

Centre of Research Excellence for the Study of Naevi

Final Report

Funded by the National Health and Medical Research Council



Acknowledgements

The Centre of Research Excellence for the Study of Naevi Investigator team would like to acknowledge the support from the National Health and Medical Research Council (APP1099021) and the support from The University of Queensland's Faculty of Medicine, which provided additional funding across the life of the Centre.

Acknowledgement of Country

We acknowledge the Traditional Owners and their custodianship of the lands on which our University stands. We pay our respects to their Ancestors and descendants, who continue cultural and spiritual connections to Country. We recognise their valuable contributions to Australian and global society.

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Introduction

The Centre of Research Excellence (CRE) for the Study of Naevi was established in November 2015 with funding from the National Health and Medical Research Council (NHMRC) (APP1099021) and concluded in May 2021. The CRE was a research collaboration between The University of Queensland, QIMR Berghofer Medical Research Institute, Cancer Council Queensland and The University of Sydney.

Naevi, commonly referred to as ‘moles’, are among the most common benign pathologies in humans and are of utmost relevance for the diagnosis of melanoma. By studying naevi, the CRE aimed to generate new knowledge on the naevus life cycle, and correlate phenotypic characteristics with germline genotypic findings to identify genetic markers – to ultimately contribute to personalised medicine via risk stratified naevi prevention and surveillance programs. In addition, the CRE aimed to facilitate collaboration, and develop the health and medical research workforce in clinical dermatology, teledermatology, epidemiology, genetics, molecular biology, behavioural science, and bioinformatics.

This report offers a snapshot of activities conducted by the researchers associated with the CRE and key outputs generated during the five years of funding (2015-2020). A large number of projects were undertaken during this time, and this report captures a selection of highlights. Overall, the CRE launched a body of work that has started to reframe the early detection of melanoma in Australia.

Professor H. Peter Soyer, CRE for the Study of Naevi Lead Investigator



The Team

Chief Investigators

Professor H. Peter Soyer

Professor Soyer is Inaugural Chair in Dermatology at UQ, Director of the Dermatology Research Centre, The University of Queensland Diamantina Institute, at the Translational Research Institute, and Director of the Dermatology Department, Princess Alexandra Hospital. He is a world leader in the field of dermatology with particular expertise in preventative dermatooncology and dermatologic imaging.



Associate Professor Rick Sturm

Associate Professor Sturm is a Principal Research Fellow of the Dermatology Research Centre, The University of Queensland Diamantina Institute.



Professor Adele Green

Professor Green is a Senior Scientist and Head of the Cancer & Population Studies Group at QIMR Berghofer Medical Research Institute. Her research is exemplary in being an authoritative source of advice for health professionals regarding skin cancer prevention (US NIH, and the BMJ's Clinical Evidence series since 2002).



Associate Professor David Duffy

Associate Professor Duffy is a Senior Research Fellow in the Genetic Epidemiology Unit at the QIMR-Berghofer Medical Research Institute.



Professor Monika Janda

Professor Monika Janda is the Professor in Behavioural Science at the Centre for Health Services Research, University of Queensland, Faculty of Medicine. She is a health psychologist with a research background in cancer prevention and quality of life research. Professor Janda is the Scientific Co-ordinator for the CRE.



Professor Joanne Aitken

Professor Aitken is Head of Research in the Viertel Cancer Research Centre, Cancer Council QLD, Director of the QLD Cancer Registry; Director of the Australian Paediatric Cancer Registry; and holds academic positions with several Universities. Prof Aitken led the world's only randomised trial of the effectiveness of a population skin screening program for melanoma involving 70,000 adults in 18 communities.



Professor Tarl Prow

Professor Prow is a research only academic specialized in biotechnology development with a focus on skin imaging/image analysis and microdevices. Formally Deputy Director of the UQDI Dermatology Research Centre, in 2017 Dr Prow started a new role as Research Professor at the Future Industries Institute, The University of South Australia.



Professor Scott Menzies

Professor Menzies is a Professor in the Discipline of Dermatology at The University of Sydney and was Director, The Sydney Melanoma Diagnostic Centre from 2003-2013. He is known as a world leader in the development of technologies for the diagnosis of primary melanoma of the skin.



Associate Professor Helmut Schaidler

Associate Professor Schaidler is a team leader at the Dermatology Research Centre, The University of Queensland Diamantina Institute.



Associate Investigators

- Associate Professor Victoria Atkinson, Medical Oncologist, Princess Alexandra Hospital, Brisbane
- Professor Boris Bastian, Dermatopathologist, University of California San Francisco, San Francisco
- Associate Professor Clara Curiel-Lewandrowski, Dermatologist, University of Arizona, Tucson
- Professor Brian Gabrielli, Mater Research Institute, The University of Queensland, Brisbane
- Professor Allan Halpern, Chief, Dermatology Service, Memorial Sloan Kettering, New York
- Professor Rainer Hofmann-Wellenhof, Professor of Dermatology, Medical University of Graz, Graz
- Associate Professor Nadine Kasparian, Psychologist, University of New South Wales, Sydney
- Professor John Kelly, Dermatologist, The Victorian Melanoma Service, Alfred Hospital, Melbourne
- Associate Professor Lois Loescher, Behavioural Scientist, University of Arizona, Tucson
- Dr Graeme Walker, Carcinogenesis Laboratory, QIMR Berghofer Medical Research Institute, Brisbane

Postdoctoral Research Fellows

- Dr Stephen Ainger
- Dr Brigid Betz-Stablein
- Dr Mitchell Stark
- Dr Aideen McInerney-Leo
- Dr Tatiane Yanes
- Dr Elsemieke Plasmeijer
- Dr Anthony Raphael
- Dr Anna Finnane
- Dr Astrid Rodriguez-Acevedo
- Dr Li Lin

Project Staff

Elizabeth Payne, Melissa Kerr, Dr Uyen Koh, Katie Lee, Joachim Torrano, Dr Saira Sanjida, Darren Smit, Kasturee Jagirdar, Dr Lisa Tom, Elizabeth Peach, Sabrina Hammerlindl, Montana O'Hara, Caitlin Horsham.

Clinical Research Assistants

Dr Antonia Laino, Dr Jenna Rayner, Dr Skye Windsor, Dr Jean-Marie Tan, Dr Matt Hishon, Dr Alison Bullen, Dr Harrison Edwards, Dr Mitchell Robinson, Dr Fleur Kong, Dr Rose Norton, Dr Alastair Ashley, Dr Pietro Bearzi, Dr Frank Chiu, Dr Ramez Barsoum, Dr Tabrez Sheriff, Dr Sarsha Mortimore, Dr Ruby Lee, Dr Hannah Gribbin, Dr Myco Tran, Laura Adams, Kaitlin Nufer, Chantal Rutjes.

Higher Degree Research Students



PhD Completions

1. McMeniman, Erin. Genotypic and phenotypic correlations in a cohort of patients with multiple primary melanoma. [PhD Thesis](#). Australia: The University of Queensland; 2020.

MPhil Completions

1. Rayner, Jenna. Gene polymorphisms associated with amelanotic/hypopigmented and nodular melanoma. [MPhil Thesis](#), The University of Queensland; 2020.
2. Nufer, Kaitlin. Classification of naevus distribution patterns in the Brisbane naevus morphology study. [MPhil Thesis](#), The University of Queensland; 2019.
3. Tan, Jean-Marie. Dermoscopic and molecular correlation of melanocytic naevi. [MPhil Thesis](#), The University of Queensland; 2018.

Ongoing Students

PhD Candidate	Institution	Topic
Clare Primiero	UQ Dermatology Research Centre (DRC)	Psychosocial and behavioural effects of reporting personalised genetic results regarding cancer susceptibility to people at high risk of melanoma
Dilki Jayasinghe	UQDRC Centre for Health Services Research	Statistical modelling of the spatial patterns and body site distributions of naevi
Nathasha Naranpanawa	UQ School of Information Technology and Electrical Engineering	Analysis and classification of skin lesions with deep neural networks for automated skin cancer detection
Fatima Al Zegair	UQ School of Information Technology and Electrical Engineering	Naevi classification using machine learning
MPhil Candidate	Institution	Topic
Dr Antonia Laino	UQDRC	Iris freckles and iris naevi as markers of cutaneous melanoma risk

Masters by Coursework Student Research Projects: Roshni Mendis (Epidemiology); Stacey Llewellyn (Biostatistics); Bin (Anna) Lvu (Data Science); Katarzyna Grochulska (Dermatology)

Medical Student Research Placements: Sam Kahler, Harpal Dhillon

Summary of Achievements

The CRE was established in 2015. Nine Chief Investigators from four research organisations aspired to improve melanoma risk prediction and early detection methods, with the ultimate aim of improving melanoma early detection and survival. The CRE took an innovative approach to tackle the early diagnosis of melanoma by focusing on naevi. During the five-year project, the CRE made some amazing progress. For example:

- The CRE established automated mole counting algorithms – we now know that people in Queensland have on average 50, and up to 332 moles on their body.
- CRE researchers found a genetic signature of moles – we now know that many moles carry very similar mutations as melanoma.
- We discovered the contribution that certain genes and certain phenotypes, compared to the combination of both, make for melanoma development.
- The CRE found that some albinism-related genes are more common in patients with amelanotic/hypomelanotic melanoma. These are melanomas that lack the dark pigmented colour.

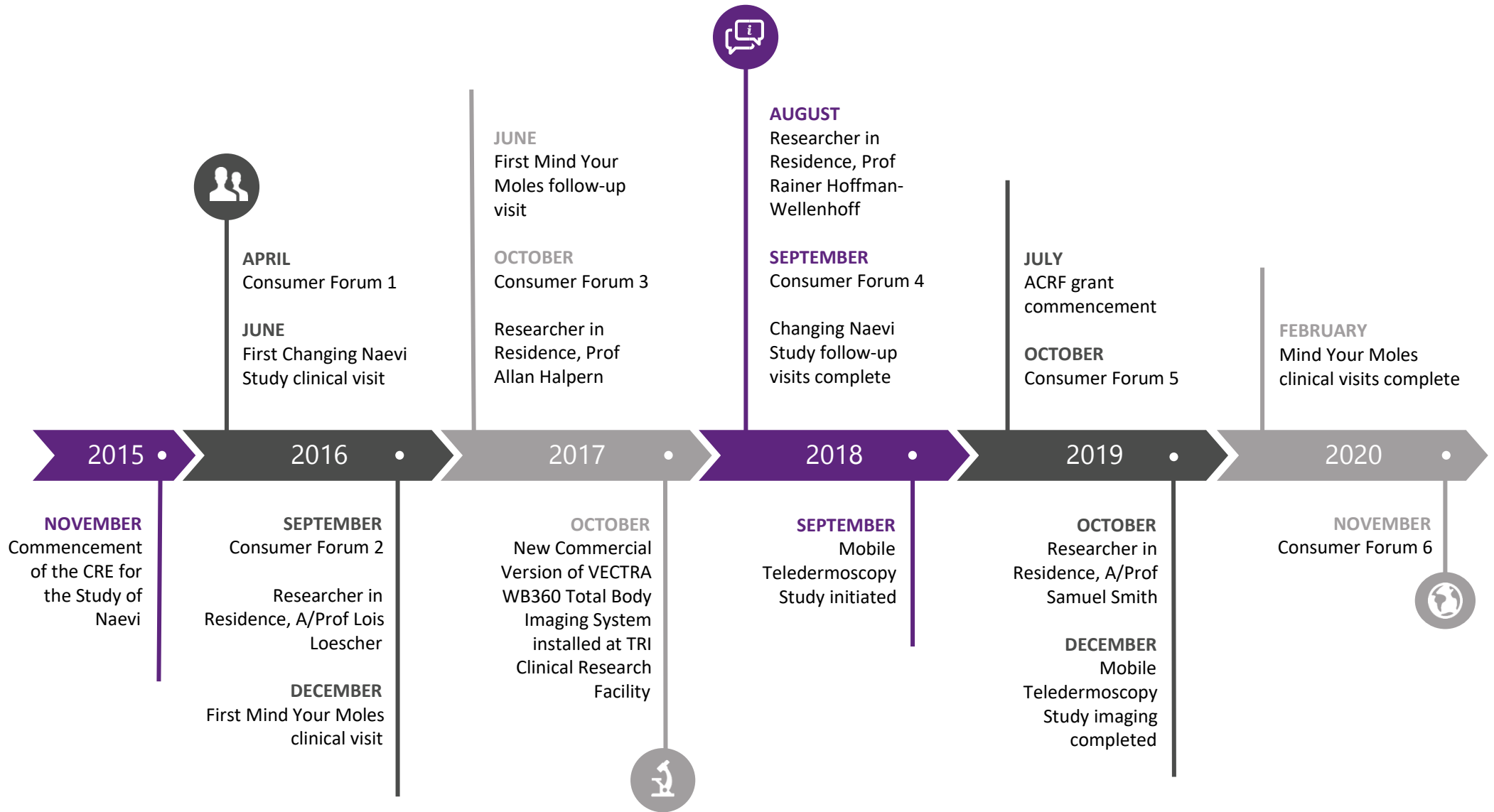
To support their research, the CRE team sourced an additional 31.2 million in funding from organisations including the Medical Research Future Fund, Princess Alexandra Hospital Research Foundation, and the Harry J Lloyd Charitable Trust.

Another major goal of the CRE was to inspire young clinicians and scientists to use the latest technology advancements in skin cancer diagnostics and care. The CRE recruited nine Postdoctoral Researchers, and provided clinical research experience to 19 medical doctors interested in dermatology. In addition, eight Higher Degree Research students were actively supported by the CRE.

Over the life of the CRE, the team produced 69 publications and made 83 presentations, including 15 at conferences and 29 at world congresses. In addition to journal publications, the CRE disseminated knowledge about its work through its website, social media, consumer forums and scientific meetings.



Timeline



What's next for the CRE?

The CRE for the Study of Naevi was one of three complementary NHMRC-funded Centres of Research Excellence, alongside the CRE in Melanoma led by CI Mann (University of Sydney); and the CRE in Telehealth led by CI Gray (UQ), which contributed to the research and clinical strength underpinning the Australian Cancer Research Foundation (ACRF) Australian Centre of Excellence in Melanoma Imaging and Diagnosis (ACEMID) initiative.



ACEMID

The ACRF ACEMID project is a three-year prospective cohort study which aims to develop more efficient and effective screening for the early detection of melanoma through the provision of an innovative 3D total body photography system. The ACRF ACEMID will deliver a next-generation, precision strategy of skin imaging technology integrated within a telemedicine network spanning 15 dermatology research nodes across Australia to reconceive how melanoma is screened for and detected.

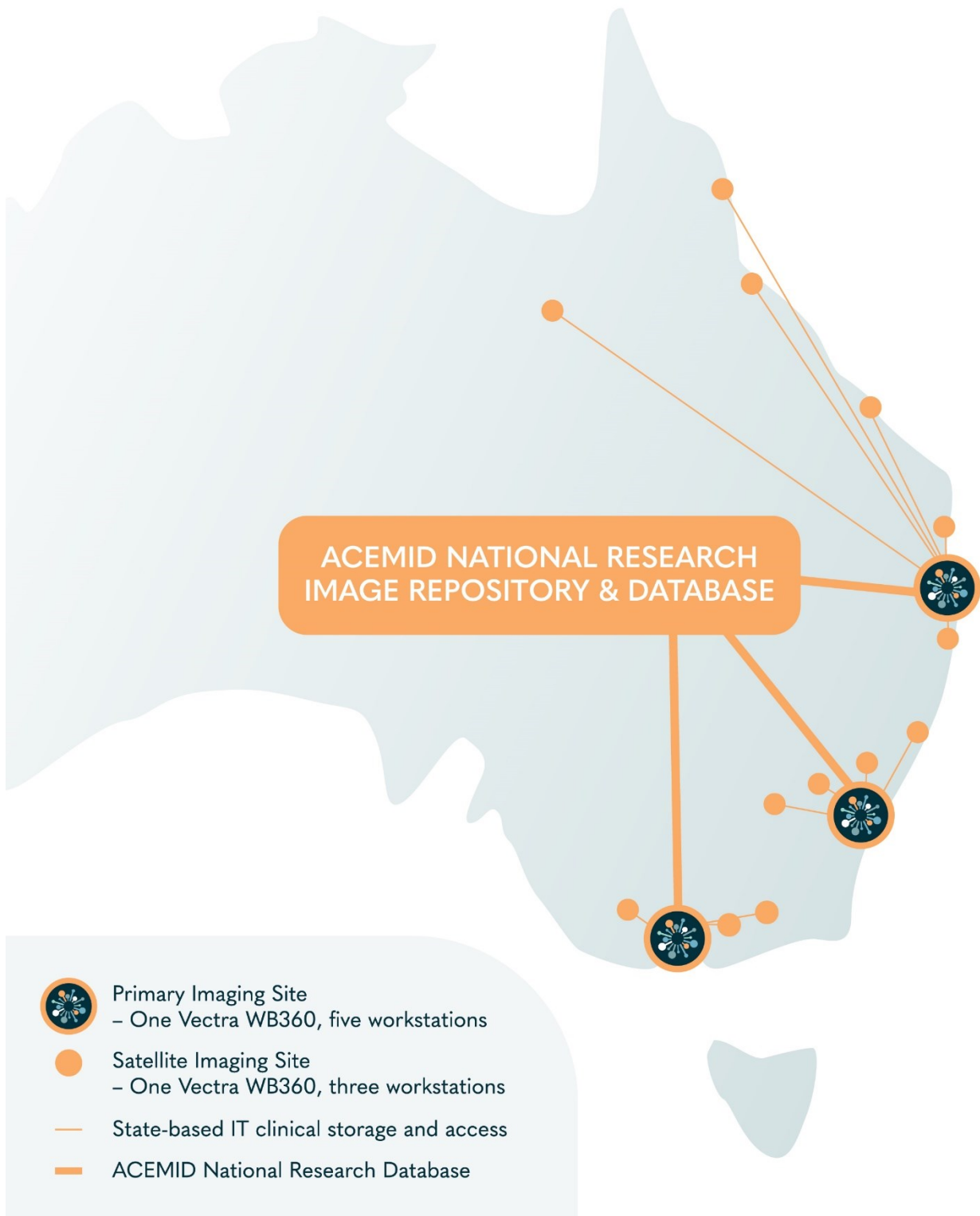
Aims of the ACEMID project:

- Establish a network of state-of-the-art 3D total body imaging systems and informatics infrastructure to form a multidisciplinary and multi-site centre of excellence; the first of its kind internationally.
- Integrate and leverage world-class research expertise that is unique to Australia to provide technologically disruptive and reliable solutions for the early diagnosis of melanoma, particularly for people at high and ultra-high risk and spanning urban and regional/rural areas.
- Provide more equitable access to melanoma diagnosis in regional Australia using telehealth supported by artificial intelligence.
- Champion a reduction in the overarching burden, morbidity and mortality associated with the 14,000 melanomas occurring yearly in Australia by helping ensure that healthcare services are targeted effectively and equitably to Australians most in need.
- Channel people into risk stratified screening or surveillance programs to enable significant personal and health care system cost-savings.
- Develop the world's largest research repository of 3D total body skin images to further improve the diagnosis of melanoma.



ACEMID

AUSTRALIAN CENTRE OF EXCELLENCE IN
Melanoma Imaging & Diagnosis



Consumer Engagement

The CRE for the Study of Naevi engaged with consumers through consumer forums, held annually at the Translational Research Centre at the Princess Alexandra Hospital or the Cancer Council Queensland in Brisbane, Australia. Consumer forums were held in conjunction with the Cancer Council Queensland and Melanoma Patients Australia, allowing the CRE researchers to gather information to inform their research.

Each consumer forum featured an invited guest speaker who gave a presentation followed by a question-and-answer session where consumers were able to ask questions to a panel. Each panel included the invited guest speaker, as well as members of the CRE research team. Consumers were highly engaged in the discussion session each year and asked a range of questions related to the topic of presentation as well as dermatology in general. In addition to the consumer forums, consumers part of the CRE Core Study, Mind Your Moles, also received 6-monthly newsletters providing updates on participant enrolment and new study findings.

Year	Title	Guest Speaker
2016	Skin Awareness in the Community	Forum 1 held in conjunction with CRE Launch
2016	Skin Checks	Associate Professor Lois Loescher <i>University of Arizona</i>
2017	Algorithms and robots, will they play a role in melanoma early detection?	Prof Allan Halpern <i>Memorial Sloan Kettering Hospital</i>
2018	Melanoma Genomics	Dr Aideen McInerney-Leo Dr Mitchell Stark Associate Professor Erin McMeniman <i>University of Queensland</i>
2019	Cancer prevention: What has psychology got to do with it?	Associate Professor Samuel Smith <i>University of Leeds</i>
2020	Artificial intelligence in dermatology: How, where, and when?*	Dr Veronica Rotemberg <i>Memorial Sloan Kettering Cancer Center</i>

* Held as online community webinar due to COVID-19



Research Programs

CRE Core Study – Mind Your Moles

Background

Melanoma is the deadliest type of skin cancer and the fourth most common invasive cancer in Australian men and women. Melanocytic naevi or 'moles' are common benign neoplasms of the skin which vary in colour, number, shape and size, both from person to person and within individuals. The presence of many melanocytic naevi, or 'moles', is the strongest predictor of melanoma risk. Therefore, improved understanding of how melanomas develop and change over time is essential to better understand melanoma development.

Mind Your Moles was the core study of the CRE Naevi. The 3-year prospective, population-based cohort study aimed to improve our understanding of the epidemiology and natural history of melanocytic naevi in adults. The project enrolled adults living in South-East Queensland, Australia, with participants randomly selected from the Australian Electoral Roll.

Clinical Study Visits

Participants in Mind Your Moles visited the Translational Research Institute Clinical Research Facility at the Princess Alexandra Hospital once every 6 months for three years to undergo 3D total body photography using the VECTRA machine. Over 1200 skin screening examinations were completed throughout the study, which ran from December 2016 to February 2020. A total of 196 participants were enrolled at baseline and 164 participants remained at the final study visit.

At each study visit, participants underwent total body photography, a clinical examination, and were asked to complete a Sun Behaviour and Health Questionnaire. Saliva samples were also collected at baseline to enable further research on genetic involvement in the development of melanoma.

Referrals

Skin images were reviewed by an experienced dermatologist and any suspicious lesions detected at the clinic visits were referred for excision and histopathology. Throughout the study, over half (55%) of participants received a referral to their own doctor for one or more lesions of concern. Males were more likely than females to receive a referral for a lesion of concern, and the average number of lesions referred per person was 2.3 (range 1-9).

Melanomas Detected

A total of six melanomas were diagnosed throughout the Mind Your Moles study. These occurred across four separate participants; three participants had one melanoma, and one participant had three melanomas. All six were melanoma in-situ, Clark Level 1, and were diagnosed within the first year of the study.



The graphic features a large orange circle at the top left containing the text "mind your moles" in white lowercase letters. Below this is the "naevi" logo in a dark blue font, with "CENTRE OF RESEARCH EXCELLENCE" in smaller orange text underneath. A central text block reads: "Queensland cancer researchers invite you to become involved in this study". Below this is a circular photograph of a woman with blonde hair, resting her chin on her hand. A teal speech bubble overlay on the photo contains a quote: "I was diagnosed with advanced Stage 4 melanoma at 22. I urge all Australians to come forward and join this study that aims to better understand the path from moles to melanoma." followed by "Emma Betts FROM DEAR MELANOMA". At the bottom right of the photo, the text "Study ID: APP1099021" is visible.

Non-Melanoma Skin Cancers Detected

A total of 39 non-melanoma skin cancers were diagnosed by the Dermatology Research Centre throughout the Mind Your Moles study. These occurred across 32 separate participants, and included 36 basal cell carcinomas, and three squamous cell carcinomas.

VECTRA Feedback

Throughout the study, participants were asked to provide feedback on their experience of the VECTRA. Overall, participants were accepting of the new technology and 75% of participants stated they believed the VECTRA was effective or very effective at monitoring changes in their moles.

Nearly all participants (95%) stated they were comfortable participating in 3D total body photography, and 98% said they would consider using the VECTRA if it were to become commercially available with their regular medical practitioner. Use of 3D total body photography for the monitoring of naevi in patients at high risk of melanoma is now being expanded through the ACRF ACEMID Cohort Study.



Program I – Genetics and Epidemiology of Naevi

The total number of naevi a person has is among the most important risk factors for melanoma. CRE Program I was based on the idea that combining clinical and dermoscopic classification of naevi with genetic information would provide new avenues for better early prediction and diagnosis of melanoma and translatable individual risk estimates. CRE Program I combined data on the changing clinical and dermoscopic classification over time, with genetic information collected from saliva and naevi. The program used data from 193 participants in the CRE Core Study (Mind Your Moles), as well as additional data from a large case-control study of naevi (Brisbane Naevus Morphology Study).

The Brisbane Naevus Morphology Study began as a pilot study of total body photography and dermoscopy in 2009, focusing on clarifying naevus morphology and its associations with genetics and melanoma risk. The aim of the study was to examine the relationship between the types of moles a person carries, including number, size, distribution, profile, and colour of melanocytic naevi, freckling score with their genetic risk of development of melanoma. The BNMS imaged and genotyped 1254 participants from south-east Queensland and collected dermoscopic images of over 28,000 naevi. The BNMS has produced 32 peer-reviewed publications, including the findings:

- Genetic associations with naevus dermoscopic subtypes, naevus count, and melanoma risk.
- A comprehensive overview of the genetics of human skin and hair pigmentation.
- Evidence that the two pathways posited in the “two pathways to melanomagenesis” model – a high naevus count/predisposition to melanocytic proliferation pathway and a sun-sensitive pigmentation/chronic UV damage pathway – act synergistically instead of additively.
- Pigmented lesions on the iris are an independent risk factor for cutaneous melanoma.
- Genetic associations of nodular melanomas.
- Evidence that Australian patients with ≥ 3 melanomas, or ≥ 2 or more melanomas diagnosed before the age of 40, should be offered *CDKN2A* testing.

Program II – Consumer Facilitated Naevi Monitoring

CRE Program II, which explored consumer-facilitated naevi monitoring, was a small mobile teledermoscopy study embedded within Mind Your Moles. The study aimed to explore how well consumers were able to monitor and capture images of clinician identified lesions on easy-to-see versus hard-to-see body areas. In addition, the study explored how well consumers accepted the use of technology-aided skin self-examination. Participants were asked to complete sequential imaging of lesions at baseline, 1-month and 3-months, using a dermoscopic device attached to their mobile phone which linked to a mobile app. Data analysis for this project is ongoing. A total of 28 participants were part of the study, none of whom had used mobile teledermoscopy before.

Initial data analysis found 64% of participants said they would use and/or purchase a mobile teledermoscope for use in the future.



Program III – Changing Naevi Study

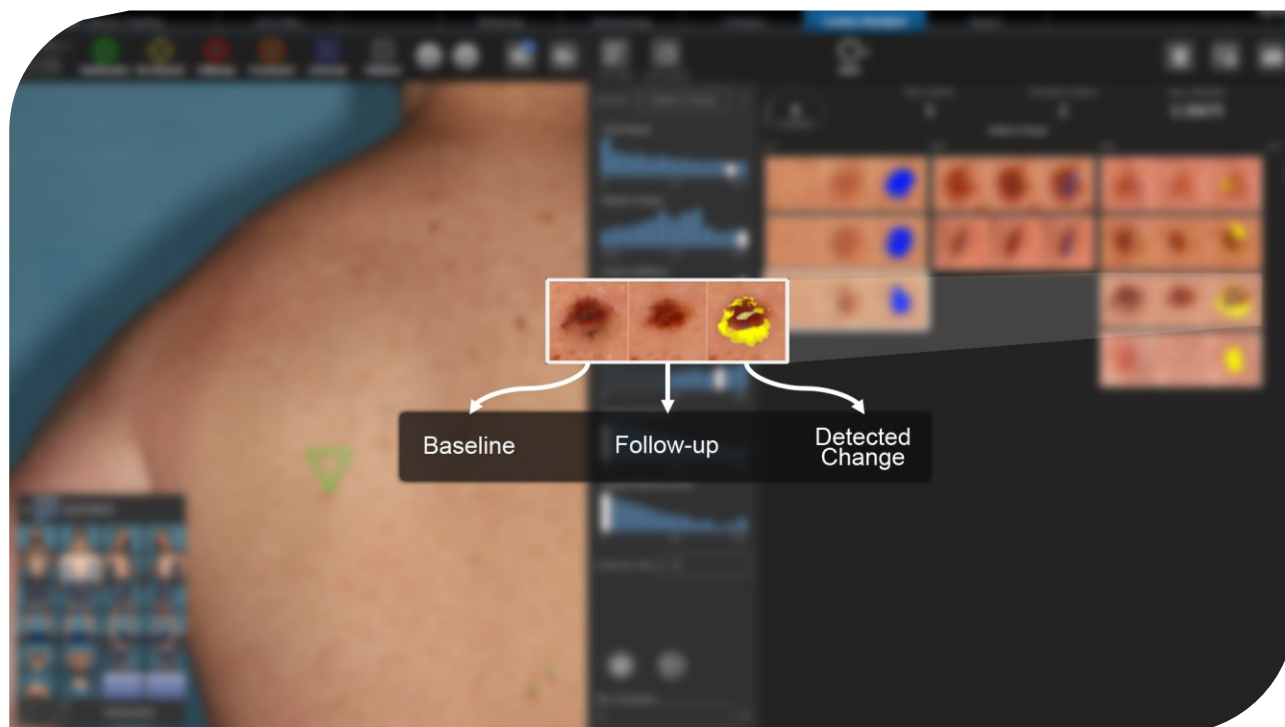
CRE Program III was a prospective cohort study which aimed to explore the evolution of naevi in people with advanced melanoma under the treatment of targeted therapies and immunotherapies, as well as people at high risk for melanoma.

Participants in this study were enrolled between May 2016 to July 2017, and followed up at four, eight, and 12 months for clinical imaging of naevi. The last follow-up visit for CRE Program III was completed in September 2018. Sociodemographic, clinical, environmental, phenotype, and behavioural characteristics data were collected.

A total of 150 participants were enrolled in Program III at baseline, and separated into two groups for analysis. The groups were: i) people with advanced melanoma (n= 43) and ii) people at high risk of melanoma (n= 107).

Forty-one participants with advanced melanoma under treatments were included in the study. Twenty-four participants were alive as of October 2020. The most frequently prescribed treatment for these participants was PD-1 blocker (pembrolizumab), however they were commonly on a combination of targeted and immunotherapies. A total of 28 advanced melanoma participants completed a 12-month follow-up visit and 387 dermoscopic images of naevi were analysed to explore the change over time. Overall, 33% of naevi changed over time, with 29% decreasing in colour or size and 4% of naevi increasing in colour or size.

A total of 107 people at high risk for melanoma were enrolled at baseline, and 64 participants completed the two to three follow-up visits for clinical imaging, with 1600 dermoscopic naevi images included in the study. In this cohort, only 6.5% of naevi changed over time. Participants who were obese, and those with fair facultative skin colour, had the highest median number of changing naevi.



Additional Projects



Mutation Signatures

CRE Program III involved performing molecular analysis of differing dermoscopic patterned naevi to identify molecular signatures that could be identified that may be responsible for phenotypic differences. In brief, a collection of fresh naevus and adjacent skin tissue specimens underwent whole exome sequencing (all genes), global methylation analysis, along with gene expression analysis (all coding and non-coding genes).

From this extensive genomics dataset, researchers identified an increase in mutations in sun-exposed naevi which corresponded with an expressed mutation signature associated with UV-exposure. In minimally sun-exposed naevi, there was an increase in differing types of mutations which corresponded with increased mutation signatures related to defects in DNA repair. DNA copy number was increased in reticular and non-specific naevi which corresponded with increased DNA methylation profiles related to genomic instability. Analysis of gene expression datasets is ongoing.

International Skin Imaging Collaboration (ISIC) Melanoma Classification Challenge

Data collected from the CRE contributed to the 2020 ISIC Melanoma Classification Challenge. The 2020 dataset included over 30,000 images from 2056 patients across three continents. Of these, the University of Queensland Dermatology Research Centre contributed 26,466 images from three prospective longitudinal studies, including the CRE Core Study (Mind your Moles) and Program III. The 2020 dataset was the first dataset to provide contextual images, where more than one lesion image from each patient was included to try to better mimic a clinical assessment. In future, the ISIC 2020 dataset will be used to develop and improve machine learning algorithms for melanoma detection and early diagnosis.

In November 2020 the CRE held a Melanoma Community Forum via webinar. Guest speaker Dr Victoria Rotemberg, an innovative dermatologist from Memorial Sloan Kettering Cancer Center in New York, presented on the topic “AI in dermatology: how, where, and when?” In her talk, Dr Rotemberg provided great insight on the use of artificial intelligence (AI) for diagnostic imaging and shared exciting results from the 2020 ISIC Melanoma Classification Challenge.

As part of her presentation, Dr Rotemberg reflected upon the potential for human-AI collaboration, and discussed some of the remaining challenges for translating AI into clinical dermatology practice. Examples of these challenges include identifying the most appropriate users of AI for skin cancer screening (e.g. patients, general practitioners, dermatologists), as well as the kind of criteria we would need to encourage adoption (e.g. improved sensitivity).

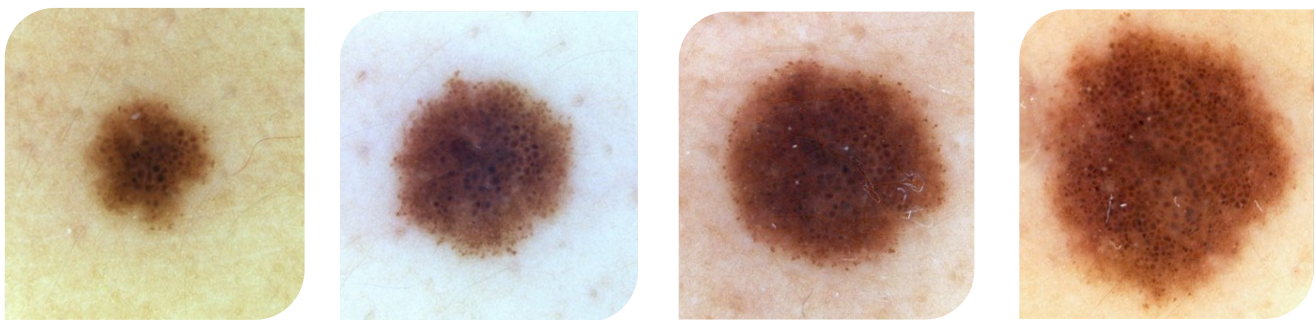
A methods paper describing how images collected in the 2020 ISIC Challenge were used to create a dermatology image dataset has been published in the journal *Scientific Data*. Images in the multicentre dataset includes clinical contextual information. A results paper is currently underway, which examines if the addition of patient- and lesion-related clinical context improves the AIs recognition of melanoma.

Lesion Change Detection

During the CRE, researchers Professor Scott Menzies and Dr Brigid Betz-Stablein initiated work on change detection using data from the Mind your Moles and Health Outcomes Programs study (HOPs). The HOPs study was a nested feasibility study of a targeted screening program using 3D total body photography and included participants at high risk of melanoma.

Change detection involved comparing pairs of dermoscopic lesion images to identify change over time. Two specially trained melanographers assessed 10,000 pairs of dermoscopic images, equating to approximately 250 hours of work. The melanographers were required to agree if change had occurred, as well as record the type of change observed and if the change was clinically significant. This human assessment was used as the “gold standard” to train the AI.

In addition, over 200 participants were imaged twice at a single timepoint, resulting in over 400 3D total body maps and images of approximately 5000 lesions. These identical body maps were used as a negative control to train the AI model. In future, this data is envisioned to form the ground truth to train an AI model to automatically assess lesion change from 3D total body photography images.



Ongoing Data Analysis

Data analysis is currently ongoing for a number of additional publications which will contribute to the body of work produced by the CRE. Topics of future publications using CRE data will include:

- Naevus distribution and sun damage
- Validation of artificial intelligence algorithms in people with skin of colour
- Distribution of naevi on the arms and lower limbs in the general population
- Principal component analysis to detect suspicious naevi
- Naevi in the iris
- Clinical outcomes and histopathological results of the Mind Your Moles study
- Consumer experience of 3D total body imaging (VECTRA feedback)
- Dermoscopic naevus patterns by body site
- VECTRA automated size measurement validation

The CRE in Action

Now is the time for dermatologists to learn genomics



Dermatologists may need to look further than red hair and freckles when identifying patients who might be genetically predisposed to skin cancer.

A University of Queensland review of the key genes affecting pigmentation could be the first step towards incorporating genomics into clinical practice to identify high-risk patients.

Associate Professor Rick Sturm, from UQ's Dermatology Research Centre, said the review was prompted by the need to provide the latest information to clinicians.

"The whole idea of how we will deliver medicine in the future is directed towards personal or precision medicine," Dr Sturm said.

"This will require clinicians to understand their patients' genetic background, so we really needed a primer for the dermatologists on the genes they should know about."

The review gives an overview of 15 genes which affect pigmentation of the skin, eyes and hair or melanoma risk.

"These include the most important genes for pigment which, when they are mutated, have a manifestation of genetic disease," Dr Sturm said.

"Dermatologists might already have some knowledge of the gene associated with red hair and freckling, which is MC1R, but there are many others."

Dr Sturm said there were now three major genes associated with naevi or moles.

"One of these genes – IRF4 – has a big effect on naevi count, and recently it's been suggested the gene can have a significant effect on the type of melanoma a patient may get," he said.

"This gene usually affects a patient's presentation in the clinic – they have lots of moles as children and not so many as adults and they get their melanoma later in life.

"In the future, clinicians will have to know the genes affecting their patients' pigmentation and naevi presentation, and that will affect how they examine or treat them."

Dr Sturm is a molecular geneticist who has been investigating gene expression, pigmentation genetics and the process of melanoma metastasis for 25 years.

"There has been such rapid progress in our knowledge on the genetics underlying skin cancer and pigmentation, but there's been little means of keeping dermatologists updated," Dr Sturm said.

"They are aware of the implications of differences in skin pigmentation for outcomes when it comes to sun-exposure, skin cancer and disease outcomes, but they need to begin to understand the underlying genetics.

"Our review is a stepping stone to incorporating genomics into the clinic to identify the patients that need management and surveillance."

The review is published in the journal [Dermatology](#).

New technology to spot skin cancer sooner

Professor H. Peter Soyer was named in the NHMRC Ten of the Best Awards, in recognition of Dermatology Research, in 2017.

The award recognises research conducted under Professor Soyer's Practitioner Fellowship, as well as the NHMRC Centre for Research Excellence for the Study of Naevi.

The number of skin cancers, including melanomas, continues to rise in Australia. Detecting melanoma early is critical for survival—rates worsen as the thickness of the melanoma increases. Professor H. Peter Soyer is a world leader in dermatology and preventative measures for skin cancers. Professor Soyer and his group at The University of Queensland have been researching and implementing a number of programs using the latest technologies to help detect skin cancers sooner.

'Melanoma is the most common cancer for 15-39 year old Australians—with the highest 'years of life lost' of any cancer.'

Professor Soyer's lifelong vision is to reduce the burden of skin cancer through early detection of melanoma and other skin cancers. One of his unique projects is the Skin Emergency Telemedicine Service (SETS) program. This service uses asynchronous Store-and-Forward Technology to provide rapid and accurate dermatological diagnosis and is critical in Queensland emergency departments.

'This eliminates the days or weeks of waiting to obtain a specialist consultation in rural Queensland,' Professor Soyer said.

Professor Soyer is also involved in several areas of pigmented naevus (moles) and melanoma research.

One development is the use of total body 3D photography and dermoscopy—allowing monitoring of moles at high risk of developing into melanoma.



'Major goals of our research are to understand the genetic basis of the clinical presentation of mole phenotypes and human pigmentation traits (skin, hair and eye colour) to provide better melanoma risk stratification,' Professor Soyer explained.

'Expertise in naevus and melanoma research has led to the NHMRC funded *Centre of Research Excellence for the Study of Naevi*,' he concluded.

Professor Soyer is now coordinating findings from current genomics, diagnostics and consumer-based research projects to identify individuals at high and ultra-high risk. These individuals will then be screened using a combination of 3D total body photography, consumer-driven mobile teledermoscopy, and minimally invasive microbiopsies for suspicious lesions.

'It is the first program of research to test these technologies in the context of the Australian health care system, and will be used to drive evidence-based changes to clinical practice,' he said.

'Based on my lifelong dedication to this field I am confident of making a significant contribution to the ultimate goal—no one should die of melanoma.'

Professor Soyer is also collaborating with the Queensland University of Technology and the University of Arizona, Tucson, USA, in developing mobile melanoma screening—using mobile phones and dermatoscopes—to detect melanoma earlier.

Melanoma is in the eye of the beholder

University of Queensland researchers have found that freckles and moles appearing on the iris indicate a high risk of melanoma, particularly in people under 40 years of age.

[Dermatology Research Centre](#)'s Associate Professor Rick Sturm said the presence of pigmented lesions was an effective predictor of the risk of melanoma that complemented traditional factors.

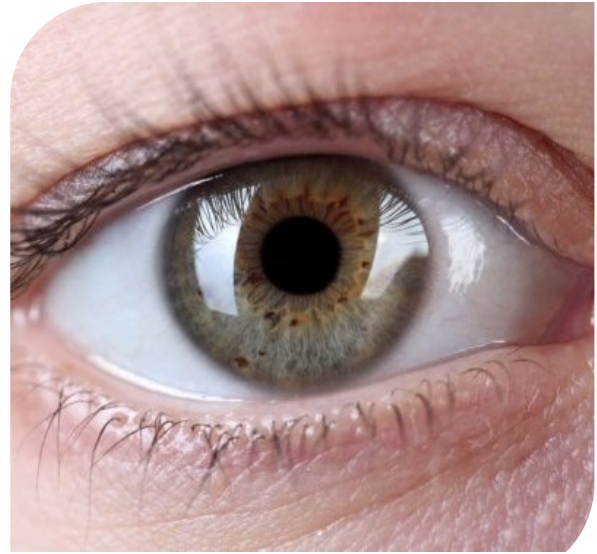
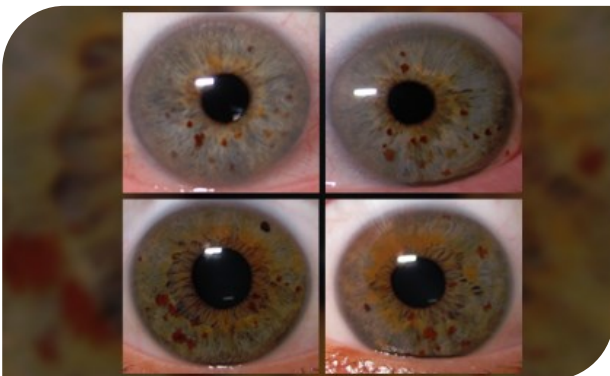
"We found the presence of three or more iris pigmented lesions was associated with a 45 percent increased risk of melanoma," Dr Sturm said.

"This association was particularly strong in people under 40.

"The presence of iris freckling and naevi (moles), provides additional information about an individual's melanoma risk over and above factors like blue eyes, red hair, fair skin and the number of moles on the skin."

The study, involving Professor H. Peter Soyer and Dr Antonia Laino, involved 1117 participants of European background living in South-East Queensland.

Dr Laino said the results showed that participants with pigmented lesions were 1.45 times more likely to develop melanoma.



"This association was particularly strong in people under 40, suggesting a genetic susceptibility," she said.

"It also suggests the potential use of these lesions as a marker for melanoma risk in younger patients (1.8 times more at risk).

"Melanoma is the most common cancer in Australians aged 15 to 39.

"Despite many new advances in treatments, long term prognosis remains poor, therefore early detection is still key in reducing the burden of the disease.

"It's very easy to look for iris pigmented lesions, and we hope that these findings will help doctors identify those people who may be at increased risk of melanoma and need a skin check."

"These lesions should be used as markers for melanoma risk in younger patients."

The UQ [Diamantina Institute](#) study is published in the [British Journal of Dermatology](#).

Sun exposure damages our skin cells DNA – the story of when repair mechanisms breakdown

Researchers from The University of Queensland have conducted the most comprehensive study looking at the genomics of skin and naevi (moles) with the goal improving the early detection of melanoma.

Overall, studies have shown that around 30% of melanomas arise within pre-existing naevi, which leaves 70% of melanomas developing on the normal skin.

[UQ Diamantina Institute's](#) Dr Mitchell Stark is the lead author of the study investigating the underlying genetic mechanisms behind why melanomas develop from some naevi, and not others.

“Moles come in many different shapes and size and some can change into a melanoma but often do not,” Dr Stark said.

“Melanoma can also form without even seeing a mole and trying to detect these changes is challenging but essential if we are to prevent melanoma formation.”

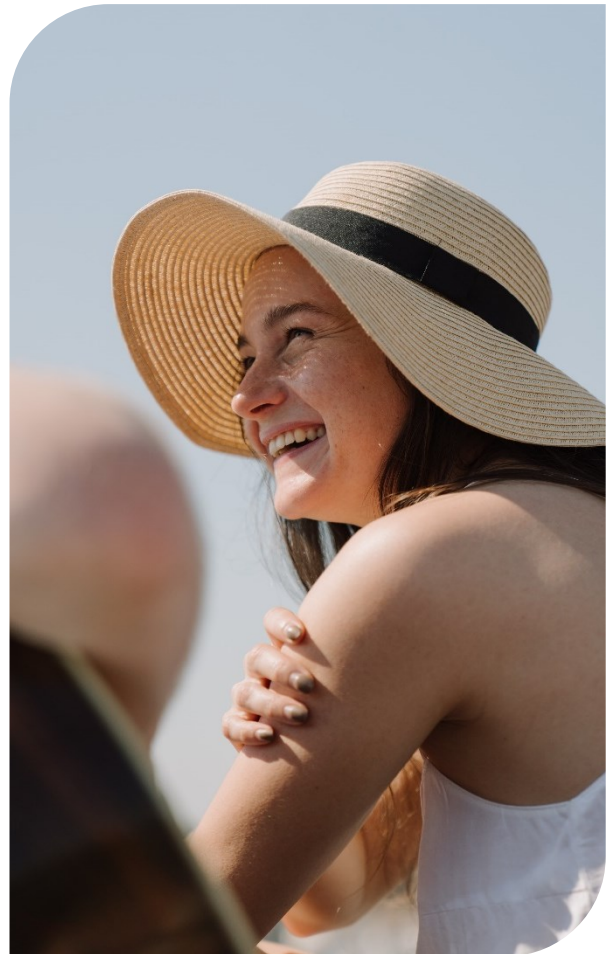
For a long time scientists have been studying the effects of sun exposure on our skin cells and it is well known that sun exposure can lead to skin cancer formation including melanoma.

“Luckily, the cells in our body have in-built sensors that detect when our DNA is damaged but sometimes these sensors do not work so well leading to damage in our DNA which may lead to cancer. In most cases, however, melanoma is not the first step and instead a mole develops.”

Scientists from UQ's [Dermatology Research Centre](#) analysed samples from participants in the Brisbane Naevus Morphology Study, and discovered that in all of the samples, moles had a lot of changes in their DNA (mutations) that were commonly seen in melanomas. The moles studied did however lack the necessary steps required for melanoma development.

“The moles sampled either were missing these necessary steps or the mole had other changes that ‘protected’ it from becoming a melanoma.”

Studies have consistently shown the number of naevi a person has is the strongest predictor of risk for melanoma. “In Australia, we have the world’s best melanoma prevention and surveillance programs leading to an increase in early detection of melanoma,”



“Moles are ‘simulators’ of melanoma and often they look just like an early melanoma and therefore they may be removed as a preventive measure.”

Dr Stark reinforced that people with a high number of moles, and other risk characteristics such as fair skin or light-coloured hair or eyes, should continue to see their treating Dermatologist or skin cancer doctor for routine skin examination.

Dr Stark is a National Health and Medical Research Council (NHMRC) Early Career Fellow.

The UQ Dermatology Research Centre is leading the Centre of Research Excellence for the Study of Naevi, and is based at the Translational Research Institute (TRI).

The research is published in the [Journal of Investigative Dermatology](#).

Physical combination multiplies melanoma risk

The risk of developing melanoma is significantly multiplied for people who have red hair and more than 20 large moles, University of Queensland researchers have found.

People with these physical traits have a one in four chance of developing the disease in their lifetime.

The study also showed that people without red hair, but who carry the variant gene for red hair, face an increased melanoma risk if they have a high number of moles.

Associate Professor Rick Sturm said the study investigated the possible correlation between established risk factors for melanoma.

“Independently, having red hair and lots of large moles over 5 millimetres in diameter are both known risk factors for developing melanoma,” Dr Sturm said.

“People with red hair have a risk two to four times higher, while those with many large moles are five times more likely to develop melanoma than the general population.

“But put the two risk factors together and the risk of developing melanoma isn’t simply added, it’s multiplied.”

Queenslanders have the highest risk factor in the world for developing melanoma, with one in 16 men and one in 24 women affected.

Dr Sturm said people with high numbers of moles should have regular physician-based skin checks, given they may be unaware they also carry the gene for red hair.

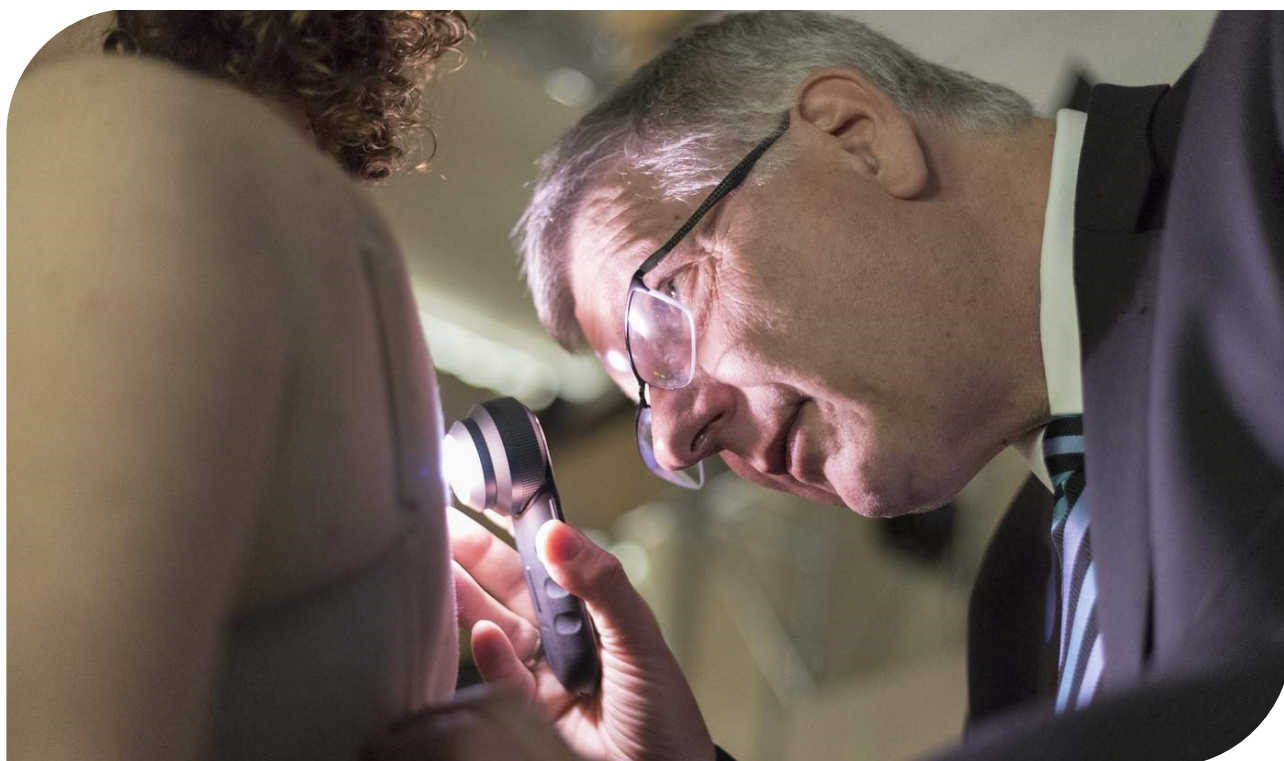
“It’s estimated that 25 per cent of the general population carry the gene for red hair,” he said.

“So there’s a need to boost awareness of the increased risk of developing melanoma for those who also have lots of moles.

“By keeping a closer eye on any skin changes in these high risk individuals, we can improve the chances of early detection of melanoma.”

The study investigated 1200 patients in the Brisbane Nevis Morphology Study, 50 per cent of whom have had melanoma.

The findings are published in the [British Journal of Dermatology](#).



Numbers count in the genetics of moles and melanomas

The University of Queensland scientists have identified a way to help dermatologists determine a patient's risk of developing melanoma.

[UQ Diamantina Institute](#) researcher Associate Professor Rick Sturm said the team uncovered the specific gene variations affecting the number and types of moles on the body and their role in causing skin cancer.

"The goal was to investigate the genetic underpinnings of different mole classes or 'naevi types' and understand how these affect melanoma risk," Dr Sturm said.

"Based on our work, the number of moles in each category can give a more complete assessment of melanoma risk rather than just the number of moles alone."

Three key mole classes, reticular, globular and non-specific were magnified under a dermoscope to assess their pattern and risk factors.

"We found people who had more non-specific mole patterns increased their melanoma risk by two per cent with every extra mole carried," he said.

"As we age, we tend to increase the amount of non-specific moles on our body, and the risk of developing melanoma increases."

Dr Sturm said globular and reticular mole patterns were also found to change over time.

"Globular patterns were shown to decrease as we get older, typically petering out after the age of 50 to 60," he said.

"Reticular moles also decreased over time but were likely to head down a more dangerous path and develop into the non-specific pattern."

A cohort of more than 1200 people, half melanoma patients, were recruited into the almost nine-year study.

Their results were then overlaid with genetic testing, which found variations in four major genes.

"We found some major relationships between genes and the number of moles and patterns when looking at the DNA," Dr Sturm said.

"Certain gene types influenced the number of different naevi types -- for example, the IRF4 gene was found to strongly influence the number of globular naevi found on the body."

The findings will help dermatologists to better understand mole patterns and provide more holistic care to patients who may be at risk of melanoma.

"For a long time, clinicians have been interested in how pigmented moles relate to melanoma and melanoma risk," he said.

"With the availability of dermoscopes and imaging, these results provide a new layer of understanding to guide clinical practice."

This research is published in the [Journal of Investigative Dermatology](#).



Melanoma risk in young Australians goes beyond the burn

Australians with melanoma detected before they turn 40 are more likely to have the cancer on non-sun damaged parts of the body compared to people diagnosed when older.

University of Queensland research found melanoma in places where sunburn was less common, such as the thighs, abdomen, buttocks and lower back, were more common in younger patients.

Diamantina Institute PhD candidate Dr Erin McMeniman said while younger melanoma patients had a greater likelihood of non-sunburn cancer due to their genetics, sun was still the greatest danger.

“It’s important for young people to be aware, particularly if they have more than 20 moles, that they have increased risk and that melanomas can grow in areas without visible sun damage.”

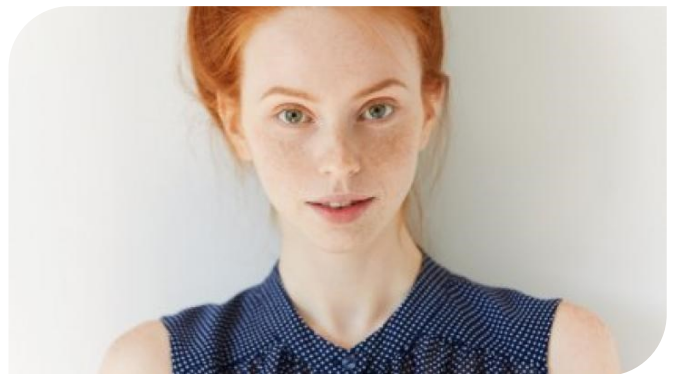
Melanoma is the most deadly form of skin cancer and affects more than 13,000 Australians each year.

The study took DNA samples and photographs from 585 volunteers with a history of one or more melanomas.

It showed people with multiple melanomas were more likely to have mutations in three genes, including the red hair gene.

“Melanoma patients are born with genetic mutations which make them more vulnerable to sun damage.

“If you live in Queensland and carry the CDKN2A mutation, the most well-known melanoma gene, you have about an 80 per cent risk of developing melanoma, whereas if you live somewhere getting very little sun, it might be much lower, or approximately 50 per cent.”



Genetic testing could help identify individuals carrying mutations and ensure they get appropriate monitoring, with early detection improving the chances of survival.

It should be considered if a person has been diagnosed with three or more melanomas, or if they have had at least one melanoma and two or more affected relatives.

“Genetic testing for melanoma isn’t available in the public system yet, but I think that will change in the future,” Dr McMeniman said.

“If people are concerned, they should see their GP or dermatologist for a risk assessment and if they are at high risk they will typically be seen every 6-12 months for a full skin examination.

“Of course, go to the doctor sooner if any marks grow or change in colour or shape - if in doubt, get it checked out.”

This research is published in the [British Journal of Dermatology](#).

"Phoning it in" is not always the answer for skin cancer detection

Australians' awareness of skin cancer using naked eye examinations is an effective way to detect suspicious moles and spots.

A new study from the University of Queensland compared people using naked eye self-monitoring to mobile teledermoscopy, a mobile phone fitted with a specialised camera, showed that both groups had a high degree of accuracy in detecting the suspicious spots.

These results were initially surprising to Professor Monika Janda, UQ Centre of Health Services Research.

"Although we had hypothesised that new mobile technologies would lead to a better detection rate, our study showed that both groups had overall high level of performance in finding spots and moles that a dermatologist also thought were worthwhile checking".

Queenslanders have the highest risk in the world for developing melanoma, with one in 16 men and one in 24 women affected.

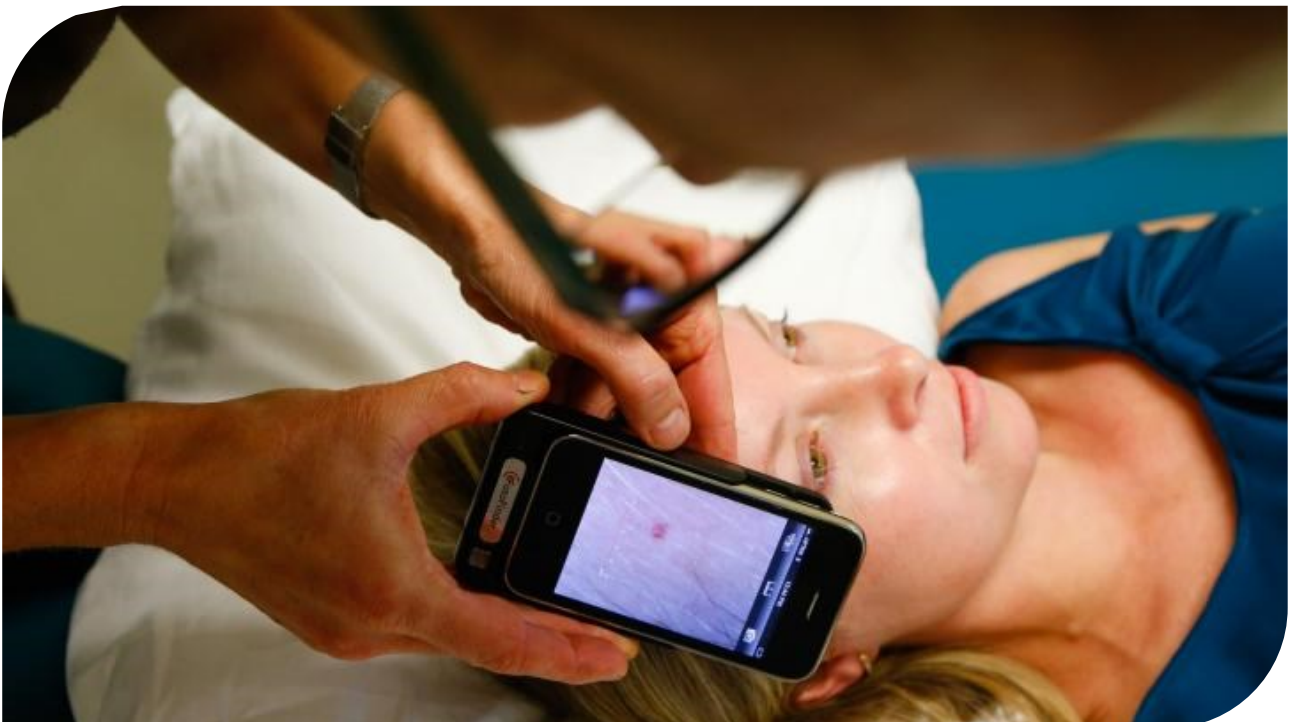
No population-wide screening program for melanoma currently exists in Australia.

As most skin cancers are first detected by the patient or their family, new methods for improving skin self-examinations at home could lead to further improvement in early detection and reduce the burden of melanoma.

"We hope that one day, with the integration of artificial intelligence, mobile teledermoscopy will be more effective at detecting melanoma and other skin cancers early". Professor Janda said.

"But as the mobile teledermoscopy group did not find more skin cancers, naked-eye skin self-examination as currently recommended still remains the standard."

The findings were published today in [The Lancet - Digital Health](#).



Pale melanomas masked by albino gene

People with pale coloured melanomas are more likely to have a gene mutation associated with albinism, University of Queensland research has found.

Study lead author Dr Jenna Rayner said albinism, a rare genetic disorder affecting one in 10,000 people, prevented brown pigment from being synthesised in the body and led to fair hair and extremely pale skin that was easily sunburned and prone to skin cancers.

“Albinism develops when there are two mutated genes, so people with one mutation usually don’t know they have it,” Dr Rayner said.

“These people may be more prone to developing pale coloured melanomas, called amelanotic, because tumours accumulate new mutations, and they already have a mutated albinism gene.”

The researchers studied DNA samples from more than 380 volunteers using whole exome sequencing, while looking for rare genetic mutations that cause albinism.

Queensland has the highest rate of melanoma in the world and more than 14,000 cases are diagnosed in Australia each year.

UQ Dermatology Research Centre Associate Professor Rick Sturm said up to eight per cent of melanomas could be amelanotic, making them difficult to diagnose and easily mistaken for non-cancerous conditions like warts or scars.

“Amelanotic melanomas are normally diagnosed in advanced stage, compared with darker melanomas, causing patients to often miss out on early treatment and their best chance of a cure,” he said.

When funding becomes available, researchers plan to collect amelanotic melanoma samples to compare their genotype with that of the patient.

Dr Rayner said it could lead to personalised medicine – where doctors would be alerted to monitor potential amelanotic melanomas in people with one albinism gene mutation.

“This could optimise early intervention and consequently improve patient outcomes,” she said.

This research has been conducted as part of the NHMRC Centre of Research Excellence for the Study of Naevi.

The paper was published in [PLOS One](#).



Presentations and Conferences

Title	Date	Conference/Presentation	Presenter
Phenotypic and genotypic risk factors for naevi and melanoma	27 – 30 July 2016	8th Annual HealthCert/University of Queensland Skin Cancer Conference and Masterclasses 2016	A/Prof Rick Sturm
Testing of viable clonal human skin cell cultures as an approach to validating microsampling of naevi	25 – 28 August 2016	Asia-Pacific Combined Dermatology Research Conference	Dr Stephen Ainger
Phenotypic and genotypic risk factors for naevi and melanoma	25 – 28 August 2016	Asia-Pacific Combined Dermatology Research Conference	A/Prof Rick Sturm
Apps and telemedicine	3 September 2016	16th World Congress on Cancers of the Skin, Vienna, Austria	Prof H. Peter Soyer
Skin cancer prevention in Australia	31 August 2016	16th World Congress on Cancers of the Skin, Vienna, Austria	Prof Monika Janda
Melanoma treatment & micro-RNA biomarkers	3 September 2016	16th World Congress on Cancers of the Skin, Vienna, Austria	Dr Mitchell Stark
Does your Research Make a Difference? Translation of Sun Protection and UVR Risk Reduction Behaviour into Community Settings	15 September 2016	CRE Naevi Seminar, Translation Research Institute, Brisbane	A/Prof Lois Loescher
Immune contexture of transforming naevi	13 October 2016	Australian Skin and Skin Cancer Research Centre, Inaugural Scientific Meeting	A/Prof Rick Sturm in place of Prof Mark Smyth
Germline variants in individuals of low and high nevus count	6 – 9 November 2016	Society for Melanoma Research Congress (SMR), Boston	A/Prof Rick Sturm
The AMPK- OGT axis prevents acquired drug resistance through inhibition of drug induced epigenetic remodelling in melanoma	6 – 9 November 2016	Society for Melanoma Research Congress (SMR), Boston	A/Prof Helmut Schaidler
The common ATM Ser49Cys variant is functionally defective for DNA damage response signalling	9 – 11 February 2017	29th Lorne Cancer Conference, Victoria, Australia	Dr Caroline Atkinson
Molecular and cellular analysis of naevogenesis	22 – 24 March 2017	Australia-Singapore Partnership in Skin Cancer Biology Conference, Biopolis, Singapore	A/Prof Rick Sturm
Genetic variation in IRF4 expression modulates growth characteristics, pigmentation and interferon-gamma response in melanocytic cells	22 – 24 March 2017	Australia-Singapore Partnership in Skin Cancer Biology Conference, Biopolis, Singapore	Dr Yash Chhabra
Freckling as a Marker of Melanoma Risk	6 – 9 May 2017	50th Annual Scientific Meeting of the Australasian College of Dermatologists, International	Dr Antonia Laino

		Convention Centre Darling Harbour, Sydney, NSW	
Combined genetic and cellular analysis of naevogenesis	9 – 10 May 2017	2017 Australasian Society for Dermatology Research (ASDR) Conference, Novotel Sydney Central, Sydney, NSW	A/Prof Rick Sturm
Update to Program 1:	19 May 2017	CRE for the Study of Naevi Scientific Meeting	A/Prof Rick Sturm
Whither mobile skin cancer detection	19 – 21 May 2017	Australasian Skin Cancer Congress, Gold Coast, QLD	Prof H. Peter Soyer
There are Moles and There are Moles	8 – 10 June 2017	Rotorua General Practice Conference and Medical Exhibition, Rotorua, New Zealand	Prof H. Peter Soyer
Targeted Melanoma Screening: Will Artificial Intelligence Take Over?	24 June 2017	Directions in Dermatology, Melbourne	Prof H. Peter Soyer
Point mutation in p14ARF-specific exon 1 β of CDKN2A causing familial melanoma and astrocytoma	5 – 8 August 2017	HGSA Annual Scientific Meeting, Brisbane Convention & Exhibition Centre, Brisbane	Dr Aideen McInerney-Leo
Human pigmentation genetics for the clinic	26 – 30 August 2017	23rd International Pigment Cell Conference (IPCC 2017), Denver, Colorado, USA	A/Prof Rick Sturm
Human pigmentation genetics to functional genomics with application across species	12 – 14 September 2017	Genomics and Collections: Adaptation to Macroevolution, CSIRO Discovery, Black Mountain, Canberra, ACT	A/Prof Rick Sturm
Debate- Detection of melanoma: the earlier the better	17 – 21 October 2017	World Congress of Melanoma, Brisbane Convention Centre, Brisbane	Prof H. Peter Soyer
Future perspectives of early detection of skin cancers	17 – 21 October 2017	World Congress of Melanoma, Brisbane Convention Centre, Brisbane	Prof H. Peter Soyer
Prevalence of common melanocytic naevi in the general population in South East Queensland	17 – 21 October 2017	World Congress of Melanoma, Brisbane Convention Centre, Brisbane	Dr Elsemieke Plasmeijer
The natural History of common melanocytic naevi in the general populations: systematic review and pilot results from the Mind your Moles Study	17 – 21 October 2017	World Congress of Melanoma, Brisbane Convention Centre, Brisbane	Dr Elsemieke Plasmeijer
Classifying dermoscopic patterns of naevi in a case control study of melanoma	17 – 21 October 2017	World Congress of Melanoma, Brisbane Convention Centre, Brisbane	A/Prof Rick Sturm
Whole exome sequence analysis of melanoma patients and unaffected control with low and high naevus count	17 – 21 October 2017	World Congress of Melanoma, Brisbane Convention Centre, Brisbane	A/Prof Rick Sturm

Genotypic and phenotypic correlations in a cohort of patients with multiple primary melanoma	17 – 21 October 2017	World Congress of Melanoma, Brisbane Convention Centre, Brisbane	A/Prof Erin McMeniman
Precision Dermatology	16 – 20 February 2018	American Academy of Dermatology Annual Meeting, San Diego, USA	Prof H. Peter Soyer
The impact of pigmentation genetics on cutaneous and ocular melanoma	6 March 2018	Focus on ocular melanoma - Networking event, Sponsored by the Mater Research Institute in collaboration with the Queensland Melanoma Collaborative, TRI Auditorium, Brisbane	A/Prof Rick Sturm
Naevus evolution to melanoma	6 March 2018	Focus on ocular melanoma - Networking event, Sponsored by the Mater Research Institute in collaboration with the Queensland Melanoma Collaborative, TRI Auditorium, Brisbane	Prof H. Peter Soyer
Circulating microRNAs in melanoma	6 March 2018	Focus on ocular melanoma - Networking event, Sponsored by the Mater Research Institute in collaboration with the Queensland Melanoma Collaborative, TRI Auditorium, Brisbane	Dr Mitchell Stark
Adaptive drug tolerance	6 March 2018	Focus on ocular melanoma - Networking event, Sponsored by the Mater Research Institute in collaboration with the Queensland Melanoma Collaborative, TRI Auditorium, Brisbane	A/Prof Helmut Schaidler
Whole-exome sequencing of acquired nevi identifies novel mechanisms for development and maintenance of benign neoplasms	14 – 18 April 2018	AACR Annual Meeting 2018, Chicago, Illinois, USA	Dr Mitchell Stark
A Prospective Longitudinal Population Based Cohort Study of the Natural History of Pigmented Skin Lesions in Adults: Preliminary Baseline Results	3 May 2018	4th International UV & Skin Cancer Prevention, Toronto 2018	Prof Monika Janda
Mobile health, consumer perception and dermatology	19 – 22 May 2018	The Australasian College of Dermatologists 51st Annual Scientific Meeting, Gold Coast Australia	Prof Monika Janda
Overview of melanoma genetics for dermatologists and results of a genetic analysis of multiple primary melanoma patients	19 – 22 May 2018	The Australasian College of Dermatologists 51st Annual Scientific Meeting, Gold Coast Australia	A/Prof Erin McMeniman
When will AI take over?	19 – 22 May 2018	The Australasian College of Dermatologists 51st Annual	Prof H. Peter Soyer

		Scientific Meeting, Gold Coast Australia	
Phenotypic and genotypic analysis of amelanotic melanoma patients drawn from a Queensland case-control study	19 – 22 May 2018	The Australasian College of Dermatologists 51st Annual Scientific Meeting, Gold Coast Australia	Dr Jenna Rayner
The impact of pigmentation genetics on cutaneous and ocular melanoma	14 June 2018	Redcliffe Hospital Grand Round, Australia	A/Prof Rick Sturm
A World Without Melanoma	15 – 18 August 2018	World Congress on Cancers of the Skin, Sydney, Australia	Prof H. Peter Soyer
Dermoscopy and Histopathological Correlations	15 – 18 August 2018	World Congress on Cancers of the Skin, Sydney, Australia	Prof H. Peter Soyer
Brisbane Naevus Morphology Study	15 – 18 August 2018	World Congress on Cancers of the Skin, Sydney, Australia	Prof H. Peter Soyer
Whole-exome sequencing of acquired naevi identifies novel mechanisms for development and maintenance of benign neoplasms	15 – 18 August 2018	World Congress on Cancers of the Skin, Sydney, Australia	Dr Mitchell Stark
The classification of naevus phenotypes in individuals at high risk of melanoma	15 – 18 August 2018	World Congress on Cancers of the Skin, Sydney, Australia	Ms Kaitlin Nufer
Will Artificial Intelligence Replace Humans in Melanoma Diagnosis?	12 – 16 September 2018	27th EADV Congress, Paris, France	Prof H. Peter Soyer
Host pigmentation factors and genotype analysis of melanoma patients: Analysis of melanocyte cell cultures	5 – 6 October 2018	Australasian Melanoma Conference 2018, Melbourne, Australia	A/Prof Rick Sturm
Whole exome analysis of naevi reveals mechanisms for development and maintenance of benign neoplasms	24 – 27 October 2018	15th International Congress of the Society for Melanoma Research, Manchester, UK	Dr Mitchell Stark
Whole exome analysis of naevi reveals mechanisms for development and maintenance of benign neoplasms	29 October – 1 November 2018	Cutaneous Biology Meeting 2018, Stradbroke Island, Brisbane	Dr Mitchell Stark
Iris freckling as a marker of cutaneous melanoma risk: a case-control study	22 – 23 November 2018	2018 Clinical and Public Health Symposium, Herston, Brisbane	Dr Antonia Laino
Functional analysis of MTAP haplotypes in melanoma risk	30 November 2018	5th Brisbane Cancer Conference, Brisbane	A/Prof Rick Sturm
IRF4 rs12203592*T/T genotype is associated with nodular melanoma	16 – 17 May 2019	2019 Australasian Society for Dermatology Research (ASDR) Conference, Melbourne	Dr Jenna Rayner

3D Total Body Photography for early detection of melanoma	16 – 17 May 2019	2019 Australasian Society for Dermatology Research (ASDR) Conference, Melbourne	Prof H. Peter Soyer
Change detection of skin lesions using 3D total body photography versus conventional dermoscopic images	18 – 21 May 2019	52nd Australasian College of Dermatologists Annual Scientific Meeting, Melbourne	Dr Stephen Thomas
Changes of melanocytic skin lesions in high-risk individuals and melanoma patients determined by 3D whole body photography and dermoscopy	18 – 21 May 2019	52nd Australasian College of Dermatologists Annual Scientific Meeting, Melbourne	Dr My Co Tran
Genes and Patterns of Nevi	10 – 15 June 2019	24th World Congress of Dermatology, Milan, Italy	Prof H. Peter Soyer
3D Total Body Photography	10 – 15 June 2019	24th World Congress of Dermatology, Milan, Italy	Prof H. Peter Soyer
Advance Imaging Technology in Early Melanoma Detection	10 – 15 June 2019	24th World Congress of Dermatology, Milan, Italy	Prof H. Peter Soyer
Artificial intelligence and 3D body imaging to identify naevi	26 July 2019	QIMR Berghofer Early Career Researcher Seminar Series, QIMRB, Brisbane	Dr Brigid Betz-Stablein
Mind Your Moles: Incorporating three-dimensional body imaging to identify naevi	20 August 2019	QIMR Berghofer Population Health Seminar, QIMRB, Brisbane	Dr Brigid Betz-Stablein
Melanoma Screening Summit	11 October 2019	ASSC Annual Scientific Meeting, held in conjunction with Global Advances and Controversies in Skin Cancer 2019, Brisbane	Prof Monika Janda
Artificial Intelligence and 3D body imaging to identify naevi	11 October 2019	ASSC Annual Scientific Meeting, held in conjunction with Global Advances and Controversies in Skin Cancer 2019, Brisbane	Dr Brigid Betz-Stablein
Applying psychological science to solving problems in cancer prevention	11 October 2019	ASSC Annual Scientific Meeting, held in conjunction with Global Advances and Controversies in Skin Cancer 2019, Brisbane	A/Prof Samuel Smith (2019 CRE Researcher in Residence)
Machine Learning in Total Body Photography of High-risk Melanoma Patients	9 – 13 October 2019	28th EADV Congress, Madrid, Spain	Prof H. Peter Soyer
AI for Melanoma Diagnosis	9 – 13 October 2019	28th EADV Congress, Madrid, Spain	Prof H. Peter Soyer
Update on the ACRF Australian Centre of Excellence in Melanoma Imaging and Diagnosis	6 November 2019	Centre of Research Excellence in Telehealth Symposium, Translational Research Institute, Brisbane	Prof H. Peter Soyer, Joachim Torrano, Dr Brigid Betz-Stablein

Increased frequency of albinism alleles in individuals with amelanotic/hypermelanotic melanoma	20 – 23 November 2019	16th International Congress of the Society for Melanoma Research, Salt Lake City, Utah, USA	A/Prof Rick Sturm
Melanoma early detection studies in Australia	20 – 23 November 2019	16th International Congress of the Society for Melanoma Research, Salt Lake City, Utah, USA	Prof Monika Janda
Genetics of human pigmentation, naevogenesis and melanoma	4 December 2019	The University of Queensland Create Change Seminar Series, University of Queensland	A/Prof Rick Sturm
DRC Research Update	6 March 2020	DRC Research Update to Doug Canfield, President at Canfield Scientific, Translational Research Institute, Brisbane	Prof H. Peter Soyer, Dr Aideen McNerney-Leo, Dr Brigid Betz-Stablein, A/Prof Liam Caffrey, Prof Monika Janda
Australian Centre of Excellence for Melanoma Imaging and Diagnosis (ACEMID) and first results of the Mind Your Moles Study	15 July 2020	UQDI Seminar Series	Prof H. Peter Soyer and Dr Brigid Betz-Stablein
A 4-decade odyssey in scientific research: from genes to dermatology	22 July 2020	UQDI Seminar Series	A/Prof Rick Sturm
Mutational signatures in benign neoplasms of the skin	22 July 2020	UQDI Seminar Series	Dr Mitchell Stark
ACEMID and Humans vs. AI	4 September 2020	2020 Huaxia International Skin Image Summit Conference, China	Prof H. Peter Soyer
Precision Prevention in Melanoma	23 September 2020	The Skin Hospital NSW	Prof H. Peter Soyer
Teledermatology in Australia	29 October 2020	29 th EADV Congress	Prof H. Peter Soyer
Clinical challenges of a 3D Total Body Imaging Telehealth Network in Australia	6 November 2020	8 th World Congress of Teledermatology 2020	Prof H. Peter Soyer
3D total body photography for the monitoring and early detection of melanoma	10 November 2020	Digital Health Institute Summit	Prof Monika Janda
Educating patients about early detection – what works?	14 November 2020	Skin Cancer College Symposium	Prof Monika Janda
The role of text messaging & social media in melanoma prevention	7 November 2020	US Melanoma Prevention Working Group Meeting	Prof Monika Janda
Mind your Moles Study: 3D total-body photography for the monitoring of naevi	9 December 2020	Herston Health Precinct Symposium	Prof Monika Janda

Research Publications

2016

Journal Publications

1. Snoswell C, Finnane A, Janda M, Soyer HP, Whitty JA. Cost-effectiveness of Store-and-Forward Teledermatology: A Systematic Review. *JAMA Dermatol.* 2016;152(6):702-708.10.1001/jamadermatol.2016.0525

2017

Journal Publications

2. Ainger SA, Jagirdar K, Lee KJ, Soyer HP, Sturm RA. Skin Pigmentation Genetics for the Clinic. *Dermatology.* 2017;233(1):1-15.10.1159/000468538.
3. Ainger SA, Yong XL, Soyer HP, Sturm RA. Testing of viable human skin cell dilution cultures as an approach to validating microsampling. *Arch Dermatol Res.* 2017;309(4):305-310.10.1007/s00403-017-1726-3.
4. Daley GM, Duffy DL, Pflugfelder A, Jagirdar K, Lee KJ, Yong XLH, . . . Sturm RA. GSTP1 does not modify MC1R effects on melanoma risk. *Exp Dermatol.* 2017;26(8):730-733.10.1111/exd.13114.
5. Finnane A, Dallest K, Janda M, Soyer HP. Teledermatology for the Diagnosis and Management of Skin Cancer: A Systematic Review. *JAMA Dermatol.* 2017;153(3):319-327.10.1001/jamadermatol.2016.4361.
6. Li X, Lee KJ, Duffy DL, Xu D, Basude MER, Hu Y, . . . Sturm RA. Acquired melanocytic naevus phenotypes and MC1R genotypes in Han Chinese: a cross-sectional study. *PeerJ.* 2017;5:e4168.10.7717/peerj.4168.
7. McWhirter SR, Duffy DL, Lee KJ, Wimberley G, McClenahan P, Ling N, . . . Sturm RA. Classifying dermoscopic patterns of naevi in a case-control study of melanoma. *PLoS One.* 2017;12(10):e0186647.10.1371/journal.pone.0186647.
8. Nguyen TM, Soyer HP, Green AC, Janda M. Do hand-addressed envelopes improve community response rates for a longitudinal study? *J Eval Clin Pract.* 2017;23(6):1422-1424.10.1111/jep.12816.
9. Plasmeijer EI, Nguyen TM, Olsen CM, Janda M, Soyer HP, Green AC. The Natural History of Common Melanocytic Nevi: A Systematic Review of Longitudinal Studies in the General Population. *J Invest Dermatol.* 2017;137(9):2017-2018.10.1016/j.jid.2017.03.040.
10. Schaidler H, Sturm RA. The evolving universe of BRAF mutations in melanoma. *Br J Dermatol.* 2017;177(4):893.10.1111/bjd.15829.
11. Janda M, Soyer HP. Automated diagnosis of melanoma. *Med J Aust.* 2017;207(8):361-362.10.5694/mja17.00618.

2018

Journal Publications

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2021

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Research Income

Secured as Chief Investigator

Year	Project Title	Granting Scheme	Amount (AUD)	Investigators
2015	CRE for the Study of Naevi	NHMRC CRE (2015-2019)	\$2,496,835 (60% for UQ)	Soyer HP Green A Aitken J Menzies S Sturm R Duffy D Janda M Prow T Schaidler H
2015	Engineering the Next Generation of Terahertz Laser Imaging Systems	ARC	\$644,767	Soyer HP
2015	Redesigning skin cancer early detection and care	NHMRC Partnership Project (2015-2017)	\$581,319	Janda M Soyer HP
2015	Funding in support of skin cancer research at UQ	Philanthropic funding from St Baker	\$600,000	Soyer HP
2015	Micromedical Device Development	NHMRC CDF	\$463,652	Prow T
2016	Predictors of mortality in thin melanomas	NHMRC Project (CCQ funded)	\$192,933	Green A Soyer HP
2016	A genomics approach for screening of patients at high risk of melanoma	QGHA Demonstration Project	\$598,845	Soyer HP Sturm R Schaidler H Janda M
2016	3D Qmelanoma - Targeted Early Detection of Melanoma Utilising a 3D Tele dermatology (3DT) Network	MABS HOPs	Funding for full time Project Coordinator for 3 years	Soyer HP Janda M
2017	Improving sun protection behaviour in young Australian adults using a digital behavioural intervention	Harry J Lloyd Charitable Trust	\$231,588 (USD) ~\$316,500 (AUD)	Soyer HP Janda M
2017	Implementation of an innovative tele dermatology network for the early detection of melanoma in high risk Australians	NHMRC Partnership Grant	\$2,500,232	Soyer HP Aitken J Janda M
2017	Personalised Early Detection of Melanoma	MRFF Next Generation Clinical Researchers Program Practitioner Fellowship (Lvl2)	\$577,188	Soyer HP
2017	Cutting-edge Imaging System for the early diagnosis of face and scalp skin cancer	Honda Foundation	\$21,362	Soyer HP

2017	Targeted and personalised early detection of melanoma using a 3D teledermatology network	PA Research Foundation Translational Research Innovation Award	\$100,000	Soyer HP Janda M Schaider H Sturm R
2018	Australian Centre of Excellence in Melanoma Imaging and Diagnosis (ACEMID)	ACRF	\$9,889,200	Soyer HP Janda M Aitken J Menzies SM
2019	Early Progression of Melanoma - Unravelling the molecular signatures in skin and precursor lesions that lead to melanoma development	New Merchant Charitable Foundation Funding	\$1,000,000 (\$200,000 per year for 5 years) Additional \$500,000 from UQ	Soyer HP
2019	Assessing diagnostic accuracy for melanoma, with compared to without access to Melanoma Surveillance Photography in high-risk individuals.	MRFF Targeted Health System and Community Organisation Research Grant	\$2,416,998	Soyer HP Janda M
2020	Targeted Early Detection of Melanoma Utilising a 3D Teledermatology Network	MRFF Rapid Applied Research Translation (RART) Program	\$233,190	HP Soyer
2021	Intelligent total body scanner for early detection of melanoma	NHMRC European Union Collaborative Research Grant	\$499,963	Soyer HP Janda M Duffy D
2021	Roadmap Options for Melanoma Screening in Australia (Melanoma-ROSA)	NHMRC Synergy Grant	\$5,000,000	Janda M Soyer HP
2021	CRE in Skin Imaging and Precision Diagnosis	NHMRC CRE	\$2,500,000	Janda M Soyer HP
2021	Establishing a familial melanoma clinic	SERTA Metro South Grant	\$300,000	HP Soyer

Funding received as collaborator

Year	Project Title	Granting Scheme	Amount (AUD)	Investigators
2015	Australian Skin and Skin Cancer (ASSC) Research Centre	UQ Strategic Funding	Per year: \$75,000 UQ Central, \$37,500 M+BS, \$37,500 UQDI, \$150,000 QIMRB. Total \$900,000	HP Soyer Janda M
2015	Identification of the molecular hallmarks of naevi progressing to melanoma	NHMRC ECF (Peter Doherty Biomedical)	\$314,000	Stark M
2015	Novel Microdevices for Controlled Blood and Skin Extraction	NHMRC ECF (Peter Doherty Biomedical)	\$314,644	Lin L
2017	Assessment of precision melanoma diagnostics	Advance Queensland Innovation Partnership (AQIP)	\$240,000	Stark M
2019-2021	Australian Skin and Skin Cancer (ASSC) Research Centre	UQ and Queensland Institute for Medical Research Berghofer (QIMRB)	\$500,000 over 3 years from both UQ and QIMBR	HP Soyer Janda M Stark M
2021	Artificial Intelligence and 3D imaging of the total skin surface in Australians with skin of colour	Australian Skin and Skin Cancer (ASSC) Research Centre	\$20,000	Betz-Stablein B
2021	Evaluating Artificial Intelligence and 3D imaging in Australians with skin of colour	Australian College of Dermatology	\$25,000	McMeniman E Betz-Stablein B Soyer HP



More information

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