

# Inherited retinal disorders now the leading cause of blindness

BY T O'NEILL

The recent paper in BMJ Open, from the UCL Institute of Ophthalmology, 'A comparison of the causes of blindness certifications in England and Wales in working age adults (16-64 years), 1999-2000 with 2009-2010' concludes that inherited retinal disorders (IRD) such as retinitis pigmentosa are now the leading cause of certifiable blindness among working age adults in England and Wales. Previously diabetic retinopathy / maculopathy was accepted as the leading cause and this change is believed to be attributable to the introduction of nationwide diabetic retinopathy screening programmes and improved glycaemic control. These findings are significant as they should place inherited retinal conditions as an even higher priority for research.

The researchers ascribe this to improvements in screening and care for people with diabetes rather than a real increase in IRD numbers, though they do say that "the progress made over the last decade in molecular genetics / diagnostics and the increasing avenues of research / clinical trials for IRD with widespread media coverage may plausibly have resulted in ... increased clinic visits and thereby registration, without a true increase in incidence rates."

"Inherited retinal disease, now representing the commonest cause of certification in the working age population, has significant clinical and research implications, including with respect to the provision of care and resources in the NHS and the allocation of research funding," said Mr Michel Michaelides, co-author of the study, Consultant Ophthalmologist at Moorfields Eye Hospital and a Clinical Senior Lecturer at the UCL Institute of Ophthalmology. "This will also importantly help to dispel the myth that inherited retinal disease is rare and thereby not relevant."

The two main causes of blindness certifications in this latest and previous studies are shown in Table 1.

RP Fighting Blindness, a national UK charity which funds pioneering

**Table 1: The two main causes of blindness certifications.**

	Study 1999/00	Study 2009/10	Change
Inherited retinal disorders	15.8%	20.2%	+4.4%
Diabetic retinopathy / maculopathy	17.7%	14.4%	-3.3%

medical research and provides a range of support services to people with retinitis pigmentosa (RP) and other related conditions, has welcomed these formalised findings, as they confirm diagnostic trends it had been noticing for some years.

David Head, Chief Executive at RP Fighting Blindness, commented: "Inherited retinal disorders such as RP have been a significant cause of sight loss in England and Wales for decades and this paper reinforces the importance of patient organisations such as RP Fighting Blindness. We hope this results in an increased awareness of inherited retinal dystrophies and stimulates further funding for both medical research and patient support services. From communicating with other leading patient groups and researchers, we feel confident that these findings will strengthen our public image and hammer home the importance of the research we support. It is vital for charities such as ours that the general public, as well as the sight loss community generally, understand why we must continue to fund cutting edge science."

Professor Sir Peng T Khaw, Director of the NIHR Biomedical Research Centre at Moorfields Eye Hospital and UCL Institute of Ophthalmology, said: "This is a very important finding. It shows that what were thought to be 'rare inherited retinal diseases' together are now the commonest cause of certifiable blindness in working adults. This emphasises the need for research and the development of new treatments such as novel drugs, gene, cell and bionic implants for these diseases. The UK government and charities have taken a leading role in developing therapies for these previously untreatable diseases, with the first person in the world treated with gene therapy for retinal disease here at the National Institute for Health Research

(NIHR) centre only a few years ago. This provides a great stimulus to encourage further patient support and research into all these exciting new treatment modalities, including the national rare disease initiatives, which give new hope to people with these conditions."

RP Fighting Blindness hopes that this serious recognition of inherited retinal blindness as being highly significant in the UK will lead to better testing and NHS funding for screening. Ultimately, this recognition would also eventually lead to a reduction in inherited blindness, with better testing and treatments now in development having the same effect as the introduction of nationwide screening programmes for diabetic retinopathy.

## RP Fighting Blindness

is an organisation dedicated to finding a cure for retinitis pigmentosa (RP), a hereditary disease of the retina which leads to deterioration of sight and ultimately to blindness. RP affects some 25,000 people in the UK alone. The charity seeks to stimulate and fund cutting-edge medical research and provides information and support services to anyone affected by RP. For further information, please visit [www.rpfightingblindness.org.uk](http://www.rpfightingblindness.org.uk)

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