NIHR | BioResource

CYS. Cystinosis

NIHR BioResource – Rare Diseases study project

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Summary

Cystinosis affects around 120 children and a similar number of adults in the UK. It is a genetic condition which leads to the accumulation of cysteine (an amino acid) in potentially any cell in the body. This can therefore cause a wide range of problems affecting organs, nerves and muscles.

One of the main problems is kidney disease, which can lead to passing lots of urine and growth problems for children. It usually leads to dialysis or a kidney transplant. The other main problem is eye disease where cysteine crystals are deposited at the front of the eye which can be painful and impair vision. Other associated problems include diabetes, thyroid disease, muscle wasting, difficulty with swallowing, breathlessness and infertility in males.
In order to try to reduce these problems, people with cystinosis need to take medication called cysteamine. Although this has transformed the outlook for people with cystinosis, it is nowhere near a cure.

People with cystinosis, even brothers and sisters who have the same mutation, have a very wide range of disease expression. Some people you could barely tell had anything wrong at all whereas others unfortunately can be very badly affected indeed.

It is hoped that, by building a cohort of patients in this project as part of the NIHR BioResource, this will enable future studies to try to unpick why some people are more severely affected than others and this might lead to ways to improve outcomes for people with cystinosis.

Recruitment Criteria

Inclusion

Cystinosis – to include non-nephropathic ocular cystinosis. (As per RaDaR.)

Exclusion

None.