



# 2015 ANNUAL REPORT

1 January - 31 December 2015

*Twin research: Discoveries for  
a healthier community*



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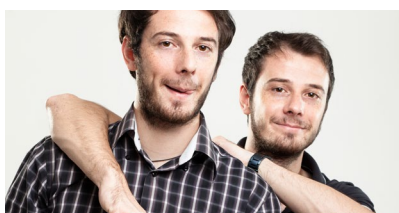
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# 2015 HIGHLIGHTS



## Membership in 2015

ATR membership grew to **45,036** twin pairs and **440** triplet sets

**1,239** new twin & **11** triplet sets

**1,832** twin pairs participated in studies



## First Twin Research Hub Launched in Sydney

Based at the University of Sydney's Charles Perkins Centre, the hub aims to grow twin research in NSW and to generate new collaborations to address major health challenges.



## Building capacity

In 2015 the ATR supported **two** PhD students

Lucas Ferreira's project aims to understand the challenges of twin research in the context of global scientific collaboration and their implications for the development of the Brazilian Twin Registry.

Shuai Li is investigating DNA methylation based on the Australian Mammographic Density Twins and Sisters Study.



## Research

**16** twin studies supported by the ATR

**40** publications

Our research covers a broad spectrum of health issues – diabetes, breast cancer, back pain, epilepsy, speech and language disorders, obesity and brain ageing.

**WELCOME  
TO THE 2015  
ANNUAL  
REPORT OF THE  
AUSTRALIAN  
TWIN REGISTRY**

# HIGHLIGHTS AND ACHIEVEMENTS

## SUMMARY 2015

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This report summarises the Australian Twin Registry's (ATR) major achievements, activities and research developments during 2015.

The 2015 reporting period covers the first year of the new Centre of Excellence Grant (2014 – 2019) from the National Health and Medical Research Council (NHMRC) that will fund the ATR for five years.

### RESEARCH

- ATR membership numbers grew again from 2014, with 1,250 new sets of twins and triplets registered in 2015.
- The ATR recruited for 16 studies in 2015; with 3 new studies looking at:
  - Personality, group membership and processes, political attitudes, and health
  - Back pain
  - Sleep and back pain

### KNOWLEDGE EXCHANGE

- Twins Plus Festival
  - This year's festival hosted the largest research component to date, with approximately 40 researchers conducting studies
- International Network of Twin Registries: Osaka
  - In 2015, more than 20 twin registries from across the world were represented
- The ATR reviewed and enhanced the range and quality of services that it provides to the research and twin community. This increased the ATR's capacity to support research that has the potential to contribute to the health and wellbeing of all Australians. This was achieved by:
  - Providing feedback to the twin community and the wider Australian community about research findings via the ATR's website, e-news, Facebook, Twitter, national media releases and the ATR newsletter

### BUILDING CAPACITY

- Charles Perkins Centre (CPC) Twin Research Project Node Launch
  - The CPC Twin Research Project Node at the University of Sydney is a unique opportunity for NSW researchers to make their mark globally. The Node will link directly to more than 60 other Project Nodes across the CPC's research strategy and provide a resource for generating new research opportunities.
- The Travel Grant Scheme supported 10 researchers to attend national and international research workshops and conferences to present their twin research.

### KNOWLEDGE TRANSLATION

- The ATR enabled the publishing of 40 peer-reviewed articles based on ATR-related studies, as well as 1 book and 8 conference proceedings.

# DIRECTOR'S REPORT

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Twin research has the potential to contribute transformative insights to our understanding of health and disease. A new NHMRC Centre of Excellence Research Grant (CRE), awarded at the end of 2014, positioned the Australian Twin Registry for the future of twin research, enabling capacity building and the development of local and international collaborations. The chief investigators, advisory board, researchers, Registry members and staff have all worked hard to achieve several milestones already.

In October, we launched a new twin research hub with the University of Sydney's Charles Perkins Centre, providing a unique opportunity for New South Wales researchers to make their mark globally. This will generate new collaborations to address major health challenges both in Australia and internationally.

Celebrating our work with twins, we also co-hosted the triennial Twins Plus Festival with the Australian Multiple Birth Association. This event

is a rare opportunity for twins, triplets and their families to come together and share the special experiences that being a multiple, or parenting a multiple, entails. It also showcases the unique contribution twins make to health and medical research and provides an opportunity to learn about what research participation entails – 2015's Festival hosted the largest research component at the event to date, with approximately 40 researchers conducting studies. I would like to thank all of the twins, triplets and their families who came along and helped us celebrate, and those volunteers who generously gave their time to help make the day a success.

Continuing to strengthen our international collaborations with other twin registries around the world, we participated in the fourth meeting of the International Network of Twin Registries, hosted by the Centre for Twin Research at the Osaka University Graduate School of Medicine in September. The International Network of Twin Registries is a working group under the

International Society of Twin Studies, and aims to develop harmonisation activities to foster active internal collaborations between twin registries around the world. More than 20 twin registries were represented at this meeting.

We have made a substantial start to our work under the CRE grant and are well-positioned to achieve our goals ahead: to enrich the Registry, progress study design, develop the twin research workforce, build local and international collaborations and translate new knowledge for the health of the community.

Warm regards,

A handwritten signature in black ink, appearing to read 'John Hopper', written in a cursive style.

John Hopper  
**Director,**  
**Australian Twin Registry**



# OUTLOOK FOR 2016

The ATR will continue to be supported by the funding structure of the Australian Centre of Excellence in Twin Research (ACETR). We will continue to drive new and innovative twin research by collaborating with leading researchers within Australia and internationally. The ATR will also continue to focus on strengthening our relationships with both the research and twin communities and, by doing so, assist with the translation of research knowledge to the community.

This will be achieved in the research community by:

## 1

Activating novel fields of enquiry and enabling interdisciplinary research across the areas of cancer, chronic disease, infection, mental health and genomics.

## 2

Increasing the scale of our international research collaborations by supporting the creation of new twin registries and engaging with existing ones across the globe. This will lead to larger, more robust studies, opportunities to study rare diseases, and also capture genetic variation across countries/races/genetic backgrounds.

## 3

Deepening our engagement and support of our community of twin pairs and their families by establishing a Stakeholder Advisory Committee. This will ensure we fully understand the needs and expectations of the twin community and incorporate their views into the ATR's overall goals.

# ABOUT



## About the ATR

Established in 1981, the ATR is a national volunteer register of twins interested in contributing to research studies. The primary goal of the ATR is to facilitate and support research studies involving twins. In 2015, the ATR maintained information on 45,032 sets of twins and triplets, and supported almost 83 research projects in the grant application, active recruiting, data collection or writing phase, covering a broad spectrum of health-related themes.

The ATR's vision is to realise the full potential of research involving twins to improve the health and wellbeing of all Australians, and in 2015 the ATR made significant progress towards achieving this vision.

“ Realise the full potential of research involving twins to improve the health and wellbeing of all Australians

## Potential for Twin Research

The ATR provides twins with the opportunity to contribute to, and make a difference in, the development of knowledge around health and medical issues that affect all Australians.

Studies involving twins play an important and unique role in developing an understanding of good health and clinical problems from a genetic and environmental perspective. Twins provide a potential resource and research tool for all medical and scientific researchers.

Twin research continues to utilise new technologies to establish the causes underlying many health and medical issues that affect Australians. Twin studies have started to play a vital role in the emerging search for epigenetic effects produced by proteins and other molecules that bind to DNA, changing gene expression. Such epigenetic effects are a recently recognised phenomenon and have been associated with many diseases, including cancer and psychiatric disorders. Studies involving twins can significantly contribute to the investigation and identification of epigenetic factors that contribute to human disease, through twins' shared environments and genetics.



# The ATR's core functions are:

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## ■ Core function 1: Research

Collaborate with researchers applying to the ATR to ensure that projects are of significant scientific merit and are appropriately described to ensure the ability of potential participants to provide informed consent.

## ■ Core function 2: The Registry

Continue the building and maintenance of an up-to-date database containing contact details and baseline information for twin members willing to participate in research.

Use judicious management and administration in approaching eligible twin members to inform them of a new research project, determine their interest in participation, and seek their permission to release their contact details to the researcher for the purpose of the project.

## ■ Core function 3: Knowledge Exchange

Develop projects and programs to value-add to research in Australia.

## ■ Core function 4: Building Capacity

Develop projects and programs to build capacity in twin research in Australia.

## ■ Core function 5: Governance

Apply governance of the ATR in a fair, transparent and equitable manner.





# RESEARCH

Collaborate with researchers applying to the ATR to ensure that projects are of significant scientific merit and are appropriately described to ensure the ability of potential participants to provide informed consent.

## PROFILE STUDIES RECRUITED FOR IN 2015

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### The Older Australian Twins Study Amyloid Imaging Project

The Older Australian Twins Study, led by Perminder Sachdev and Julian Trollor of the Centre for Healthy Brain Ageing at the University of New South Wales, has been recruiting ATR twins (65 years old and older) for more than nine years and has made a substantial contribution to understanding brain ageing. Brain ageing and brain diseases are determined by multiple genetic factors that interact with environmental influences. Since identical twins share 100 percent of their genes, whereas non-identical twins share half their genetic information, detailed comparisons of these two groups has the potential to discover new genes involved in cognitive decline or resilience.

The OATS aims to find out what influences memory and thinking as

we age. It investigates environmental influences such as lifetime physical and mental activity, socioeconomic environment and nutrition. It also investigates how biological factors such as hypertension and antioxidant levels, interact with genes to influence brain ageing. Since, over time, the expression of genes varies depending on different influences in the environment, by studying twins, OATS aims to determine which influences on the ageing process are genetic, which are environmental, and how the two interact.

In 2015 a new OATS sub-study was established to investigate the deposition of amyloid plaques in the brain using positron emission tomography (PET) scans. Amyloid plaques are thought to predict memory decline with age. Performing these scans on twins will help the researchers establish how amyloid plaques relate to performance in memory and thinking ability. The researchers also aim to determine if there is a genetic component, and if

there are any potentially modifiable environmental factors that may be contributing to the development of the plaques.





(Left to right) Evangelos Pappas, Mauricio Magalhaes, Manuela Ferreira and Paulo Ferreira

## The Australian Twin Back Pain Study

Nearly 80 percent of the population have had, or will experience lower back pain, making it the most commonly suffered pain condition and the third most common long-term condition.

Researchers, Dr Paulo Ferreira and Marina de Barros Pinheira, from the University of Sydney are undertaking a study with the Australian Twin Registry to better understand the types of physical activities that cause

and protect low back pain as well as whether treatments aimed at increasing sleep quality are able to help people with back pain.

Dr Ferreira reports that low back pain is the most disabling condition worldwide in terms of number of years lived with disability and “it significantly affects people’s lives and adds a lot of suffering and burden”. Moreover it costs \$4 billion annually in direct and indirect costs. With an increasing number of people leading more sedentary lifestyles in demanding and stressful occupations, and a growing lack of

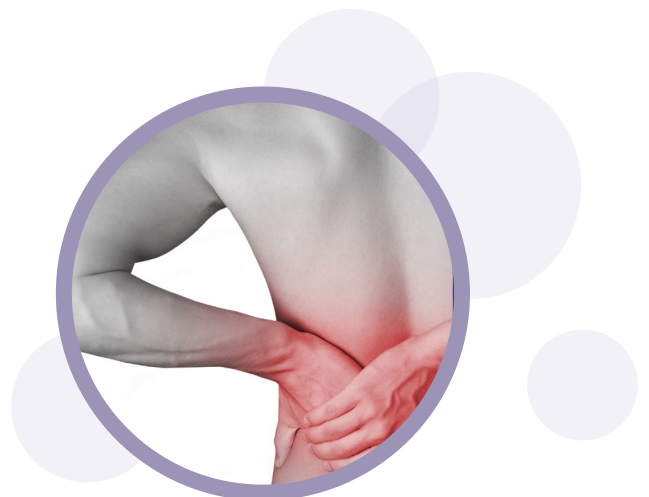
opportunities for social interaction and support, the incidence of low back pain looks set to soar.

His team is interested in studying twins (18 years old or older) who have and who have not suffered back pain in order to identify what factors are involved in this condition.

Dr Ferreira argues that twins are ideally suited for such a project “because they allow us to investigate the contribution of familial factors in the development of back pain and the response of people to treatment approaches.”

“ With an increasing number of people leading more sedentary lifestyles in demanding and stressful occupations, and a growing lack of opportunities for social interaction and support, the incidence of low back pain looks set to soar.

-Dr Ferreira



# CURRENT RESEARCH STUDIES

The ATR continues to enable research through the recruitment of twins for health-related studies. In addition to driving key research projects, it also provides a service by which researchers can collaborate with us to recruit twins and connect with the twin community.

Studies are classified into three areas:

- Those in the initial stages of planning and development;
- Those involved in active recruitment;
- Post-recruitment and statistical support

There were 26 active and ongoing studies utilising ATR services and/or involving ATR members in 2015. This includes the active processing of three Expressions of Interests (EOIs) for new research, four new research applications (as a result of approved EOIs) and three amendments processed throughout the year. In total, there were 16 active recruiting studies with participants in 2015, four of which were completely new, and the ATR provided ad hoc support to a further 57 studies in varying stages of study development, data collection data analysis and writing up.

A complete count of all studies, by status, as of 31 December 2015, is shown in Table 1 and a count of active recruiting studies in the past seven years is shown in Figure 1.

Study Status reached by end of 2015	Number
Application (EOIs, Full Application, Protocol Change)	10
Recruiting	16
Data Collection	4
Data Analysis	43
Writing Up/Publishing	10
<b>TOTAL</b>	<b>83</b>

Table 1: Studies by Status 2015.

## Number of Active Recruiting Studies 2006-2015

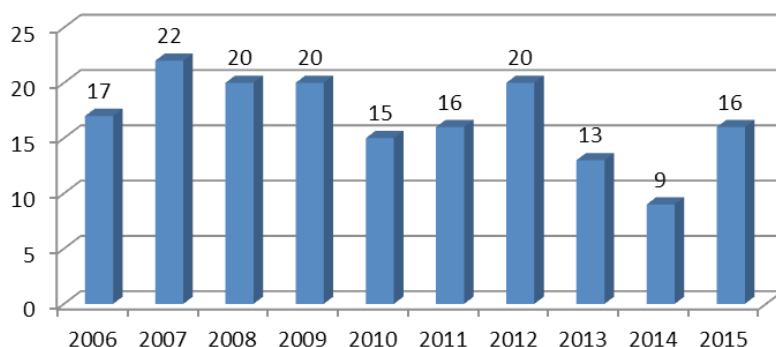


Figure 1. Number of Active Recruiting Studies, Reported as at Annual Report Publication.



*Growing Pains and Functional Pain Disorders*  
 David Champion, Department of Anaesthesia and Pain Medicine, Sydney Children's Hospital

"We could not have asked for anything more."

"The assistance has been invaluable and essential."



*Sleep and Low Back Pain*  
 Paulo Ferreira, Discipline of Physiotherapy, Faculty of Health Sciences, The University of Sydney

"The ATR, in my opinion, fulfils its mandate expertly. It understands the needs of researchers, and has the right tools to assist them."

"The ATR was invaluable to our project. At the most basic level, the ATR's existence made possible the entire collection of the data analysed in our study. On a deeper level, the ATR had the tools on hand to assist with the proper and rigorous analysis of this data."



*Beauty and the Eye of the Beholder*  
 Jeremy Wilmer, Psychology Department, Wellesley College, Wellesley, Massachusetts

"It has always been such a pleasure to work with every person at ATR. It is an incredible organization and an incredible group of people."

# RESEARCHER SATISFACTION

The 2015 Annual Researcher Satisfaction survey was administered as part of the Annual Progress Report submitted by researchers. It requested feedback relating to the previous 12 months on:

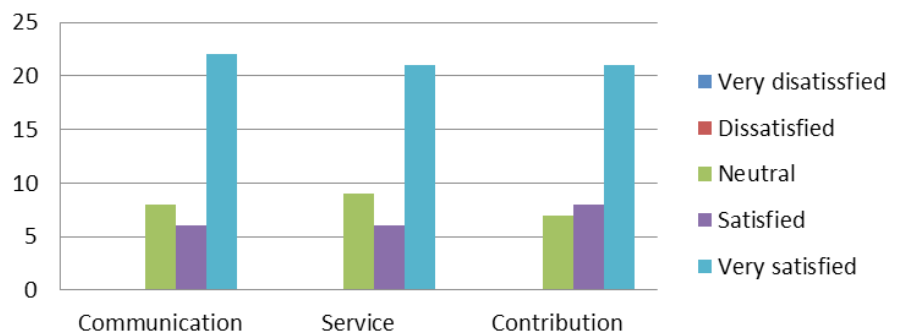
- (1) the researcher's overall satisfaction with communication with the ATR;
- (2) the researcher's overall satisfaction with the services that the ATR provided; and
- (3) the value of the contribution that the ATR made to the overall research project

Responses were recorded as

1 - Very Dissatisfied / 2 - Dissatisfied / 3 - Neutral / 4 - Satisfied / 5 - Very Satisfied

We received feedback from 36 research groups for 2015. Overall, researchers were very satisfied with the contribution provided by the ATR and also the communication and service the ATR provided.

## Scores for Overall Satisfaction of Researchers who have interacted with the ATR in 2015



**Figure 2. Overall Satisfaction Scores from Researchers, Evaluating Communication with ATR, and Service and Contribution Provided by the Registry.**





# THE REGISTRY

Continue the building and maintenance of an up-to-date database containing contact details and baseline information for twin members willing to participate in research.

Use judicious management and administration in approaching eligible twin members to inform them of a new research project, determine their interest in participation and seek their permission to release their contact details to the researcher for the purpose of the project.

# DATABASE

The ATR maintains an up-to-date register of twins (or in the case of twins under the age of 18, their parents) willing to consider involvement in scientific studies. This register is supported by a comprehensive database, which retrieves updated membership data to allow accurate record keeping and analysis of trends and results.

The ATR continues to update and improve internal database processes and mechanisms to better assist staff in providing a cost effective and efficient service to twins and researchers.

# MEMBERSHIP

The ATR volunteer members are an integral part of the organisation, and management of the membership is a core component of its function. Twins and Higher Order Multiples (HOMs), including triplets, quadruplets and quintuplets of all ages, sex combinations and zygosity are eligible to enrol with the ATR. The current status of members of the ATR is summarised in Table 2. Junior members represent approximately (25%) of the entire Registry, the remaining (75%) being senior members.

The top row of Table 2 details display T1 (twin one) status, and the left most column details T2 (twin two) status.

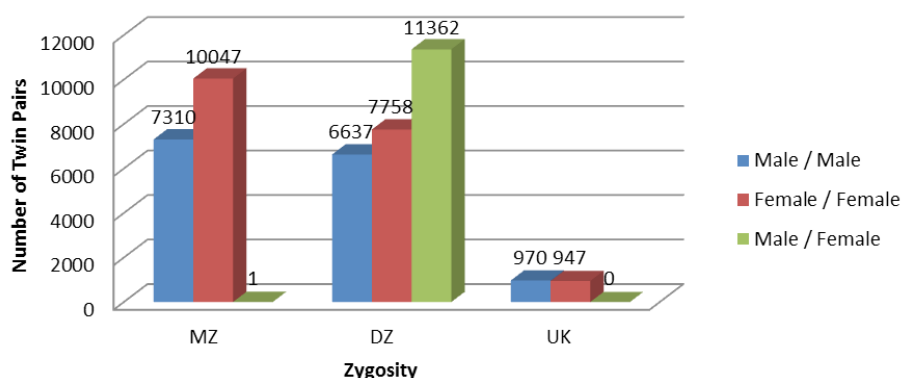
		Twin 1				
Twin 2	T1/T2 Status	Active	Inactive	Deceased	Lost/Pending	Total
	Active	33067	579	435	1648	35,729
	Inactive	512	1724	302	137	2,675
	Deceased	400	260	597	68	1,325
	Lost/ Pending	1690	133	74	3406	5,303
		35,669	2,696	1,408	5,259	45,032

**Table 2: Twin Pair Registrations.**

- Inactive: Twin member(s) have requested to withdraw from registry activities.
- Pending denotes members for whom we do not have current details; so they are 'pending' an update.

The current numbers of active twin pairs by sex and zygosity are shown in Figure 3. It does not show the 860 'lost' or 'unknown' twin pairs.

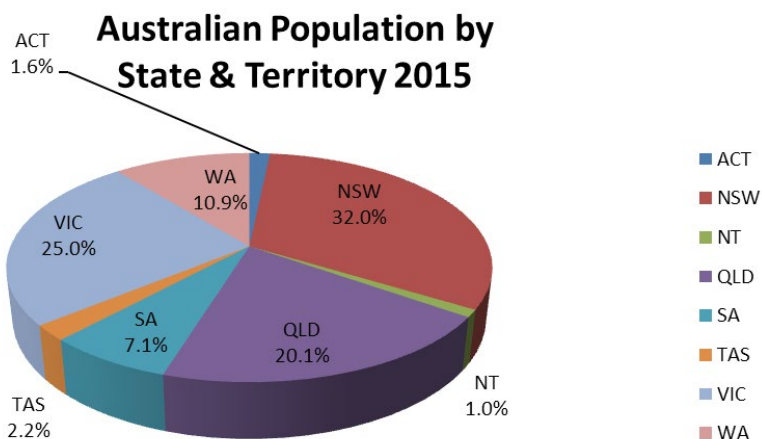
### Active Twin Pairs by Sex and Zygosity 2015



**Figure 3. Active Twin Pairs by Sex and Zygosity 2015.**

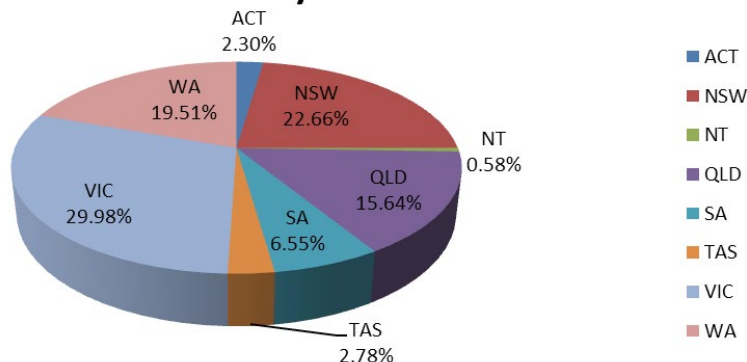
\*MZ = monozygotic, DZ = dizygotic, UK = unknown

The distribution of active twin pairs by location is shown in Figure 5, together with the overall distribution of the Australian population by State and Territory (as reported by the Australian Bureau of Statistics in 2015) in Figure 4. Comparison of the two graphs shows that most populated states are also the states where most active ATR members reside.



**Figure 4. Australian Population by State 2015.**

### Distribution of Active Twin Pairs by State & Territory 2015

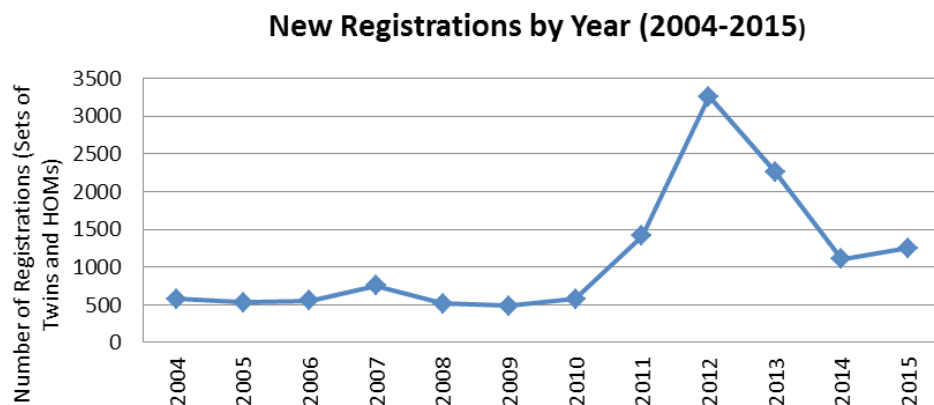


*Note: Because some twins live in separate states or one twin member in a pair lives overseas, this excludes twins overseas, or with no address available.*

**Figure 5. Distribution of Active Twin Pairs by State 2015.**

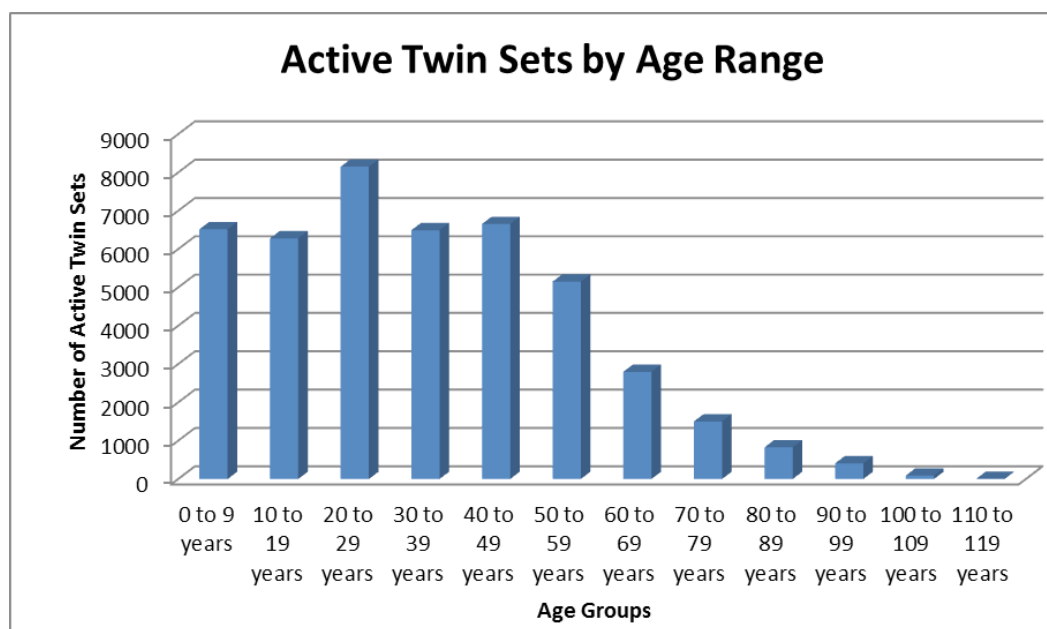


The numbers of new registrations by year, since 2004 are represented in Figure 6.



**Figure 6. Number of New Twin and HOM Sets Registered with ATR by Year Since 2004.**

*\*2012 peak: Merge of Western Australian Twin Registry with Australian Twin Registry*



**Figure 7. Active Twin Pairs Shown by Age Group**

The distribution of active pairs of twins in the Registry, is shown by age groups in Figure 7. The majority of new members enrolled with the ATR are within their first year of life and mainly recruited through AMBA and the Twin Pregnancy Booklet.

# STUDY APPROACHES

Mail outs to prospective participants for individual studies are a core component of the Registry's daily operations. Scheduling of mail outs and the total number of approaches sent is dependent on the requirements of the researcher. During 2015, 46 mail outs were conducted and a total of 10,465 letters or emails were sent (**Table 3**).

Study ID	Project Leader	Institute	Study Title
2005-003-3	Prof. Perminder Sachdev	University of New South Wales	OATS Amyloid Imaging Project
2008-001-4	Prof. Ego Seeman	Austin Health	Effect of Menopause on Bone Structure in Twins
2012-003	Prof. Brian Byrne	University of New England	A Twin Study of the NAPLAN
2013-002	Dr Fiona Barlow	University of Queensland	Genetics and Personality as Predictors of the Extent to which We Join, and Identify with, Groups.
2014-004/ 2015-004	Dr Paulo Ferreira	University of Sydney	Efficacy of a Sleep Quality Intervention in People with Chronic Low Back Pain Controlling for Genetics: A Preliminary Randomised Co-Twin Controlled Trial / AUTBACK
ATR-002	Prof. John Hopper	University of Melbourne	ATR Junior Health and Lifestyle Questionnaire
ATR-001	Prof. John Hopper	University of Melbourne	ATR Senior Health and Lifestyle Questionnaire
2012-012	Prof. Russell Keast	Deakin University	Why Do Some People Overconsume Fatty Foods
2007-005-7	A/Prof. David Champion	Sydney Children's Hospital	Adolescent Primary Dysmenorrhea: Genetic Risk and Associations with Primary Pain Disorders and Related Conditions
2007-005-8	A/Prof. David Champion	Sydney Children's Hospital	Paediatric ADHD: Genetic Risk and Associations with the Primary Pain Disorders and Related Conditions
2012-014	Prof. John Hopper	University of Melbourne	A Study of Breast Cancer and Epigenetics

2007-004	Prof Dianne Fatkin	Victor Chang Cardiac Research Institute	Role of Genetic and Environmental Factors in Atrial Fibrillation
2008-002-3	A/Prof. Velandai Srikanth	Peninsula Health	Type 2 Diabetes and Brain Function – a Co-Twin Study of Brain Metabolism
2012-001	Dr Peter Simm	Murdoch Childrens Research Institute	Epilepsy and Bone Health
2008-006	Prof. Sam Berkovic	Epilepsy Research Centre	Genetics of Syncope and Breath Holding(Syncope & Breath Holding)
2008-006-2	A/Prof. Lata Vadlamudi	Royal Brisbane and Women's Hospital	Genetics of Epilepsy
2013-005	Dr Angela Morgan	Murdoch Childrens Research Institute	Genetics of Speech Disorders
2008-001-2	Prof. Ego Seeman	Austin Health	Menopause and Bone Health

**Table 3: Study Approaches.**

## FOLLOW UP

As part of its services, the ATR offers researchers several follow-up options. In 2015, email continued to be the dominant tool for sending reminders to ATR members. Although email is potentially far more efficient and

economical, phone and mailed reminders boost response rates. SMS text reminders will be piloted in 2016 to become a key reminder tool in future years.



## CURRENCY AND ACCURACY OF MEMBERSHIP CONTACT DETAILS

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Maintaining active members at the ATR requires constant work to ensure we have current membership contact details. This is done in a number of ways such as: providing opportunities to update details on the website and through the E-News; following up return to sender

mailed communication through study approaches, the printed newsletter and via phone calls.

## RECORD UPDATES

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Maintaining active members at the ATR requires constant work to ensure we have current membership contact details. In 2015 we updated 7,392 individual twin and other contacts (i.e. parent or secondary carer) member records. This is mainly done online through the website

and the E-News; but also through following up return to sender mailed communication through study approaches, the printed newsletter and phone calls to the twins' or second and third contacts.

## HEALTH & LIFESTYLE QUESTIONNAIRE

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The Health and Lifestyle Questionnaire (HLQ) was launched in 2014, in part, to better describe the ATR membership through the publication of summary statistics to both the twin and the researcher communities. These statistics include zygosity, age, gestation term and medical conditions. To date, over 6,100 adult twins, including over 1,500 pairs and over 3,000 parents of twin children have completed the HLQ.

The HLQ enables identification of potential participants for specific research studies to reduce costs to researchers and unnecessary approaches to ineligible ATR members. It also enhances fast-track research by

providing de-identified data to researchers for ethically approved research analyses.

There has been a lot of interest in the HLQ data and two research groups conducted studies using this de-identified data in 2015. In addition to applications for health and lifestyle questionnaire data, information from the questionnaire has been used to target recruitment for an additional three studies



## KNOWLEDGE EXCHANGE

Apply best practice to translating research  
outcomes to the end users



# TWINS PLUS FESTIVAL



*Australia's oldest identical triplets Florence, Betsy and Dorothy, aged 75, at the 2015 Twins Plus Festival*

Held on March 22nd 2015, the Twins Plus Festival hosted the largest research component at the event to date. Ten research groups held stands and approximately 40 researchers registered to attend. Research studies included: speech disorders, brain injury, fat-taste, back-pain, eating disorders, singing, autism, peri/postnatal, epilepsy, and childhood pain disorders. Four studies conducted recruitment through the Twins Plus Festival. The research stands were highly popular and several researchers successfully recruited or took expressions of interest from substantial numbers of twin pairs.

## INTERNATIONAL NETWORK OF TWIN REGISTRIES: OSAKA

The fourth meeting of the International Network of Twin Registries (INTR) was hosted by the Centre for Twin Research at the Osaka University Graduate School of Medicine, 28-29th September, 2015. The INTR is a working group under the International Society of Twin Studies (ISTS), and aims to develop harmonisation activities and foster active internal collaborations using twin registries around the world.

In 2015, more than 20 twin registries were represented at this meeting. Working groups were formed to address topics relevant to fostering

twin research such as innovation and communication, stakeholder and industry engagement and novel funding. A newsletter has been established and has successfully promoted new ideas and proposals for multi-centre twin studies within the network. An online platform to facilitate data and knowledge sharing between researchers is also being planned.

Due to growing interest in INTR from the scientific community, an INTR session within the International Congress of Twin Studies has been proposed for 2016 in Brisbane.

Lucas Ferreira, ATR PhD student, supported by the Centre of Research Excellence Grant has undertaken to establish an Innovation and Communication Working Group to revamp the current Worldwide Twins website. The website aims to develop an online platform to facilitate the networking of global collaborations and promote research findings to the wider twin community.

[www.worldwidetwins.net](http://www.worldwidetwins.net)



# COMMUNICATIONS

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The ATR continued online initiatives such as the quarterly E-news to provide feedback on study findings and activities to ATR members and to researchers. The E-News plays an important role in raising awareness of the role of twins in research and translating this to the wider community.

In 2015, three E-news were sent to ATR members and four E-news were sent to researchers.

## IN PRESS – ARTICLES IN NEWSPAPERS, MAGAZINES, TV, RADIO AND OTHER MEDIA INCLUDING ONLINE

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In 2015 media coverage was achieved for the ATR and other substantial events. There were 67 press articles in newspapers, magazines, TV, radio and other media including online. Media coverage related to the Twins Plus Festival, the CPC Twin Research Project Node, zygoty, current research studies and the Registry itself.



# GENETIC TESTING DOUBLY IMPORTANT FOR TWINS



*Sue Sukkel with identical twins Lilly and Abbie.*

“Are they identical or not?” is usually the first question asked of new parents about their twins. While it may be a matter of curiosity for most parents, for others it has become lifesaving knowledge.

Mother of two-year-old twins, Kylie Tyrell, discovered the importance of knowing her twins’ genetic identity when one was diagnosed with a rare immune disorder. Kylie had only recently found out for sure her twins were identical and this led to the other twin, who wasn’t displaying any symptoms, to be tested and the discovery both twins had the disorder.

A joint study by the Australian Twin Registry and the Murdoch Children’s Research Institute involved twins who were uncertain of their zygosity and undertook a DNA test to find out for sure. It found that nearly one-third of the twin pairs were incorrect when asked to identify their zygosity prior to the test.

“We found a substantial proportion of parents and twins had been misinformed by their own parents or medical professionals,” according to the ATR’s Deputy Director, Associate Professor Dr Jeff Craig.

He says this knowledge can have implications for the bonding of twins, tissue compatibility in organ transplantation, assessing disease risk, the personal right to know your identity, legal and educational reasons, estimating the likelihood of the mother or close relatives giving birth to further sets of twins, and to avoid embarrassment when asked by family, friends and strangers.

Mother of eight-year-old twins, Sue Sukkel, participated in the study and has first-hand experience of the impact of twin identity confusion. She always believed her twins, Lilly and Abbie, were non-identical after being told so at their birth in hospital by the doctor and midwife. But with so many friends and family members convinced the twins were identical because they looked so similar, Sue decided on genetic testing.

“When the results arrived in the mail advising they were identical, I was so overwhelmed that I burst into tears,” Sue said. “I was so pleased that the girls will grow up with this knowledge. As an adopted child myself, I know what it is like to be unsure about your genetic heritage and how that can affect you.”

Associate Professor Craig said the confusion often arose due to wrong assumptions that identical twins always share a placenta in the womb and always look and behave identically. But nearly one-third of identical twins (as in the case of the Sukkel twins) and all same-sex non-identical twins have separate placentas.

According to Dr Craig, the only way to know for sure whether same-sex twins are identical or fraternal is to have a DNA fingerprint (zygosity) test done. Genetic testing is usually done by collecting a mailed kit with a cheek swab. The ATR provides this service at a special discounted rate of \$120 per twin pair – find details at the ATR’s website [www.twins.org.au](http://www.twins.org.au) “It is interesting how that initial wrong advice played out in our lives,” Sue Sukkel said. “I used to tell my family and friends, ‘you are crazy to think the girls are identical’, as I was so convinced they weren’t. The girls were teased and embarrassed at school because they would argue they weren’t identical and their class mates didn’t believe them.

“In the end, I think it is important to know the truth and to have certainty for the girls and our family. As they say, knowledge is power.”



# BUILDING CAPACITY

Develop projects and programs to build capacity in  
twin research in Australia.



## CHARLES PERKINS CENTRE TWIN RESEARCH PROJECT NODE

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*Jeffery Craig, Perminder Sachdev, Justine Gatt, John Hopper, Paulo Ferreira, Jan Hartvigsen and Brian Byrne*

On October 30th the ATR launched the Twin Research Project Node at the Charles Perkins Centre (CPC) at the University of Sydney. The Node will link directly to more than 60 other Project Nodes across CPC's research strategy and provide a resource for generating new research

opportunities. Over 100 researchers attended the event, generating much interest in possible new collaborations.

The launch comprised a series of presentations from seven NSW twin researchers and plenary speaker,

Jan Hartvigsen, Professor and Head of the Research Unit for Clinical Biomechanics at the University of Southern Denmark. A panel of twins also discussed their experiences with research.

## INTERNATIONAL RESEARCH AND RESEARCH TRAINING FUND

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The Brazilian Twin Registry (BTR) has been successfully established in collaboration with the Australian Twin Registry under support from the University of Melbourne's International Research and Research Training Fund grant. The BTR's website was created along with a

platform for database management. Recruitment in Brazil has started for the replication of the AUTBACK study. The Brazilian Twin Registry protocol was presented by Associate Professor Dr. Paulo Ferreira at the 4th International Network of Twin Registries in Osaka, Japan, aiming

at future international collaboration. The first study protocol has been submitted to the Low Back Pain Forum 2016. Lucas Ferreira's PhD project on international collaboration and knowledge translation, and implications for twin research and the BTR, is ongoing.



# VICTORIA PRIZE FOR SCIENCE AND INNOVATION



“ Professor Hopper has made seminal contributions to understanding the roles of genetic and environmental factors on Australia’s major cancers and other diseases

-Lily D'Ambrosia

The ATR’s Director, Professor John Hopper, has won Victoria’s top prize for scientific discoveries and technological innovations that improve lives.

A distinguished genetic epidemiologist and twin researcher, Professor Hopper received the Victorian Government’s prestigious Victoria Prize for Science and Innovation (Life Sciences Category). The award recognises the important role innovation plays in Victoria’s economic future and the need for people to be skilled in science, technology, engineering and mathematics.

“These are Victoria’s most prestigious awards for scientific discoveries and technological innovations

that advance knowledge, produce commercial outcomes and benefit our community,” said Victorian Minister for Industry, Lily D’Ambrosia, as she presented the award to Professor Hopper (picture above).

“Professor Hopper has made seminal contributions to understanding the roles of genetic and environmental factors on Australia’s major cancers and other diseases with significant clinical and population health significance,” Minister D’Ambrosia said.

In accepting the award, Professor Hopper joins a select group of 23 other leading Victorian scientists who have received the Victoria Prize since 1998.

In receiving the award, Professor Hopper stressed that “this is recognition of the many researchers, twins, and families who have contributed to research over more than 30 years of the ATR’s history.” “None of these achievements would have been possible without the generous donation of information and bio specimens from more than 100,000 Australians with the undertaking these would be used for research of global significance.”



*Liang-Dar (Daniel) Hwang*

"I would like to thank the Australian Twin Registry for awarding me the Australian Centre of Excellence in Twin Research Travel Grant Scheme that supported my attendance at the International Congress of Twin Studies (ICTS) satellite meeting, the Behavior Genetics Association (BGA) annual meeting and its pre-conference OpenMx workshop."

"Attendance at these two international meetings provided me with great learning and networking opportunities. It also allowed me to present my work and made my research more widely known by other researchers in the field. The OpenMx workshop equipped me with enough knowledge and confidence to move from class Mx to OpenMx. I cannot wait to start my next project using this new tool."

## RESEARCH TRAVEL GRANT SCHEME

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Grants for Round 14 of the Australian Centre of Excellence in Twin Research Travel Grant Scheme (ACETR) were awarded in September 2015. There were a large number of applicants who applied for funding to assist travel to national and international research conferences.

Congratulations to the following successful recipients:

- Dr Ali Ghasem-Zadeh, Department of Endocrinology, Department of Medicine, Austin Health
- Amabile Borges Dario, Physiotherapy Department, The University of Sydney
- Baptiste Couvy-Duchesne, Neuroimaging Genetics, Queensland Brain Institute, University of Queensland
- Dr Daniela Ribeiro, Orthodontic Unit, University of Adelaide
- Liang-Dar (Daniel) Hwang, Genetic Epidemiology Group, QIMR Berghofer Medical Research Institute
- Dr Lucia Colodro Conde, Quantitative Genetics, QIMR Berghofer Medical Research Institute
- Marina de Barros Pinheiro, Faculty of Health Sciences, University of Sydney
- Mihiri Silva, Vascular Biology, Murdoch Children's Research Institute
- Shuai Li, Centre for Epidemiology and Biostatistics, Melbourne School of Population and Global Health, The University of Melbourne
- Dr Yuk Jing (Jane) Loke, Early Life Epigenetics, Murdoch Children's Research Institute

## ATR DATA INDEX PROJECT

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The ATR is a collaborative development partner of The Ark, an open-source web-based tool that allows researchers to manage health and medical research data without specialised database skills or programming expertise. The system provides data management for core research information including demographic, phenotype, biospecimen and pedigree data. In addition it supports typical registry requirements such as tracking participant consent and correspondence, whilst also being able to generate custom data exports and reports. The Ark is a highly configurable system designed to provide the majority of data management functionality for a research registry or study.

# FBE-MDHS SEED PROJECT

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In 2015, the University of Melbourne's Faculty of Business and Economics and the Faculty of Medicine, Dentistry and Health Sciences provided seed funding (\$7,500) to encourage collaborative projects between the two faculties. Together, Jinhua Li (Melbourne Institute of Applied Economic and Social Research, Faculty of Business and Economics), Nicola Reavley (Centre for

Mental Health, Melbourne School of Population and Global Health) and Katrina Scurrah (Australian Twin Registry, Melbourne School of Population and Global Health) were successful in their application for a pilot study to test the feasibility of using a 'Children of Twins' study examining intergenerational transmission of mental health problems in Australia.

## FUNDING PRIORITIES FOR NEW INITIATIVES

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### Twin Pregnancy App

With around 9000 new parents of twins every year in Australia, the ATR saw the demand for a twin pregnancy mobile app. The app is designed to build on the success of the ATR's *Twin Pregnancy Booklet* but uses mobile technology, given our audience are young parents who engage regularly with digital devices and are time poor.

The app is being developed by the ATR, the University of Melbourne and Murdoch Children's Research Institute, in collaboration with the Australian Multiple Birth Association and will:

- Provide an evidence-based expert guide to twin pregnancy, birth and early parenting of twins

- Translate relevant research findings and, in time, to conduct research
- Serve as a central reference, trusted resource and networking hub connecting twin families

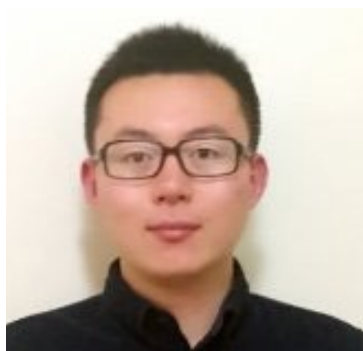
We are now seeking funding for the development of this app. To find out more or to make a contribution, please contact Lynette Walker: [lynettew@unimelb.edu.au](mailto:lynettew@unimelb.edu.au) or refer to our website: <http://www.twins.org.au/contact>.



## PhD students

To build capacity, the ATR is developing the twin research workforce by supervising and supporting PhD students. The ATR currently hosts two students and is looking to host another.

Our current PhD students are:



### Shuai Li

Shuai Li is investigating DNA methylation based on the Australian Mammographic Density Twins and Sisters Study. His interests include the genetic and/or environmental determinants for DNA methylation, lifestyle behaviour's influence on DNA methylation, DNA methylation and breast cancer.

So far, Shuai has been the recipient of five awards during his time with the ATR at the University of Melbourne:

- The Roger Williams Memorial Award for the Best Presentation at the 2015 Annual Meeting of the International Genetic Epidemiology Society
- International Postgraduate Research Scholarship
- Australian Postgraduate Award (International)
- The Richard Lovell Traveling Scholarship
- Graduate Research Studentship

Shuai is originally from China and completed a Master's in Epidemiology at Peking University prior to commencing his PhD.



### Lucas Ferreira

Lucas Ferreira's project aims to understand the challenges of twin research in the context of global scientific collaboration and their implications for the development of the Brazilian Twin Registry. It will use a mixture of qualitative research methods to address these aims, including interviews with researchers within the International Network of Twin Registries.

Lucas is originally from Brazil and completed a Master's in Technology and Innovation Management at the University of Queensland.

He is the recipient of a CAPES Scholarship, Ministry of Education, Brazil.

To find out more or to make a contribution, please contact Kate Murphy: [murphyks@unimelb.edu.au](mailto:murphyks@unimelb.edu.au) or refer to the contact details on our website: <http://www.twins.org.au/contact>.



# HOW GIFTS IN YOUR WILL ARE MAKING A DIFFERENCE



*A life-threatening illness led this twin to consider how she could make a lasting contribution to finding cures to help others.*

We thank twin and ATR member, Meryl (as she wishes to remain anonymous, we have not used her full name), for her bequest to the ATR and for telling her story to encourage other twins to consider making a bequest to twin research.

## Meryl's story:

"My twin sister and I joined the ATR over 20 years ago and have been more than happy to participate in the ATR's research when called on.

"I have recently returned to Australia after living in England since 1995. In 2003 I was diagnosed with Non Hodgkinson Lymphoma. The treatment I received over 11 years led to the full remission of the condition in 2014.

"Without the advanced medical technologies that I was able to benefit from, I might have not survived. During the years of treatment I have been acutely aware that

these treatments were the result of intensive painstaking research by many research organisations. Whilst much of the research would have been funded by governments and the pharmaceutical industry, I am also aware that donations and bequests by generous people have also helped fund the research.

"My bequest to the ATR is my way of thanking everybody involved in medical research for the gift of good health that I received and helping to fund further research programs."

A bequest to the ATR will help us to continue contributing to the forefront of health and medical research for generations to come. Twin research has the power to transform our understanding of disease and to

contribute to cures to benefit all of humankind.

Meryl is a twin and ATR member who suffered a life-threatening illness and chose to leave a bequest to the ATR as a lasting contribution to finding cures and helping others. Your gift through a bequest can also make a very real difference. If you would like to confidentially discuss including the ATR in your Will, you can phone Kate Murphy on 03 9035 4625 or email her at [murphyks@unimelb.edu.au](mailto:murphyks@unimelb.edu.au). Kate can discuss with you, the ways in which you can leave a bequest to us and what your bequest will achieve. Further information is also available at: <http://www.twins.org.au/support-us/bequests>.





# GOVERNANCE

Apply governance of the ATR in a fair, transparent and equitable manner.

# ATR MANAGEMENT

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The ATR's leadership team is led by 7 Chief Investigators from institutes around Australia, supported by a management team and Associate Investigators and overseen by an Advisory Board.

## Chief Investigators:

- Professor John Hopper, Director of the Australian Twin Registry, University of Melbourne
- Associate Professor Jeffrey Craig, Deputy Director of the Australian Twin Registry, Murdoch Children's Research Institute
- Professor David Mackey, University of Western Australia
- Professor Stephen Simpson, University of Sydney
- Professor Brian Byrne, The University of New England
- Dr Paulo Ferreira, University of Sydney
- Ms Susan Carrick, CPC, University of Sydney

- A/Prof Jeff Craig, Deputy Director ATR, Group Leader, Early Life Epigenetics at the Murdoch Children's Research Institute
- Kate Murphy, ATR Manager
- Jenny Boadle/ Janine Lam, ATR Study Coordination & Liaison
- Shaie O'Brien, ATR Project Support Officer

Any member of ATR Management with a potential conflict of interest is required to declare this interest prior to any relevant discussions. Persons with a conflict of interest in any study are excluded from review or application approval processes of that study. Members of the Advisory Committee are available to help act as independent reviewers. In the event that the Director or Deputy Director is involved in a study as a researcher, they take no part in the approval process. If both are involved or unavailable, an independent person is brought in to oversee the processing of the application

As of 31 December 2015, the ATR Management comprised:

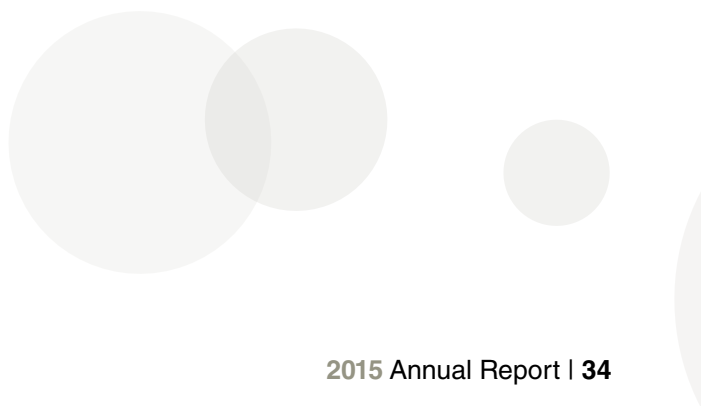
- Prof John Hopper, AM, Director, ATR; NHMRC Senior Principal Research Fellow; Director (Research), Centre for MEGA Epidemiology, University of Melbourne

# ADVISORY BOARD AND CHARTER

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In 2015, the Advisory Board members were:

- Mr Vincent Pollaers, Chair, Advisory Board
- Dr Paul Jelfs (Australian Bureau of Statistics, Australian Capital Territory)
- A/Professor Paul Lancaster (University of Sydney, Honorary Associate Professor)
- Prof Margaret Otlowski (University of Tasmania, Tasmania)
- Mrs Karen Willetts (AMBA Representative, New South Wales)
- Ms Sue Carrick (Twin Representative, New South Wales)





## Vince Pollaers (Chair)

Vincent Pollaers has been Chairman of the Australian Twin Registry since 2006. In 2010, he was also awarded an honorary fellowship in the School of Population and Global Health at the University of Melbourne for his contribution to medical and scientific research. He currently holds the position of General Counsel and Company Secretary of Greencross Limited, a leading ASX200 pet care company.

Past career highlights include: Group Human Resources Manager, General Counsel and Business Development Executive – Services, Mammoth Pet Holdings Pty Ltd; a corporate lawyer with Freshfields, London; General Counsel, IBM Australia/NZ; Managing Director, McKinney Rogers Asia Pacific.



## Dr Paul Jelfs

Dr Paul Jelfs (PhD) is the General Manager of the Population and Social Statistics Division at the Australian Bureau of Statistics. Paul has extensive experience in Commonwealth and State Government agencies in both information management and service delivery. He has degrees in Applied Economic Geography (UNSW) and a PhD in Epidemiology (UNSW).

On a personal level, his blended family includes two sets of twin girls aged 20 and 31. All four twins are members of the ATR and have participated in twin studies.



## Dr Paul Lancaster

Paul Lancaster is Honorary Associate Professor at the Menzies Centre for Health Policy, Charles Perkins Centre, University of Sydney. He trained as a paediatrician at UNSW teaching hospitals; then was appointed Staff Paediatrician and Director of Newborn Services at the Royal Hospital for Women in Paddington. In 1979 he was appointed founding Director of the National Perinatal Statistics Unit, School of Public Health, University of Sydney.

He has been a member of the Advisory Board of the Australian Twin Registry at the University of Melbourne for 20 years.





## Professor Margaret Otlowski

Professor Otlowski is the Dean and Head of School of the University of Tasmania's Faculty of Law, with particular interest in the area of health law and ethics. Her research involvements include her work as Chair of the University Human Research Ethics Committee, Deputy Director for the Centre of Law and Genetics, and a member of the Royal Hobart Hospital's Clinical Ethics Committee.

She has served as member of two of the National Health and Medical Research Council's principal committees for two successive trienniums (2009-2012 and 2012-2015): the Human Genetics Advisory Committee and the Australian Health Ethics Committee. She has been appointed as Fellow of the Australian Academy of Law.



## Karen Willetts

Karen's role as a mother of (now 14-year-old) twin girls and vast experience at a local, state and national level within the Australian Multiple Birth Association (AMBA), means she is ideally suited to her position on the ATR's Advisory Board as a 'voice' to address the needs of the families of multiples. She continues to play an active role within the organisation and brings a wealth of experience and knowledge to the ATR.



## Sue Carrick

Sue Carrick leads Susan Carrick and Associates as a Special Adviser, Research Strategy and Partnerships to leading Australian research foundations.

Career highlights include roles with the National Breast Cancer Foundation as Director Research Investment, and General Manager Register4; management positions with the NHMRC Clinical Trials Centre, University of Sydney; and a lecturer at the Faculty of Nursing University of Sydney.

She is one of a male/female twin pair.

## ATR TEAM



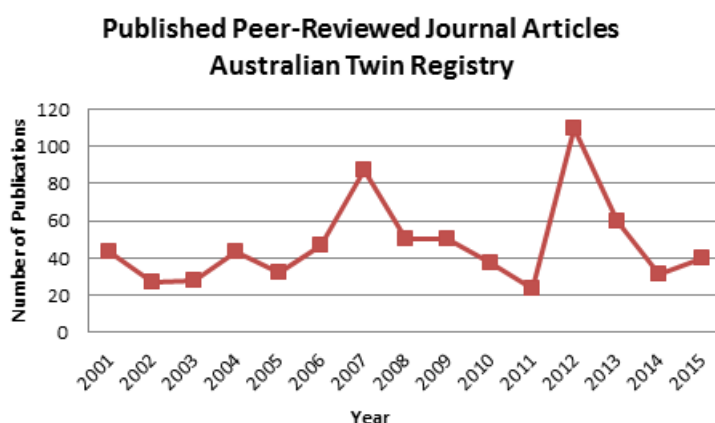
The ATR is administered by the University of Melbourne and is situated in the Centre for Epidemiology and Biostatistics, in the Melbourne School of Population and Global Health. In 2014, the ATR team was made up of a Director, Deputy Director, ATR Manager, ATR Study Coordinator, a Marketing Officer, Project Support Officer,

graphic designer, two administration assistants, two phone staff, a research assistant, one PhD student and a part-time Database Manager.

The ATR provided an honorarium to the part-time Deputy Director.

## PUBLICATIONS

An important measure of the output of the ATR is the number of publications arising from studies supported by the facility. In 2015, the ATR recorded 40 peer-reviewed journal articles (Figure 16), one book, and eight conference proceedings, for a total of 49 publications. There have been more than 700 publications in the last 15 years. The ATR's CRE goal is to increase annual twin publications by 20 percent. The list of all 2015 publications can be found in Appendix 1.



**Figure 16: Published Peer-Reviewed Journal Articles  
Australian Twin Registry**

# DONATIONS

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The ATR welcomes donations towards the administration and management of the Registry. Donors are provided with a receipt. Donations may be earmarked for a specific activity. We are very grateful for all the support we receive from Registry members and the wider community who have donated in 2015 towards a total of \$3,875.

The ATR is deeply grateful for the generosity of all its donors who have supported our initiatives throughout the year.

You can help make a difference by contributing online, by mail or phone. More information is available at:  
<http://twins.org.au/support-us>.



# APPENDIX

## APPENDIX

# APPENDIX 1

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## ATR 2015 PUBLICATIONS LIST

### BOOKS

Townsend, G., Pinkerton, S., Rogers, J., Bockmann, M. & Hughes, T. (2015 ). Twin Studies: Research in Genes, Teeth and Faces. University of Adelaide Press.

### CONFERENCE PUBLICATIONS

Bjørnerem, A., Bui, M., Wang, X., Ghasem-Zadeh, A., Hopper, J., Zebaze, R., & Seeman, E. (2015 ). Does Cortical Bone Loss Precede Menopause? 2015 ASBMR Annual Meeting, USA Plenary Sessions. Seattle, Washington.

Gatt, J. (2015). Understanding And Promoting Resilience: A Neuroscience Perspective. Psychiatry Professional Grand Rounds. Westmead Hospital, Sydney.

Gatt, J. (2015). Using Twin Studies To Understand The Genetics And Neuroscience Of Resilience And Wellbeing. ATR Twin Node Launch. Charles Perkins Centre, University of Sydney.

Li, S. (2015). The Variation Of DNA Methylation At Vast The Majority Of CpG Sites Are Due To Individual Factors But Not Genetic Or Shared Environment Factors. The 24th Annual Meeting of the International Genetic Epidemiology Society. Baltimore, USA.

Li, S., Wong, M., Apicella, C., Stone, J., Dite, G., Joo, J., Giles, G., Southey, M., & Hopper, J. (2015). Familial Correlation In DNA Methylation Is More Due To Sharing The Womb Than Sharing Genes: A Twins And Sisters Study. The 6th Biannual Australian Epigenetics Alliance Flagship Conference. Hobart, Australia.

Li, S., Wong, M., Apicella, C., Stone, J., Dite, G., Joo, J., Giles, G., Southey, M., & Hopper, J. (2015). Is Familial Correlation In DNA Methylation Due To Sharing The Womb At The Same Time Or Sharing Genes? A Twins And Sisters Study. GeneMappers 2015. Perth, Australia.

Li, S., Wong, M., Apicella, C., Stone, J., Dite, G., Joo, J., Giles, G., Southey, M., & Hopper, J. (2015). Is Familial Correlation In Genome-Wide Methylation Due To Sharing The Womb Or Genes. American Society of Human Genetics Annual Meeting 2015. Baltimore, USA.

Mackey, D. (2015). Twins – Heritability and Endophenotypes Helping Drive Gene Discovery in Eye Disease. Healthier Kids: Insights from twin research. Melbourne, Australia.

### JOURNAL ARTICLES

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Boraxbekk, C., Ames, D., & Kochan, N., et al. (2015). "Investigating The Influence Of KIBRA and CLSTN2 Genetic Polymorphisms On Cross-Sectional And Longitudinal Measures Of Memory Performance And Hippocampal Volume In Older Individuals." Neuropsychologia, **78**(10-17).

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Craig, J., Segal, N., Umstad, M., Cutler, T., Keogh, L., Hopper, J., Rankin, M., Denton, J., Derom, C., Sumathipala, A., & Harris, J. (2015). "Zygosity Testing Should Be Encouraged For All Same-Sex Twins." BJOG: An International Journal of Obstetrics and Gynaecology, **122**(12): 1573–1719, e1571–e151.

Cutler, T., Murphy, K., Hopper, J., Keogh, L., Dai, Y., & Craig, J. (2015). "Why Accurate Knowledge Of Zygosity Is Important to Twins". Twin Research and Human Genetics, **18**(3): 298-305.

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