

Inherited Retinal Disease: an horizon scanning review

NIHR Horizon Scanning Research &
Intelligence Centre

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NIHR Horizon Scanning Research & Intelligence Centre (NIHR HSRIC)

Aim:

to provide advance notice to the Department of Health (England), health service policy-making bodies and research funders of significant new and emerging technologies, up to three years prior to launch on the NHS.

Remit:

Pharmaceuticals, devices, diagnostic tests and procedures, surgical and other interventions, rehabilitation, public health and health promotion activities.

Patient & Public Involvement & Engagement at NIHR HSRIC

- Patient and Public Involvement & Engagement Strategy published in June 2013. Aims:
 - To identify areas of work where the HSRIC can build and strengthen mutually beneficial relationships with patients and the public, and professionals working within the PPI arena.
 - To add value to HSRIC work and outputs through patient and public involvement.
 - To ensure accessibility and effective dissemination of the work and outputs of the HSRIC through patient and public engagement.
 - To ensure PPI is strategic, meaningful and appropriate.
- Progress Report to end of 2014 published in March 2015
- Patient and Public Involvement & Engagement Strategy 2015 – 2016 published in April 2015
- <http://www.hsruc.nihr.ac.uk/for-the-public/>

Horizon scanning reviews - overview

- Aim to identify all relevant new and emerging health technologies being developed:
 - in a technology area
 - at a specific point on a patient pathway
 - across a disease area.
- Often in response to a customer request to:
 - inform commissioning and planning decisions
 - identify current research activity
 - identify the need for further research
- A number of reviews have also been produced in areas where the NHIR HSRIC has identified new or high levels of activity in an area or where there is uncertainty e.g. regenerative medicines.

Horizon Scanning for Inherited Retinal Diseases

- James Lind Alliance Sight Loss and Vision Priority Setting Partnership (PSP) identified a number of priorities that related to emerging health technologies:
 - Can a treatment to stop dry AMD progressing and/or developing into the wet form be devised?
 - Can new therapies such as gene or stem cell treatments be developed for corneal diseases?
 - Can a treatment to slow down progression or reverse sight loss in inherited retinal diseases be developed?
- Liaised with Fight for Sight to identify review aim:
to identify new and emerging health technologies that aim to slow or stop disease progression and/or reverse sight loss in people with inherited retinal diseases.

Review Inclusion Criteria

- Pharmacological technologies—where these were in clinical trials with a relevant patient group and clinically relevant patient outcomes; typically phase II and III clinical trials.
- Medical technologies—where these were ‘emerging’ (expected to be CE marked and/or launched within the UK within ~ 2 years), ‘new’ (CE marked and usually only available for clinical use for less than one year), in the launch or early post-marketing stages, or ‘new and poorly adopted’ (that is, technologies within 2 years of launch and available in only two or three UK National Health Service (NHS) centres).

Methods

- **Identification:** Between November 2013 and January 2014, potential treatments were identified using online searches, databases, suggestions from clinical experts, and patient and carer focus groups, and direct contact with commercial developers known to be active in this field.
- **Filtration:** Using inclusion criteria
- **Prioritisation and commentary on impact:**
 - Fight for Sight (www.fightforsight.org.uk) arranged and facilitated two patient and carer focus group discussions:
 - 10 people attended: 5 men and 5 women; 60% were adults affected by IRDs (all aged over 40) - 2 were blind, 3 visually impaired, and one still had good vision; 40% were parents of children (aged between 4-15 years) with IRDs - one was blind, one severely visually impaired, and two at early stages of sight loss.
 - 10 clinical experts were invited to comment on the technologies

Results: Identification

- 40 new and emerging technologies were identified :
 - 10 medical technologies;
 - 9 gene therapies;
 - 5 pharmacological technologies;
 - 5 regenerative and cell therapies;
 - 11 very early developments (typically phase I or pre-clinical development stages)

Results: prioritisation

- Clinical experts provided comments on 30 technologies – the patient group provided comments on 7 of the technologies and general comments on policy and implementation.
- Both agreed that the technologies likely to have the most impact in the future are gene therapies and regenerative and cell therapies
- Safety, possible risks and complications important to patients and carers. Any remaining vision is precious so they would not want to do anything that risked damage and further loss of vision.
- The appearance of technologies was important to younger people in particular. Even where a technology is effective, if people do not wish to wear the new device because of its appearance, it will never be fully adopted
- Comments highlighted the hope that patients hold for some technology developments that are early in development, or less likely to succeed.

Dissemination

What's coming?

Horizon Scanning Centre

New and emerging technologies for inherited retinal disease

Inherited retinal diseases are now the most common cause of blindness in working age adults in England and Wales, and the second commonest in childhood. Currently, there is no cure or specific treatment. Management of these conditions consists of early diagnosis, specialised genetic counselling, treatment of any associated genetic conditions, as well as visual rehabilitation, support and training (e.g. to use visual aids).

We looked for new and emerging technologies that aim to slow or stop disease progression and/or reverse sight loss by consulting clinical experts and developers, and by searching special other online sources. Clinical experts and two patient focus groups (facilitated by Fight for Sight) reviewed the technologies we identified and commented on their potential for future impact (on patients, NHS systems and resources) and barriers to adoption. The patient focus groups provided valuable insights into the technologies from a potential user's perspective.

We found forty new and emerging technologies. These included nine medical devices, five pharmacological (drug) technologies, and five therapies. The other eleven technologies identified were at various stages of development. The majority are in or anticipated to be in clinical trials; some are already available to a small number of patients. Several technologies of particular interest to clinical experts and potential users and these are detailed in our [full report](#) that provides details of the specific technologies we identified on the HSC website.

Gene therapy

Gene therapy has the potential to slow and reverse retinal degeneration. It is the most effective in treating conditions before the degenerative process has affected large numbers of retinal cells. It promises both a reduced risk of disease progression and the potential for long term effectiveness following a single administration of a gene that delivers a new gene that could be more cost effective than repeated administrations. Most inherited retinal degenerations result from mutations in specific genes and further genetic research could expand the range of conditions potentially treatable using this approach. Current trials are at early stages and follow up will be needed to understand the effectiveness and safety of the approach.

Medical devices

A number of innovative retinal implants were identified with potential to slow or stop disease progression and/or reverse sight loss.

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National Institute for Health Research

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New and emerging health technologies for inherited retinal diseases



James Lind Alliance
@LindAlliance

Follow

New work by the @OfficialNHSC and @fightforsightUK to identify treatments in the pipeline for 4 inherited retinal diseases
goo.gl/ad4o8W

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New Treatments on the Horizon to Treat Inherited Retinal Diseases

16 July 2014

The National Institute for Health Research (NIHR) Horizon Scanning Centre (HSC), working with Fight for Sight, has identified 40 potential new treatments which are currently being developed for inherited retinal diseases in their latest horizon scanning exercise. These potential treatments may be able to help thousands of patients.

Inherited retinal diseases are now the most common cause of blindness in working age adults in England and Wales and the second most common in childhood. Currently, there is no cure or treatment.

Fight for Sight, the main UK charity dedicated to funding pioneering eye research to prevent sight loss and treat eye disease, was keen to help support the NIHR. In 2012-13, the charity co-ordinated the James Lind Alliance (JLA) Sight Loss and Vision Priority Setting Partnership (PSP) looking to set priorities for eye research. In this major consultation those with first-hand experience of sight loss (patients, relatives, carers and eye health professionals) were asked what were the most important questions they felt should be addressed by medical research.

The top priority in relation to inherited retinal diseases was: "Can a treatment to slow down progression or reverse sight loss in inherited retinal diseases be developed?"

UNIVERSITY OF Southampton

Health Research and Studies Coordinating

the Horizon Scanning Centre for Inherited Retinal Diseases

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Health Research



National Institute for Health Research

UNIVERSITY OF BIRMINGHAM

Feedback from patients and carers

- Participants were really impressed, if not slightly overwhelmed, by how much is going on.
- Because the information was to be read by people with sight loss, tables were inaccessible for those who need to use the main reading software. Fight for Sight removed the tables before circulating to patients and simplified the language where possible to make it more accessible.
- The information contained in the report was very complex for a patient audience. The use of language and jargon was exclusive meaning that only those with a scientific background were able to fully understand the information.
- Some words which appear to be 'lay' can have different meanings to different people e.g. bionic
- The information as currently presented is too distant from the needs of patients. They want to know how it applies to them and their condition.

Feedback from Fight for Sight

- We have been able to share the horizon scanning report with our research community and condition specific support charities to help them understand better the landscape in which they are working.
- The report alongside the PSP has allowed us to engage with the key funders of eye research to identify the gaps in research and where investment is needed.
- We will also be able to focus more on effecting the eye research funding landscape.
- The exercise of running the focus groups, expanded our understanding of the experience of inherited retinal diseases and why many have focused on the care and support they can receive rather than believing in the power of research to make a difference.

Further information

- Full report available at <http://www.hsric.nihr.ac.uk/>
- Paper published in Eye –
 - Smith J, Ward D, Michaelides M, Moore A T and Simpson S. [New and emerging technologies for the treatment of inherited retinal diseases: a horizon scanning review](#). Eye advance online publication 26 June 2015; doi: 10.1038/eye.2015.115
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Any questions.....

