

Patient-Led Initiative to Identify the Molecular Cause of Rare Inherited Retinopathies

IRDiRC – April 16, 2013 Dublin
Avril Daly, Fighting Blindness/EURORDIS



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Our vision is to cure blindness, support those experiencing sight loss and empower patients.



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Darkness to Light and Hope

Ireland and the Irish people play a pivotal role in global research into treatments and cures for blindness.

Our research is patient led!



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The Fighting Blindness Story

- Started in 1983 as a group of families affected by blindness getting together for support.
 - Now a global leader in the search for cures and treatments for genetically inherited and age-related forms of blindness.




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Fighting Blindness Research

- In 1989, researchers in TCD funded by Fighting Blindness discovered the first gene responsible for causing retinitis pigmentosa. Since then, over 35 other genes have been discovered, as well as hundreds of individual mutations.
- Genetic research has been successful here because Ireland has so many large families, making it easier to trace the genetic course of the disease.
- Since this first project, we have continued to progress our knowledge of degenerative eye conditions and further this research into better diagnosis, treatments and cures.



R.P. Ireland—Fighting Blindness
NEWSLETTER MAY 1992 Vol. 10. No. 1

CONGRATULATIONS TO TRINITY TEAM



Back Row: Karen Molloy, Pat Manley, Rajendra Kumar-Singh,
Second Row: Mark Lawlor, Prof. Peter Humphries, Dr. Peter Creighton, Cathy Wang,
Third Row: Dr. Paul Kenna, Beth Sharp, Siobhán Jordan,
Front Row: Marian Humphries, Dr. Denise Shiels, Dr. Jane Farrar.

We would like to extend congratulations to Professor Humphries and the Trinity research team on their recent breakthrough in the discovery of a genetic mutation in the protein known as peripherin and we wish them continued success in their quest to find a treatment and cure for R.P.

“CO-OPERATION AND CO-ORDINATION CAN CURE”



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CURE



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Rare & Inherited Eye Conditions We Are Concerned With

- Retinitis Pigmentosa
- Leber's Hereditary Optic Neuropathy
- Usher Syndrome
- Leber's Congenital Amaurosis
- Stargardt's Disease
- Choroideremia
- Retinoschisis
- Retinopathy of Prematurity



Our Research Activities

- Since 1983, we have funded 40 research projects in universities and colleges throughout Ireland, with one in the UK.
- We advocate for sustainable infrastructures to expedite the development of therapies.
- We advocate to ensure equitable access to these therapies when they become available.



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Today's Research, Tomorrow's Cures

“When Fighting Blindness started, there were only questions. Today, they have the tools for a cure in their hands.”

- Paul Norton,
Fighting Blindness Member



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SUPPORT



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“Losing your sight is like losing someone you love. It’s like a part of your heart is gone. You have to adapt both emotionally and mentally.”

- Gerry Kerr,
Fighting Blindness Board Member



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Insight Counselling – Services Available



- Individual therapy
- Family member group therapy
- Spouse group therapy
- Outreach patient workshops
- Regular monthly group meeting in Cork
- Weekly support
- Clinical settings
- Phone appointments



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EMPOWER



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Empowering Patients Through EU Outreach

- In 1984, Fighting Blindness became a member of Retina International, and in 1988, FB worked with other organisations to establish GRDO in Ireland to advocate for a National Centre for Medical Genetics to be established. NMCG was founded in 1994.
- In the 1990s, FB began to investigate what was happening with groups concerned with rare diseases in the EU and how activities in MS could apply to our national systems. FB joined EURORDIS in 1997.
- FB built coalitions in Ireland to advocate for better funding and infrastructure for research and access to care pathways for people (IPPOSI, MRCG).



Understanding the Complexity of Genetic Eye Disease



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Gene Identification for Inherited Retinal Conditions

- We now know that the 1989 discovery of rhodopsin as the first gene to be linked to retinitis pigmentosa was the tip of the iceberg. Retinal degenerations (RD) are some of most complex genetic disorders of all. For example, in rhodopsin alone, there are 150 known mutations.
- Mutations in genes currently identified are believed to account for 60% of retinal degenerations. Current work is focused on identifying the remaining genes.
- As we enter the age of clinical trials, there is a urgent need to establish the molecular causes of disease in our patient population.
- For the individual, this will lead to a complete diagnosis of an individual's condition and a better prognosis of the way the disease may develop in the future.



Building a Bridge to Treatments

- In Ireland and abroad, there are numerous clinical trials planned for gene therapy and other possible treatments for retinal degenerations.
- A prerequisite for inclusion in such trials is a clinically and genetically well characterised population, as it is essential that researchers can identify patients with the particular gene mutation in question so that candidates for the trials can be selected.
- This approach will screen for known genes previously implicated as causative of RD together with a number of candidate genes and will also document the incidence of extremely rare gene mutations.



Pilot Study: Trinity College, Dublin

- Next generation exome sequencing pilot study funded by the Health Research Board, Ireland (HRB). Collaborative project between Professor Jane Farrar of TCD and Mr. Paul Kenna, RVEEH.
- Our researchers have validated gene mutations in control samples from the Netherlands, and 10 Irish patients have been successfully screened.
- This pilot study has successfully identified and flagged causative mutations, paving the way for a reproducible and reliable methodology for large-scale whole exome sequencing and analysis.



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Target 3000: Gateway to Vision

- Target 3000 launched in March 2012. The goal of the project is to identify causative mutations across the entire Irish inherited RD population.
- The challenge is that there is no register of exactly how many people in Ireland are affected by RD, but current estimates put it at 3000 individuals.
- We have successfully staged a series of patient engagement programmes and we have a well-informed and knowledgeable patient base who are extremely supportive of the initiative.
- Funding strategy
- Roll out



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13 | Presentation title | Preserve Sight | Gene | Surveys | Acknowledgements Only
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Sustaining a Long-Term Initiative

- As a patient-led organisation with experience of funding research over three-year cycles, this represents a long-term commitment.
- This project is a legacy and does not have a finish date, as we will continue to add to the data.
- We have a responsibility to our patient base to carry out the process effectively.
- Understanding data protection, consent and ownership of materials: Ensuring our register is compliant, can be part of an ERN and is built for sustainability from an IT perspective.



Patients Lead RD Research *

From EURORDIS 2010 Survey on Patient Organisations (POs) and Research: 309 POs respondents, 110 diseases

- POs have a high commitment to research and are keen observers of all its areas from basic, therapeutics, social and human sciences.
- POs understand that research is a long-term process and engage in sustainability of their projects.
- POs play an important role as catalysts of research.
- POs provide two types of essential support to research: Financial and Non-Financial. POs are natural go-betweens for scientists from various fields from the more basic research to therapeutic applications, crucial support in clinical.
- POs have limited budgets, “fill the gaps” by supporting the type of research that appears less attractive to the public or private sector.



Patients Lead RD Research *

From EURORDIS 2010 Survey on Patient Organisations (POs) and Research: 309 POs respondents, 110 diseases

- POs are fully aware that to understand the disease (basic research) and to cure the disease (therapeutic research) are two axes that must be supported concurrently, as progress in one contributes to the other.
- The long history of several POs and their continuous commitment to basic research demonstrates that they understand that research is a slow and long-term process.
- POs do not want to (and they cannot) replace public or private research institutions, but rather collaborate with them as fully recognised partners bringing important specific contributions.



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Who We Work With

Internationally

EURORDIS www.eurordis.org

EPF <http://www.eu-patient.eu/>

Retina International <http://www.retina-international.org/>

In Ireland

GRDO www.grdo.ie

IPPOSI www.ipposi.ie

MRCG www.mrcg.ie



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Where to Find Us

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Thank You



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