Southeast England ahead on genetic tests for inherited eye conditions

31 March 2015

Close up of light shining in eye landscape

New research from The University of Manchester published in the *Journal of Community Genetics* reveals a stark variation in genetic testing services for inherited eye disease in England.

The study, which was part-funded by Fight for Sight, shows that service provision in the North-east is much lower than expected based on population size and demographics, while in London and the South-east, it's much higher.

Genetic tests have been available on the NHS for over a decade for a limited number of inherited retinal dystrophies such as retinitis pigmentosa. However, new technology, known as 'next-generation sequencing' (NGS), has made it possible to map many genes simultaneously, saving time and money.

NGS means that many more patients with inherited retinal dystrophies could receive accurate genetic diagnoses and appropriate genetic counselling on how the condition might affect their families. But in order to plan for an expansion in NHS service provision, it is necessary to know how well existing services are working.

In the current study, the research team looked back at genetic testing in 2003-2011 for common mutations in six genes linked to dominantly inherited and X-linked retinitis pigmentosa. They quantified the variation in testing rate between the nine NHS regions in England, based on population size and demographics.

Results showed that by 2011, 4.5 per 100,000 males and 2.6 per 100,000 females in England had been tested. However, there was a wide variation in testing rates between the regions.

In north-east England there were approximately half as many tests as expected, whereas in the south-east, the rate was over a third more than expected. Only in the west Midlands and east England were test rates in line with the overall rate for England.

"It is likely that a number of factors have contributed to this variation in access to genetic services," said Professor Graeme Black from the Centre for Genomic Medicine at The University of Manchester, who led the research. "For instance, the at-risk population is not uniform across England; the way in which diagnostic tests are made available to clinicians varies between regions; and it's unclear whether there is variation in the way that clinicians and genetic counsellors explain the tests to patients.

"However, it is clear that we are unlikely to achieve equal access across the regions by chance. We need a consistent approach in providing information to patients about the availability and perceived value of testing and we need a strong evidence base to support the value of genetic testing on grounds of clinical and economic utility.

"In this way we can begin to develop a single, national strategy that will make it possible to fulfil

the huge potential of next-generation sequencing to improve patient care and drive research forward."

The study is part of the wider £0.5 million REGARD programme, funded by Fight for Sight, which aims to build an optimal, patient-led model of care services for people with inherited retinal diseases. In one strand of research, the team has developed a new NGS test for inherited eye disease that targets 180 different genes.

The REGARD programme is particularly important as commissioning for genetic eye services changes within the NHS. The recently established NHS specialised commissioning task force requires firm evidence of patient outcomes and the cost-benefit case for improving services. Data from REGARD will help build the evidence needed, for example, to introduce the 180-gene test throughout the NHS.

"Whilst the variation in services across England is shocking, I'm glad to see that we now have some hard evidence with which to build the case for a clear NHS strategy on inherited retinal dystrophy," said Dr Dolores M Conroy, Director of Research at Fight for Sight.

"These results go hand in hand with developing the next-generation sequencing technology that enables specific diagnoses for patients and finds new targets for developing treatment. We still have a long way to go but this is a very necessary step along the way."

More information: "Variation in healthcare services for specialist genetic testing and implications for planning genetic services: the example of inherited retinal dystrophy in the English NHS." *J Community Genet*. 2015 Apr;6(2):157-65. DOI: 10.1007/s12687-014-0210-4

Provided by University of Manchester