Researchers uncover new forms of blindness

April 8 2014

Scientists from the University of Leeds have discovered six new forms of inherited blindness, each one resulting from mutations in a different gene important in eye development and vision.

The research, carried out by experts from the University's School of Medicine over the past three years, focuses on identifying new genes which, when mutated, cause these blinding disorders.

Pinpointing the exact cause gives clinicians the ability to offer their patients more information about how their condition will progress, what risk there is to relatives, and in some cases can point to specific treatment.

A grant from the National Eye Research Centre, together with funding from other sources, supported a team led by Professor Chris Inglehearn, Dr Manir Ali and Dr Carmel Toomes. They studied local families, particularly from the West Yorkshire Pakistani community where such conditions are common, in order to track down the genes involved.

Using DNA technology, they sequenced every gene in patients from families in which multiple members were blind from birth due to conditions such as Leber's congenital amaurosis, cone-rod dystrophy or global defects of eye development.

Professor Inglehearn said: "Finding these genes is important in many ways. As well as directly benefitting families it allows researchers to build a more comprehensive list of the genes needed for the eye to develop and function properly.

"This greater understanding, together with next generation DNA sequencing technology, is driving a diagnostic revolution in inherited eye diseases.

"As the cost of sequencing comes down, it will soon be feasible to sequence all patients. This still doesn't lead us straight to the mutations that cause the condition, but as we and others identify more of the genes involved, we get progressively better at interpreting their DNA code to work out what the problem is in each patient."

The National Eye Research Centre is now fundraising to support the Leeds team on the next steps in the research, in which the greater knowledge gained is used to screen patients on a larger scale, developing and testing new methods to reduce cost and bring closer a time when such tests are available to all patients and their families on the NHS.

"As well as improving diagnostics, the greater understanding of the

biology of vision gained from such studies also informs the search for new forms of therapy," Professor Inglehearn added.

"Since new therapies are often specific to particular forms of inherited blindness, it is essential for each patient to know which condition they have, so that when new treatments are being tested they can enrol on clinical trials, helping to test the new therapies and potentially benefiting from them themselves."

Mike Daw, of the National Eye Research Centre, said: "This is exactly the type of research that my charity is proud to support and demonstrates how advances in DNA sequencing might lead to individually targeted treatments which have the potential to improve the vision and outcomes for hundreds, and perhaps thousands, of people."

Provided by University of Leeds