown)***

**Did You Know?**

That when you visit our web shop at

http://www.buy.at/cystinosis and make a purchase the retailer will make a donation to the Cystinosis foundation UK at no extra cost to you! Currently we have raised £60 this way! ***Remember every purchase counts!***

FundRaising Standards Board

Send Gavin items suitable for Prizes or Selling. This will help him with his fundraising activities.

If you need any help with fundraising contact Gavin below:

***Gavin Reddington***
***2 Sitwell Villas***
***Morton, Alfreton***
***Derbyshire***
***DE55 6GX Gavin@greddington8.orangehome.co.uk***
01246 860 133

***DISCLAIMER: Information provided, either verbally or in written form, is purely of an advisory level and we recommend that you consult your doctor before making any decisions regarding the future of the patient concerned.***