

# Royal Victoria Eye & Ear Hospital Research Foundation

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# **COMMITTEE MEMBERS**

Chairperson	Noel Horgan
Treasurer	Mark Cahill
Lecturer in Ocular Genetics	Paul Kenna
Committee Member	Aoife Doyle
Committee Member	Conor Murphy
Committee Member	Susan Kennedy
Committee Member	Andra Bobart-Hone
Committee Member	David Mooney
Committee Member	
Committee Member	Marie Tighe
Chief Electrodiagnostic Technician	Hilary Dempsey
Senior Counselling Psychologist	
Research Registrar	Micheal O'Rourke
Research Nurse	Claire English
Research Nurse	Anne Caslin
Research Coordinator	Emma-Jayne Verner
Research Photographer	Laura Hughes
Research Secretary	Cathy King
Research Accounts	Josephine Lavelle

# SERVICES AVAILABLE

Clinical Electrophysiology Service – to assess retinal and brain stem function Colour Vision Testing – Farnsworth Munsell 100 hue Dark Adaptation Visual field testing for glaucoma patients (Humphrey Analyser) A-Scans for assessing power of lens implants for cataract surgery Assessment of visual function in Medico Legal cases Full eye examination for patients and their families suffering from Retinitis Pigmentosa and other inherited retinal degenerations Pattern E.R.G for pre-clinical visual function loss in Glaucoma Optical Coherence Tomography for Assessment of Macular Disease and Glaucoma GDX for Retinal Nerve Fibre assessment in Glaucoma Counselling Services for visually impaired people and their families

#### INTRODUCTION



Since its establishment in 1974 the Research Foundation at the Royal Victoria Eye & Ear Hospital, Dublin has had a particular interest in various forms of inherited and metabolic retinal diseases. It was amongst the first centres in the country to develop electrodiagnostics to aid in the diagnosis and assessment of patients with blinding conditions. Today, the Unit possesses state-of-the-art electrodiagnostic equipment and can carry out the full range of electrophysiological tests of retinal and visual function to the highest international standards. The Research Foundation has expanded over the last number of years and includes research on all types of eye disease including retinal disease, ocular oncology, ocular inflammation, corneal disease and eye complications of systemic disease, in addition to research in diseases of the ears, nose and throat.

The Research Foundation has a long history of collaboration with the Ocular Genetics Unit in Trinity College Dublin where researchers were instrumental in identifying the first disease-causing gene in any form of Retinitis Pigmentosa (RP), rhodopsin, in 1989. These genetics studies, which are on-going, have contributed significantly to research world-wide in this field. The Research Foundation is also involved in studies to determine the genetic basis for age related Macular Degeneration as a significant cause of vision loss in patients over the age of 50. A Phase1B trial of an orally delivered medication for a form of autosomal dominantly inherited Retinitis Pigmentosa has recently commenced. This is the first gene-informed trial for any form of dominantly inherited Retinitis Pigmentosa world-wide. At the Foundation, we aim to continue to be at the forefront of research which will eventually result in the development of sight-saving treatments for patients.

The Research Foundation is the principal investigating site in Ireland for a number of drug trials involving anti-VEGF medications for the treatment of retinal diseases. These Phase 3 clinical trials investigate the use of Ranibizumab in patients with Macular Oedema, Branch Retinal Vein Occlusion and Central Retinal Vein Occlusion. The Research Foundation has funded a trial project also to improve the diabetic retinopathy screening service in the hospital. It is hoped that some of the lessons learned from this programme would be adapted by the proposed National Screening Programme which is scheduled to start by the HSE towards the end of 2012.

The ocular inflammation/cornea service of the Royal Victoria Eye & Ear Hospital works in partnership with the National Institute for Cellular Biotechnology (NICB) at Dublin City University, the Department of Immunology at the Royal College of Surgeons Ireland and the Department of Rheumatology at St Vincent's University Hospital. This collaboration brings together clinical and scientific skills from a range of disciplines that are helping to improve our understanding of a number of inflammatory eye conditions and corneal diseases.

The annual Eithne Walls Research Meeting was held on Thursday 21st June 2012. This meeting, held in memory of Dr Eithne Walls, is a forum for the young doctors in the hospital to present their research and to develop their ability to undertake research and present it to their peers. Dr Farahida Ibrahim was awarded the Eithne Walls Memorial Medal for her paper on "Diabetic Retinopathy Screening Using Manual and Automated 3 Step Grading of Fundus Photographs with Supplementary OCT in an Outpatient Setting" and Sorcha Ni Dhubhghaill was awarded the Research Foundation Clinical Prize for her work on "Genetic & Demographic Risk Factor Profile in the Irish Age Related Macular Degeneration Population".

Members of Eithne Walls Family, staff of the hospital and a number of dancers from Riverdance productions ran the Mini Marathon on Monday 4th June 2012 to raise funds for the Eithne Walls Research Fund. These events continue to grow from strength to strength each year and we look forward to continuing to build on these important dates in the hospital's calendar, which allow us to remember our dear colleague and foster continued interest in ophthalmic research.

The Royal Victoria Eye & Ear Hospital Research Foundation looks forward to having further submissions for research projects undertaken at higher degree level by young doctors in training. At the back of the progress report there is a covenant form and we would hope that people would consider donating to the Foundation using the covenant method.

# 2012

# **CURRENT RESEARCH**

# Genetics Research

Working with Irish families with autosomal dominant forms of the inherited retinal degeneration, Retinitis Pigmentosa (RP) and in collaboration with the Ocular Genetics Unit at Trinity College Dublin, Foundation researchers were instrumental in identifying the first disease-causing gene in any form of RP, rhodopsin, in 1989. Subsequently other RP causing genes were identified in families who were characterised at the Foundation, namely the Peripherin/RDS and the mitochondrial MTTS2 genes. Most recently, a mutation was identified in the RPE65 gene in an Irish family, ascertained at the Foundation, with an autosomal dominant form of Retinitis Pigmentosa.

At present, patients with inherited retinal degenerations face inexorable loss of vision, in many cases resulting in total blindness. However, as a result of the molecular genetic advances with which the Foundation has been intimately involved, realistic prospects now exist to give hope that treatments will become available in the foreseeable future. At the Foundation, we aim to continue to be at the forefront of research which will eventually result in the development of sight-saving treatments for these patients.

#### **Projects**

#### 1) Age-related macular degeneration (AMD)

Age-related macular degeneration is one of the most common causes of visual impairment in the Irish population. Although many risk factors for the condition have been identified, recent research has highlighted the significance of genetic factors in increasing the risk of an individual developing this disease. A large-scale study into the genetics of AMD is presently underway at the Foundation to characterise the importance of genetic risk factors in the Irish AMD population.

#### 2) Retinitis Pigmentosa (RP)

#### ADRP with Choroidal Involvement associated with Asp477gly Mutation within the Rpe65 Gene

In a collaborative research effort between the Research Foundation at the Royal Victoria Eye and Ear Hospital and the Ocular Genetics Unit at Trinity College Dublin investigators identified a new gene responsible for a form of autosomal dominant Retinitis Pigmentosa. The results were published in October 2011. Linkage testing using Affymetrix 6.0 SNP Arrays mapped the disease locus in TCD-G, an Irish family with autosomal dominant retinitis pigmentosa (adRP) ascertained and clinically characterized at the Foundation to an 8.8 Mb region on 1p31. Candidate gene and exome sequencing resulted in the identification of an Asp477Gly mutation in exon 13 of the RPE65 gene tracking with the disease in TCD-G. The Asp477Gly mutation was not present in Irish controls, but was found in a second Irish family identified at the Foundation, provisionally diagnosed with Choroideraemia, but in whom no Choroideraemia gene mutation had been found. Mutations in RPE65 are a known cause of recessive Leber congenital amaurosis (LCA) and recessive RP, but no dominant mutations have been reported. This important paper (A dominant mutation in RPE65 identified by whole-exome sequencing causes retinitis pigmentosa with choroidal involvement. Bowne SJ, Humphries MM, Sullivan LS, Kenna PF, et. al. Eur J Hum Genet. 2011 Oct;19(10):1074-81. doi: 10.1038/ ejhg.2011.86. Epub 2011 Jun 8) documented for the first time a dominantly acting mutation in this gene in 2 families with a clinical picture somewhat reminiscent of Choroideraemia. Approximately 20% of patients with a diagnosis of Choroideraemia do not have mutations in the X-linked gene for Choroideraemia. It is likely that mutations in RPE65 may be responsible for the disease in these patients.



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Gene therapy for LCA patients with RPE65 mutations has shown great promise, raising the possibility of related therapies for dominant-acting mutations in this gene.

#### 3) Registry of Irish Patients with Leber Congenital Amaurosis and Early Onset Severe Retinal Dystrophy Wellcome Trust – HRB Centre for Clinical Research, Molecular Medicine Ireland, P. Kenna (RVEEH), M. Cahill (RVEEH), D. Keegan (MMH), I. Flitcroft (MMH).

This collaborative research co-ordinated by Mr. P. Kenna of the Research Foundation at the Eye and Ear Hospital and Mr. David Keegan of the Mater Misericordiae Hospital aims to identify Irish patients with Leber Congenital Amaurosis or Early Onset Severe Retinal Dystrophy (EOSRD), conditions in which gene mutations in the RPE65 gene have been identified. The aim is to document the incidence of these rare inherited disorders in the Irish population and to identify those individuals who might benefit from the encouraging results of the on-going clinical trials of gene therapy in these conditions.

#### 4) Genetic Characterisation of a population of Irish Retinal degeneration patients

The study, funded by the Health Research Board of Ireland aims to analyse the DNA of a cohort of Irish patients with a variety of inherited retinal degenerations using next generation gene sequencing technology. This is a collaborative effort between Mr. P. Kenna of the Research Foundation, The Royal Victoria Eye and Ear Hospital and Prof. G. Jane Farrar at the Genetics Department, Trinity College Dublin.

As a result of this work a Phase 1b trial of an orally delivered 9-cis retinal analogue involving Irish patients with this condition and sponsored by QLT Inc. of Canada has just commenced. This is the first geneinformed treatment trial for any form of autosomal dominant Retinitis Pigmentosa and further highlights the importance of the long-standing work of the Foundation in this area.

Patients with a variety of inherited retinal degenerations who have been clinically characterised at the Foundation have been recruited over the past 2 years into a research project funded by the Health Research Board for next generation genome sequencing. This ambitious project will hopefully result in the identification of disease-associated gene mutations in these patients. While the study is designed to give a picture of the spectrum of genes causing inherited retinopathies in a cohort of Irish patients the results will be of value to the participants and allow them to consider participating in future gene-based treatment trials. Undoubtedly, over the next 5 to 10 years, as a result of the progress in elucidating the genetic basis of many of these currently untreatable disorders, treatment options are likely to become available. The Foundation is proud of its contribution over many years to this pioneering work.

Mr. Eoin Hanney, an Irish clinical genetics student at the University of Wales, is conducting a survey of patients attending the Foundation with Retinitis Pigmentosa into their experiences and attitudes to genetic testing. This important study will hopefully influence the development of future genetic testing services in Ireland. This is undoubtedly going to become a major clinical need for Irish patients as more treatment options, many based on knowledge of the disease causing gene, become available.

- Mr Paul Kenna

# Retinal disease

The Research Foundation has funded a trial project also to improve the diabetic retinopathy screening service in the hospital. The Research Foundation is the principal investigating site in Ireland for a number of drug trials involving anti-VEGF medications for the treatment of retinal diseases. This collaboration with pharmaceutical companies is a new avenue for the Research Foundation and we hope to develop this facility in the future.

## **Projects**

#### 1) Objective three-steps grading of digital fundus photographs of diabetic retinopathy.

This project is using objective three-step grading of fundus photographs of patients with diabetic retinopathy. 450 hospital-based patients have been screened to date. The three step process increases the quality of the grading of the screening program. A quality component has been the use of OCT to detect diabetic macular oedema. This project will be expanded in 2012 to include 2 primary practices located in Ranelagh and Churchtown. It is hoped that some of the lessons learned from this programme would be adapted by the proposed National Screening Programme which is scheduled to start by the HSE towards the end of 2012.

#### 2) RETAIN Study

This is a phase 3 clinical trial investigating the use of Ranibizumab (anti-VEGF medication) for the treatment of macular oedema. The Research Foundation is the principal investigating site for the study. The project started in January 2011 and will continue until January 2013. 5 patients have enrolled in the study which requires monthly visits and extensive investigation of each patient.

#### 3) BRIGHTER Study

This is a phase 3 clinical trial examining the use of Ranibizumab in the treatment of macular oedema secondary to branch retinal vein occlusion. The Research Foundation will be the principal investigator on site for this trial in Ireland. It is proposed that recruitment for the study will commence in August 2012.

#### 4) CRYSTAL Study

This is a phase 3 clinical trial examining the use of Ranibizumab in the treatment of macular oedema secondary to central retinal vein occlusion. The Research Foundation will be the principal investigator on site for this trial in Ireland. It is proposed that recruitment for the study will commence in August 2012.

- Mr Mark Cahill

# Pathology / Oncology

This is a collaboration between the pathology services at the Royal Victoria Eye & Ear Hospital and Dublin City University looking at potential prognostic biomarkers in eye cancer. The pathology and ocular oncology unit at Royal Victoria Eye & Ear Hospital are also working closely with the National Institute for Cellular Biotechnology NICBI on a Research Foundation supported research programme in identifying specific proteins in patients with eye melanoma that help to predict the spread of cancer outside the eye, with the aim of improving long term patient survival.



#### **Projects**

1) Proteomic analysis of tumours and vitreous fluid from uveal melanoma P.Ramasamy (MD Study), Prof C.Murphy, Mr N.Horgan, Dr P.Meleady, M.Clynes

2) Establishment of a database and tissue microarray bank of 500 archived uveal melanoma tumours funded by SFI grant to the consortium Molecular Therapeutics for Cancer, in association with Paul Moriarty and Mr Noel Horgan. In collaboration with Professor Clynes's Research Group (DCU) and with Professor Beatty's Group (WTI), Mr Moriarty and Mr Horgan are conducting a study of uveal melanoma proteomics funded by the Research Foundation and donation from Professor Beatty.

3) A collaboration with Dr Brian Hennessey, RCSI on molecular sequencing of head and neck tumours funded by SFI grant to the consortium Molecular Therapeutics for Cancer.

- Professor Susan Kennedy

#### Ocular immunology, inflammation and corneal research

The Research Foundation supports long term collaboration between the ocular inflammation/cornea service of the Royal Victoria Eye & Ear Hospital, the National Institute for Cellular Biotechnology (NICB) at Dublin City University, the Department of Immunology at the Royal College of Surgeons Ireland and the Department of Rheumatology at St Vincent's University Hospital. This collaboration brings together clinical and scientific skills from a range of disciplines that are helping to improve our understanding of a number of inflammatory eye conditions and corneal diseases.

#### **Projects**

#### 1) Herpes simplex keratitis research

Herpes simplex keratitis (HSK) represents the single most important inflammatory disease of the cornea with respect to its impact on vision and health related quality of life. It is characterised by repeated episodes of inflammation in the cornea, the clear window at the front of the eye, which leads to corneal scarring and, in many cases, loss of vision. It is caused by the common cold sore virus, known as Herpes Simplex Virus type 1. Our research into this condition aims to improve our understanding of how the herpes virus interacts with our immune system, particularly our innate immunity which is our first line of defense. By improving our understanding of this interaction, we hope to identify new targets for treatments of this disease and improve the outlook for sufferers of HSK.

#### **Projects**

1) Evasion of the innate immune response by herpes simplex virus in the cornea: molecular mechanisms mediating interferon down regulation and virus survival

2) Effect of corneal Herpes Simplex Virus-1 infection on Toll-Like Receptor expression in human peripheral blood mononuclear cells

Investigators: Conor Murphy, David Shahnazaryan, Ciaran de Chaumont, Con Malone, Joan Ni Gabhann and Caroline Jefferies.

Affiliations: Royal Victoria Eye and Ear Hospital and Royal College of Surgeons in Ireland.

# Giant cell arteritis research

Giant cell arteritis (GCA) is the most common form of primary systemic vasculitis (inflammation of blood vessels). Patients with GCA endure significant morbidity associated with the disease and its treatment. Significant deficits exist in our understanding of this disorder, particularly the underlying causes and mechanisms of the disease. Through a multidisciplinary approach we are developing a large database of GCA patients which will facilitate clinical and translational research studies, audit and participation in international multi-centre clinical trials. We are performing radiological assessments of patients with suspected GCA, including MR angiography, CT angiography and temporal artery ultrasound, in order to improve diagnostic accuracy. In addition, we are performing laboratory investigations on the blood and temporal artery biopsy specimens of patients with GCA with the aim of improving our understanding of how this disease occurs at a molecular level. This will hopefully help us to use more specific and effective treatments in the future and help us to understand why some patients do not respond well to conventional therapy with steroids. In the future we will perform genetic studies that we hope will provide information about the underlying causes and long term prognosis of the disease.

## **Projects**

1) Improving outcomes in giant cell arteritis through clinical collaboration

2) Increasing diagnostic accuracy in GCA through imaging

3) Blood Vessel Instability and Oxidative Damage in Giant Cell Arteritis

4) Investigating pro-inflammatory mechanisms of GCA using an ex-vivo temporal artery culture model

*Investigators*: Conor Murphy, Eamonn Molloy, Ursula Fearon, Douglas Veale, Geraldine McCarthy, Lorraine O'Neill and Jim Meaney.

*Affiliations:* Royal Victoria Eye and Ear Hospital, Royal College of Surgeons In Ireland, St Vincent's University Hospital, University College Dublin, St James' Hospital and the Mater Misericordiae Hospital.

# Primary Sjögren's Syndrome Research

Primary Sjögren's Syndrome (pSS) is an autoimmune disease that destroys the specialised secretary glands that produce saliva and tears, causing dry eyes and dry mouth as well as generalized symptoms of aches, pains and lethargy. There is currently no cure for pSS and the exact cause is unknown. In this study we are expanding our understanding of this disease at a molecular level by investigating the role of toll-like receptors on blood cells from patients with pSS, as well as minor salivary gland biopsies (when taken for diagnostic purposes), tear samples and ocular surface washings. This study is being funded jointly by the Health Research Board (HRB) and the RVEEH Research Foundation.



#### Project

1) Profiling Toll-like receptor responses in patients with primary Sjögren's syndrome

*Investigators:* Conor Murphy, Caroline Jefferies, Con Timon, Eamonn Molloy, Joan Ni Gabhann. *Affiliations:* Royal Victoria Eye and Ear Hospital, St Vincent's University Hospital and Royal College of Surgeons in Ireland.

## Anterior uveitis and spondylarthropathy research

Acute anterior uveitis (AAU) is characterised by the acute onset of inflammation in the front compartment of the eye, leading to pain, light sensitivity and blurred vision. It is a common reason for presentation to ophthalmic emergency departments. In approximately half of cases there is an identifiable systemic disease, most commonly the seronegative spondyolarthropathies (SpA). This is a group of inflammatory joint diseases that predominantly affect the spine but have many other manifestations including skin and bowel problems.

This collaboration with St. Vincent's Unversity Hospital Department of Rheumatology has led to the development of an assessment algorithm called the Dublin Uveitis Evaluation Tool (DUET) that enables the earlier recognition of SpA. With early detection come early and more effective treatment and disease control, and hence better quality of life. Our laboratory studies on the causative mechanisms of AAU are also providing us with some fascinating insights into the disease.

#### **Projects**

**1)** Validation of the Dublin Uveitis Evaluation Tool (DUET), a new algorithm for the detection of undiagnosed spondylarthropathies in patients presenting with acute anterior uveitis in a primary care ophthalmology setting.

**2)** To investigate the role of regulatory microRNA and dendritic cell function in the pathogenesis of acute anterior uveitis

3) Prospective evaluation of vision and health-related quality of life in patients with acute anterior uveitis.

**4)** Peripheral blood mononuclear cell activation status and functional characteristics in patients with acute anterior uveitis.

*Investigators:* Conor Murphy, Micheal O'Rourke, Pathma Ramasamy, Muhammad Haroon, Mary Connolly, Douglas Veale, Ursula Fearon, and Oliver Fitzgerald.

*Affiliations:* Royal Victoria Eye and Ear Hospital, Royal College of Surgeons In Ireland, St Vincent's University Hospital, and University College Dublin.

# Genetic analysis of patients with congenital hereditary endothelial corneal dystrophy (CHED)

CHED is a very rare inherited disease that manifests early in life with clouding of the front window of the eye, the cornea, as well as poor vision and nystagmus (wobbly eyes). It typically presents between the age of 2 and 5 years and causes lifelong bilateral blindness. In previous work, Dr Collette Hand, Lecturer in Genetics at UCC, located the abnormal gene causing CHED to chromosome 20. Since then, the affected gene has been identified and called SCL4A11. A large number of mutations in this gene have been described in different populations. In this study, we wish to identify the nature of the mutation in the SCL4A11 gene in a large Irish family with the condition.

Investigators: Conor Murphy, Mairide McGuire, Collette Hand.

*Affiliations:* Royal Victoria Eye and Ear Hospital, Royal College of Surgeons In Ireland, and the Department of Molecular Genetics, University College Cork.

- Professor Conor Murphy

# Corneal Tissue Engineering Research

#### **Projects**

**1)** Gene expression profile of cultured limbal-cornea epithelial stem cells and cultured limbal fibroblast cells.

2) The role of cell culture set up in the growth of cultured limbal-cornea epithelial stem cells.

*Investigators:* William Power, Conor Murphy, Martin Clynes, Finbarr O'Sullivan & Clare Gallagher *Affiliations:* Royal Victoria Eye and Ear Hospital, Royal College of Surgeons In Ireland

- Mr William Power

# Systemic Disease

The Research Foundation was chosen by Servier, a leading French pharmaceutical company, to do a Phase III multi-centre clinical trial to analyse the retinal effects of Ivabradine, an IF inhibitor used as cardiac rate limiter in heart disease. This trial commenced in October 2009 and will be ongoing for the next three years.

#### **Projects**

**1)** Long-term (3 years) ophthalmic safety and cardiac efficacy and safety of Ivabradine administered orally. P.Kenna, L.Cassidy and H.Dempsey

- Professor Lorraine Cassidy



# Medical Research Charities Group

The Medical Research Charities Group is a collaboration of Medical Research and patient support charities similar to the Research Foundation. It is the aim of this organization to support, promote, and fund medical research for improved Health outcomes. The Medical Research Charities Group and the Health Research Board has approved Joint Funding Schemes for financing a number of research studies. Going forward the Research Foundation will be submitting a number of projects for funding in 2012. The Research Foundation is delighted that this scheme will supplement the research work in the diagnosis, treatment and prevention of ocular disease.

#### Flora Women's Mini-Marathon

On 1 June 2009 Dr Eithne Walls, along with her two friends and colleagues Dr Aisling Butler and Dr Jane Deasy, was a passenger on the ill-fated flight AF447 which disappeared over the Atlantic as it travelled from Rio de Janeiro to Paris.

Eithne was a doctor at the Royal Victoria Eye and Ear Hospital and just starting out on her exciting journey to fulfilling a lifelong ambition of becoming an eye surgeon. She was a gifted, vibrant and special person and her loss has had a profound impact on all who were privileged to know her. Following this tragic accident Eithne's family established a research fund at the Research Foundation in her memory. This fund helps to support the vital research work of the Research Foundation into eye disease.

In 2012 a group of Eithne Walls Family, staff of the hospital and a number of dancers from Riverdance productions ran the Mini Marathon on Monday 4th June 2012 raising €8000 for the fund. Last year a team of 40 runners joined by staff from the hospital participated again raising €7300 for the Research Foundation.



#### Dublin City Marathon

Dr Martha McGann ran the Dublin City Marathon on Monday 29th October 2012 to raise funds for the Eithne Walls Research Fund. Not only did Martha meet her desired time but she also managed to raise over €2000 for the Research Fund. The Research Foundation would like to personally thank and congratulate Martha on such an achievement. It is only through the generous support of fundraisers like Martha that we can continue vital research work.



# Eithne Walls Research Meeting

The third annual Eithne Walls Research Meeting was held on Thursday 21st June 2012 in the Education & Conference Centre where all basic specialist trainees in ophthalmology in the hospital were invited to make a presentation based on their clinical or basic science



research in ophthalmology. Dr Farahida Ibrahim was awarded the Eithne Walls Memorial Medal for her paper on "Diabetic Retinopathy Screening Using Manual and Automated 3 Step Grading of Fundus Photographs with Supplementary OCT in an OPDR Setting". A new prize was also awarded this year; the Research Foundation Clinical Prize was awarded to Sorcha Ni Dhubhghaill for her work on "Genetic & Demographic Risk Factor Profile in the Irish AMD Population".

The meeting was a great success, and followed on from the Women's Mini Marathon in which a large number of Eithne's family, friends and former colleagues participated to fundraise for the research fund established in her memory. These events continue to grow from strength to strength each year and we look forward to continuing to build on these important dates in the hospital's calendar, which allow us to remember our dear colleague and foster continued interest in ophthalmic research.

# Research Foundation funded Registrars

The Research Foundation funded Dr Micheal O'Rourke's research on 'The Dublin Uveitis Evaluation Tool (DUET) – an algorithum for earlier diagnosis of spondyloarthropathies by Ophthalmologists in acute anterior uveitis'.



Dr Pathma Ramasamy received funding from the Research Foundation on his project on 'Proteomic analysis of uveal melanoma tissue and vitreous fluid'. Pathma was delighted to receive the award for best presentation at the annual ICO conference, best presentation in the oncology section at EVER 2012 and a travel award from EVER 2012.



# Counselling Service at Research Foundation

A counselling and support service is provided to the Research Department in the Royal Victoria Eye and Ear Hospital every Wednesday morning from 10.00 a.m. to 1.00 p.m. for newlydiagnosed patients and their families and also for patients on follow-up visits.

This service is provided by Insight Counselling Centre, an initiative of Fighting Blindness. The Centre was established in 2002 with the objective of providing practical help, emotional support and psychotherapy to individuals experiencing sight loss, as well as to their families. Sight loss impacts in very many different ways and at many different levels on individuals and their families. The onset of sight loss can change one's relationship with self and others and



reaching out for support in these circumstances can be very beneficial. The Centre provides a safe place for exploring matters of concern and help in finding a way through what can be a difficult and frightening time.

On-going counselling is also available Monday to Friday at the Insight Counselling Centre premises at 4 South Great George's Street. The service is provided free of charge by Fighting Blindness and is open to referrals from the clinical staff.

Mary Lavelle Senior Counselling Psychologist Insight Counselling Centre 4 South Great George's Street Dublin 2

# Peer reviewed publications

Amin M, Ali R, Kennedy S, Timon C. Ear Nose Throat J (2012) Inflammatory myofibroblastic tumor of the nose and paranasal sinuses masquerading as a malignancy, 91 (5): E1-3

Browne B.C, Eustace A.J, Kennedy S, O'Brien N.A, Pedersen K, McDermott M.S.J. Larkin A.S.M, Ballott J, Mahgoub T, Sclafani F, Madden S, Kennedy J, Duffy M.J, Crown J, O'Donovan N, (2012) **Evaluation of IGF1R and phosphorylated IGF1R as targets in HER2-positive breast cancer cell lines and tumours**, Breast Can Res Treat, 136(3):717-27

Chadderton N, Palfi A, Millington-Ward S, Gobbo O, Overlack N, Carrigan M, O'Reilly M, Campbell M, Ehrhardt C, Wolfrum U, Humphries P, Kenna P.F, Farrar, G.F. (2012) Intravitreal delivery of AAV-NDI1 provides functional benefit in a murine model of Leber hereditary optic neuropathy. Eur J Hum Genet. 2012 Jun 6. doi:10.1038/ejhg.2012.112. [Epub ahead of print] PubMed PMID: 22669418.

Doyle SL, Campbell M, Ozaki E, Salomon RG, Mori A, Kenna P.F, Farrar G.J, Kiang A.S, Humphries M.M, Lavelle E.C, O'Neill L.A, Hollyfield J.G, Humphries P (2012) NLRP3 has aprotective role in agerelated macular degeneration through the induction of IL-18 by drusen components. Nat Med. 2012 May;18(5):791-8. PubMed PMID: 22484808.

Germano S, Kennedy S, Rani S, Gleeson G, Clynes M, Doolan P, McDonnell S, Hughes L, Crown J, O'Driscoll L, (2012) MAGE-D4B is a novel marker of poor prognosis and potential therapeutic target involved in breast cancer tumorigenesis, Int J Cancer, May 1;130(9):1991-2002, doi: 10.1002/ijc.26200.

Gleeson M, O'Marcaigh A, Cotter M, Brosnahan D, Vulliamy T, Smith OP (2012) **Retinal vasculopathy in autosomal dominant dyskeratosis congenita due to TINF2 mutation**, British Journal of Haematology 2012

Hanrahan F, Campbell M, Nguyen A.T, Suzuki M, Kiang A.S, Tam L.C, Gobbo O.L, Dhubhghaill S.N, Humphries M.M, Kenna P.F, Humphries P. (2012) **On further development of barrier modulation as a technique for systemic ocular drug delivery.** Adv Exp Med Biol. 2012;723:155-9. Review. PubMed PMID: 22183328.

Humphries MM, Kenna PF, Campbell M, Tam LC, Nguyen AT, Farrar GJ, Botto M, Kiang AS, Humphries P (2012) **C1q enhances cone photoreceptor survival in a mouse model of autosomal recessive retinitis pigmentosa.** Eur J Hum Genet. Jan;20(1):64-8. doi: 10.1038/ejhg.2011.151. Epub 2011 Aug 24. PubMed PMID: 21863053; PubMed Central PMCID: PMC3234518.

Kumar D, O'Hare B, Timon, C, Mukhtar M, Kelly D (2012) **Bilateral pneumothoraces and pulmonary oedema following tracheonstomy induced by acute tracheal obstruction,** BMJ Case Reports 2012; 10.1136/bcr-2012-006557,

Linge A, Kennedy S, O'Flynn D, Beatty S, Moriarty P, Henry M, Clynes M, Larkin A, Meleady P, (2012) Differential expression of fourteen proteins between uveal melanoma from patients who subsequently developed distant metastases versus those who did not, Invest Ophthalmol Vis Sci, 9;53(8):4634-43



McAnena L, Murphy C.C, O'Connor J (2012) Tarantula keratitis: a case report. Ir J Med Science Dec 5 2012.

Nguyen A.T, Campbell M, Kenna P.F, Kiang A.S, Tam L, Humphries M.M, Humphries P (2012) Calpain and photoreceptor apoptosis. Adv Exp Med Biol. 2012;723:547-52. Review.PubMed PMID: 22183376

Palfi A, Chadderton N, McKee A.G, Blanco Fernandez A, Humphries P, Kenna P.F, Farrar G.J. (2012) Efficacy of codelivery of dual AAV2/5 vectors in the murine retina and hippocampus. Hum Gene Ther. 2012 Aug; 23(8):847-58. doi: 10.1089/hum.2011.142. Epub 2012 Jul 20. PubMed PMID: 22545762.

Tam LC, Kiang AS, Campbell M, Keaney J, Farrar G.J, Humphries M.M, Kenna P.F, Humphries P (2012) **Protein misfolding and potential therapeutic treatments in inherited retinopathies**. Adv Exp Med Biol. 2012;723:567-72. Review. PubMed PMID: 22183379.

#### Presentations

Chan T, De Chaumont C, Jefferies C.J, Ni Gabhann J, Murphy C.C (2012) **Investigating the impact of HSK on TLR and IFN signaling in peripheral blood mononuclear cells,** RAMI Winter Meeting 2012.

Chan T, De Chaumont C, Jefferies C.J, Ni Gabhann J, Murphy C.C (2012) **Investigating the impact of HSK on TLR and IFN signaling in peripheral blood mononuclear cells,** RCSI Annual Research Day 2012.

De Chaumont.C, Tyson C, Shahnazaryan D, Malone C, NiGabhann J, Jefferies C.J1, Murphy C.C (2012) Investigating the effect of corneal Herpes Simplex Virus-1 infection on Toll-Like Receptor expression in human peripheral blood mononuclear cells, RCSI Annual Research Day, 2012.

Haroon M, Ramasamy P, O'Rourke M, Murphy C.C, FitzGerald O (2012) **Uveitis and Spondyloarthroptahy**, SpondyloArthritis Knowledge and Learning (SpArKLe) meeting 18th October 2012. [Education in Spondyloarthritis designed for the Rheumatology community in Ireland].

Haroon M, Ramasamy P, O'Rourke M, Murphy C.C, FitzGerald O, (2012) **Uveitis and SpA**. SpondyloArthritis Knowledge and Learning (SpArKLe) meeting 28th September 2012 [Education in SpA designed for Allied health rheumatology professionals].

Kennedy, S (2012) **Proteomics of Uveal Melanoma,** International Association of Pathology Capetown, South Africa, September 2012

Kennedy, S (2012) Cilary body teratoid medulloepithelioma occurring in an adult, International Academy of Pathology, Capetown, South Africa, September 2012

Kennedy, S (2012) Keratoconus treated by microwave therapy, British Academy of Ophthalmic Pathology, 29-30th March 2012, Sheffield, UK

Malone C, Shahnazaryan D, Conlon R, Anyiam A, Smith S, Ní Gabhann J, Murphy C.C, Jefferies, C.J (2012) **Targeting Toll-Like Receptor Pathways in Herpes Simplex Keratitis**, Irish College of Ophthalmologists Annual Conference, 2012.

Molloy D, Connolly M, McCormick, Haroon M, Veale D.J, Murphy C.C, Molloy E.E, Fearon U (2012) Acute serum amyloid A and TLR2 acitvation induces pro-inflamatory mechanisms in a novel ex-vivo temporal artery explants culture model of giant cell arteritis. European League Against Rheumatism (EULAR) 2012.

Molloy D, Connolly M, McCormick J, Haroon M, Veale D, Murphy C.C, Fearon U, Molloy E (2012) Blood Vessel Instability and Oxidative Damage in Giant Cell Arteritis. American College of Rheumatology 2012.

O'Rourke M, Connolly M, Fearon U, Murphy C.C (2012) Molecular mechanisms in the pathogenesis of acute anterior uveitis, Rheumatology-Ophthalmology meeting, RVEEH, Oct 2012.

O'Rourke M, Haroon M, FitzGerald O, Murphy C.C (2012) Acute anterior uveitis: an opportunity for earlier diagnosis in spondyloarthropathy. M RAMI Winter meeting, Dublin. December 2012.

O'Rourke M, O'Connor J, Kennedy S, Murphy C.C (2012) A description of the pathological features of corneal specimens of patients requiring tectonic penetrating keratoplasty for rheumatoid melt, Irish College of Ophthalmologists Annual Conference 2012.

O'Rourke M, O'Connor J, Murphy C.C, Kennedy S (2012) **Pathological findings in rheumatoid corneal melt**. EU Cornea, Milan 2012.

O'Sullivan, F (2012) PDA/IMB ESOF 2012 Satellite Conference. Making Gene & Cell Therapy Medicines a Reality. Repairing the Window of The Eye. July 10-11, 2012 Dublin, Ireland.

**O'Sullivan**, F (2012) The 2012 Ireland-Taiwan Symposium on Cancer and Stem Cell Biology. Corneal stem cells for transplant therapy. September 20th, 2012 Dublin, Ireland.

Ramasamy P, HenrM, Linge A, Horgan N, Murphy C.C, Clynes M, Kennedy S, Meleady P, (2012) **Proteomics** of Uveal Melanoma, Biotechnology in Action, National Institute for Cellular Biotechnology, 2012.

Ramasamy P, HenrM, Linge A, Horgan N, Murphy C.C, Clynes M, Kennedy S, Meleady P, (2012) **Proteomics** of Uveal Melanoma, European Vision and Eye Research (EVER) Conference 2012.

Ramasamy P, Henr M, Linge A, Horgan N, Murphy C.C, Clynes M, Kennedy S, Meleady P, (2012) **Proteomics of Uveal Melanoma**, Irish College of Ophthalmologists Annual Conference 2012 (Winner best presentation award).

Shahnazaryan D, Jefferies CJ, Murphy CC (2012) **The role of viral regulatory protein ICPO in herpes simplex virus type 1 (HSV-1) keratitis**, Irish College of Ophthalmologists Annual Conference, Dublin, Ireland 2012.

Treacy M.P, Treacy M.G, Dimitrov B.D, Seager F.E, Stamp M.A, Murphy C.C (2012) A method for prescription of inexpensive spectacles by non-specialist healthcare workers: S-Glasses. Irish College of Ophthalmologists Annual Conference 2012.

# 2012

# Submitted for publication

Power W, (2012) Gene expression profile of cultured limbal-cornea epithelial stem cells and cultured limbal fibroblast cells. Target journal - Molecular Vision

Power W, (2012) The role of cell culture set up in the growth of cultured limbal-cornea epithelial stem cells. Target journal - Experimental Eye Research

#### Awards

Mr Pathma Ramasamy, Clinical Research Fellow RVEEH (Prof C Murphy, Mr N Horgan, Dr M Clynes, Dr P Meleady, Prof S Kennedy)

1. Best paper presentation, Irish College of Ophthalmologists Annual Conference 2012

2. Travel grant for best paper, oncology section, European Association for Vision and Eye Research Congress 2012

Dr Muhammad Haroon, Clinical Research Fellow SVUH (Dr O Fitzgerald in collaboration with Prof C Murphy)

1. Awarded the Innovation Prize for the best abstract at the British Society of Rheumatology for his work on uveitis associated with spondylarthropathy.

# Invited Lectures

(1) Dr Susan Kennedy Ophthalmic pathology for Post-Graduate Medical Trainees in Pathology 4/12/12 RCPI, Kildare Street

(2) Dr Susan Kennedy Ophthalmic pathology for Post-Graduate Trainees in Ophthalmology 20/12/12 at the Research Centre, RVEEH

#### Book Chapters

Brosnahan D, Endophthalmitis, Chapter 14 Pediatric Ophthalmology and Strabismus. Eds Creig S Hoyt and David Taylor. Elsevier 2012

O'Rourke M, Haroon M, Murphy C.C, Fitzgerald O (2012) Anterior uveitis in sponyloarthropathy: The ophthalmologist's role in earlier detection. Ankylosing Spondylitis: Symptoms, Treatment and Potential Complications [Accepted for publication Dec 2012: Nova]



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- 3. Your company could help sponsor a project or a piece of equipment at our Research Unit.

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