PC Project has a new leader!

We are delighted to announce to all the members of the International Pachyonychia Congenita Consortium (IPCC), that PC Project has recruited a new leader who will be familiar to many of you. In February 2017, Irwin McLean, who discovered the first genetic mutations that cause PC in the 1990s, was appointed by PC Project’s Board of Trustees as Chief Executive Officer (CEO) and Chief Scientific Officer (CSO). His appointment is very much supported by PC Project’s founder – the truly amazing Mary Schwartz, who has been tremendously generous and is much loved by the PC community across the globe. The appointment followed the departure of the previous CEO Cindy Atha, in January, who moves on to other things. We would like to take this opportunity to thank Cindy for all of her hard work and efforts while she was CEO and wish her health, happiness and every success in all that the future brings.

Irwin McLean PhD DSc FRS FRSE FMedSci MAE
Professor of Genetic Medicine, University of Dundee
Principal Investigator, Division of Biological Chemistry and Drug Discovery, School of Life Sciences
Scientific Director, Centre for Dermatology and Genetic Medicine
Honorary Consultant Clinical Scientist in Dermatology and Genetics, UK National Health Service
CEO/CSO, PC Project

Fighting for a cure. Connecting & helping patients. Empowering research.
**BIOGRAPHY OF THE NEW CEO/CSO**

Although he has a very Scottish name and works in Dundee, Scotland, Irwin McLean is originally from Northern Ireland and trained at The Queen’s University of Belfast where he graduated in Microbiology in 1985. He gained his PhD in Human Genetics from Queen’s in 1988, based on his studies of protein abnormalities involved in muscular dystrophies. Following a period of postdoctoral work 1988-1991 in the Department of Medical Genetics, also at Queen’s in Belfast, he learned a range of DNA technologies and started to apply them to identify the causes of inherited skin diseases, which were completely unknown at that time. He moved to the University of Dundee in 1992 and there, working as a postdoc in Birgit Lane’s group, he made a number of key discoveries in the field of hereditary skin conditions. This work included the identification in late 1994 of the first mutations in the genes encoding keratins K16 and K17 as the cause of PC (McLean et al., *Nature Genetics* 9:273-278, 1995). This discovery also predicted that mutations in K6 would likely also cause PC. Indeed, this was proved when the first mutations in K6a were discovered by Paul Bowden at Cardiff University in Wales in 1995. Later, Irwin and his long-time co-worker and friend, Frances Smith, and their research group, discovered that mutations in K6b and K6c also cause PC, thus completing the genetic basis of this genodermatosis.

In 1996, Irwin set up an independent research laboratory in Jouni Uitto’s department at Thomas Jefferson University, Philadelphia. Based on his success there, in 1998 he was awarded a Wellcome Trust Senior Research Fellowship and relocated his group to the University of Dundee where he was based at its Medical School for ten years. Since 2008, his laboratories have been based in the School of Life Sciences, which is the top-rated university research unit for biomedical sciences in the UK government’s recent national research quality assessment (Research Excellence Framework).

In 2013, Irwin was elected as a Fellow of the Royal Society (FRS), which is the UK’s national academy of science and the world’s oldest organisation of this type. In 2015, he was also awarded the Royal Society’s Buchanan Medal “for his major contribution to our understanding of the genetic basis of heritable skin diseases” which is only awarded to a single individual once every two years. Irwin has published more than 300 articles in the scientific and medical literature to date, including many in the top genetics and dermatology journals, reporting his lab’s many pioneering discoveries in the field of inherited skin diseases. He is also an elected Fellow of the Academy of Medical Sciences (FMedSci), the Royal Society of Edinburgh (FRSE) and in 2016, he was elected as Member of Academia Europaea (MAE), which is a Europe-wide academy of science.

Irwin has been very heavily involved in PC Project from its outset, and has been on the Medical and Scientific Advisory Board (MSAB) from its inception in 2004, as well as the Steering Group that has guided the organization’s research priorities in recent years. Because he discovered the genes for PC, Irwin has a very strong personal commitment to supporting PC research and PC families. In particular, he is committed to the development of therapies for PC, and has been involved in this undertaking for over a decade.

Prior to the existence of PC Project, Irwin’s laboratory was one of very few working on the disease and functioned as a *de facto* international diagnostic lab for PC. This effort was led by Frances Smith and since the launch of PC project in 2004, she has continued to do all the genetic diagnosis worldwide for PC as well as any other patients who present to PC Project with closely related conditions. This work is critically important for the International PC Research Registry (IPCR), which we need in order to study the natural history of PC and to allow future clinical trials of new medicines to help treat or even cure PC. This activity has also contributed to the discovery of additional keratoderma genes.

Since 2004, Irwin and his group have been running the European PC Patient Support Meetings (PSMs) in Scotland, France and other locations in Europe and he has also attended a number of the North American PSMs and one in Asia. This extensive involvement in the meetings has led to especially close patient contact. In addition, Irwin runs patient meetings for the skin blistering disease epidermolysis bullosa (EBS; caused by mutations in K5 or K14) as well as other genodermatoses closely related to PC. He is always in
search of ways to increase awareness of rare, neglected skin diseases like PC and others across the medical and scientific communities. Considering these disorders as a group helps him to leverage funding for keratin diseases such as PC, EBS and many other rare diseases involving mutations in any of the 54 human keratin genes.

Since he arrived back in Dundee in 1998, Irwin has generated research funding in excess of $50 million. The ability to fund-raise is particularly important for PC Project’s long-term future and is especially important as we move into clinical trials of new medicines for PC that are in development. Irwin has always been extraordinarily generous with his time, and therefore, it will come as no surprise that he is taking no salary for his post as CEO/CSO of PC Project. He has also agreed not to accept any research funding from PC Project at any time in the future, so that he has no conflict of interest. Because of Irwin’s international reputation in the dermatology field, the University of Dundee has very generously allowed Irwin to work for PC Project from within his academic post and also to allow PC Project’s headquarters to be run from Irwin’s offices at no additional cost. Irwin has completed and passed a very detailed conflict of interest assessment within the University. In addition to his voluntary agreement to waive a salary and future research funding from PC Project, he has also stated publically that he will never block access to PC Project’s patient registry by any academic or commercial entity who has a credible research project or experimental therapy that has passed peer review by the Medical and Scientific Advisory board. For example, he is already supporting Palvella Therapeutics who are gearing up for a clinical trial in PC towards the end of 2017 or early 2018.

Irwin McLean’s research group is now pushing hard to develop therapy for PC and related skin conditions. In 2013, Robyn Hickerson moved from Transderm Inc to Dundee, where she now has her own independent research position and is spearheading the efforts of the McLean/Hickerson labs to get new medicines into patients with PC and closely related conditions. Robyn and Irwin have recently been awarded almost $3 million in new research funding to move their work rapidly towards the clinic. We look forward to sharing more information about this research in future newsletters.

Irwin is already settling into his new post of CEO/CSO of PC Project and starting to make some changes to the organization to strengthen it and ensure its success. Holly Evans will take on the role of Patient Support Officer (for the Americas) and will remain in Salt Lake City. Holly has 10 years of experience in the day-to-day running of PC Project and should be familiar to all the readers of this Newsletter. The PC Project office will have a new address and telephone numbers from March 1st 2017 and Holly will remain as the main point of contact for PC Project. The headquarters of the organization will be based at Dundee and Irwin will be supported by his Personal Assistant, Margaret Barton, who is also Patient Support Officer (for Europe). Frances Smith has taken on the role of Chief Geneticist and will be running the genetic diagnostic program for PC Project, which is critically important to allow us to continue building our patient registry and to enrol patients in clinical trials. In addition, Irwin and Margaret have already raised considerable new funding to underpin PC Project’s activities around the world.

We will keep you posted of any further exciting developments as they emerge. Having a motivated scientist leading the organization, who has a close personal interest in PC and in helping patients, is a very positive move for PC Project. Irwin is in the process of shutting down several other research programs in his laboratory and is giving up other directorships so that he can focus more of his time on PC Project and developing therapy for keratin diseases. Watch this space – exciting things are happening!
The IPCC symposium will include the following sessions and speakers (listed alphabetically by session):

**Session 1 Horizons in PC Research**
MEETING CHAIR—Irwin McLean (University of Dundee, Dundee)
KEYNOTE SPEAKER 1—Edel O’Toole (Queen Mary University of London, London)
KEYNOTE SPEAKER 2—Eli Sprecher (Tel-Aviv Sourasky Medical Center, Tel-Aviv)

**Session 2 Translational horizons**
Pierre Coulombe (Johns Hopkins Bloomberg School of Public Health, Baltimore)
Wes Kaupinen (Palvella Therapeutics, Philadelphia)
Bishr Omary (University of Michigan, Ann Arbor)
Dennis Roop (University of Colorado, Denver)

**Session 3 Scanning the PC pain and itch horizons**
SPECIAL GUEST LECTURE—Howard Chang (Stanford University, Stanford)
Thomas Magin (University of Leipzig, Leipzig)
Michael Polydefkis (Johns Hopkins Medicine, Baltimore)

**Session 4 Towards new horizons – brainstorming session**
We hope you will join us for a wonderful meeting!

**Recent Publications**

Author’s Reply: *Pachyonychia Congenita Type 1: Case Report and Review of the Literature.*  

A novel KRT6A mutation in a case of *pachyonychia congenita* from India.  

Mutations in desmoglein-1 cause diverse inherited palmoplantar keratoderma phenotypes: Implications for genetic screening.  

Isolated recessive nail dysplasia caused by FZD6 mutations: report of three families and review of the literature.  