

## A National Cohort Study of Adult Cystinosis Patients

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*.

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).

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](mailto:advice@cystinosis.org.uk)).

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