



2014 ANNUAL REPORT

1 January - 31 December 2014



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Discoveries for a
healthier community



CONTENTS

HIGHLIGHTS AND ACHIEVEMENTS	3	PARTICIPATION	26
		<i>Study Approaches</i>	27
		<i>Telephone Follow Up</i>	28
		<i>Record Updates</i>	29
		<i>Adverse Effects and Complaints</i>	31
ATR OVERVIEW	4	VALUE-ADD	32
<i>Outlook for 2015</i>	5	<i>ATR Data Index Project</i>	34
<i>About the Australian Twin Registry</i>	6	<i>The Ark</i>	34
<i>Potential for Twin Research</i>	6		
<i>Values</i>	7	GOVERNANCE	36
		<i>ATR Management</i>	37
THE REGISTRY	8	<i>Advisory Board and Charter</i>	37
<i>Database</i>	9	<i>ATR Staff</i>	40
<i>Membership</i>	9	<i>Dispute Resolution Process</i>	41
<i>Recruitment</i>	12	<i>ATR Budget</i>	41
<i>Currency and Accuracy of Membership Data</i>	15	<i>Donations</i>	41
<i>Media Activities</i>	15		
<i>In Press – articles in Newspapers, Magazines, TV, Radio and other media including online</i>	15	Appendix 1: Researcher Reports (2014)	42
<i>Twin Pregnancy Booklet</i>	17	Appendix 2: In Press Publications List (2014)	49
SCIENTIFIC MERIT	18	Appendix 3: Publications List (2014)	50
<i>Current Research Studies</i>	19	Appendix 4: Response Rates (2014)	55
<i>Researcher Reports</i>	22		
<i>Publications</i>	23		
<i>Meetings and Conferences</i>	23		
<i>Research Travel Grant Scheme</i>	24		

2014/15 highlights



Membership in 2014

ATR membership grew to 43,797
twin pairs and 429 triplet sets

1,103 new twin & triplet sets

14,404 twins participated
in studies



Australian Centre for Excellence in Twin Research

The Federal Government announced the country's first ACE in Twin Research which will provide ongoing support for the Australian Twin Registry. This will position the ATR to generate transformative research to benefit the lives of twins and all Australians.



Research

12 twin studies supported
by the ATR

Our research covers a broad spectrum of health issues — diabetes, breast cancer, speech and language disorders, obesity and addiction

Healthier Kids: Insights from Twin Research
Conference attracted 200 researchers
and twins



Twins Festival

Twins research, support services and entertainers featured at this year's festival attended by 2000 twins, triplets and their families and friends from around Australia and the world.



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WELCOME TO THE 2014 ANNUAL REPORT OF THE AUSTRALIAN TWIN REGISTRY (ATR)

This report summarises the Australian Twin Registry's major achievements, activities and research developments during 2014.

The 2014 reporting period coincides with the fifth and final year of the renewed Enabling Grant (2010 – 2014) from the National Health and Medical Research Council (NHMRC) that has funded the ATR for the last five years.

Highlights and Achievements Summary 2014

- » The ATR was successful in its application for a Centre of Research Excellence (CRE) grant from the National Health and Medical Research Council (NHMRC) for the period 2015-2020. This will enable the ATR to drive multidisciplinary collaboration across the research community to further empower twin research in Australia and globally, and address health challenges for the whole population
- » The ATR applied and was successful for the International Research and Research Training Fund (IRRTF) to work with Dr Paul Ferreira, University of Sydney, and his collaborators in Brazil to conduct a twin study of lower back pain and to help the establishment of a Brazilian Twin Registry.
- » ATR Membership numbers grew, again exceeding the goal set in the Enabling Grant, with 1,103 new sets of twins and triplets registered in 2014.
- » The ATR recruited for 9 studies in 2014; with 2 new studies looking at environmental factors and how they interact with genes to predict NAPLAN results and the genetic basis of singing ability. Three new de-identified data studies were possible with the release of the Health and Lifestyle Questionnaire.
- » The ATR enabled the publishing of 31 peer-reviewed articles based on ATR-related studies, as well as 2 book chapters and 17 conference abstracts and posters.
- » The *Healthier Kids Conference: Insights from Twin Research* was held at the Royal Children's Hospital in Melbourne on Friday, December 5th and was a great success. The conference had a full itinerary of 23 presenters and over 250 attended, including 95 delegates (30 twins).
- » The Travel Grant Scheme supported 19 researchers to attend at national and international research workshops and conferences relating to twin research.
- » 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:
 - » Developing the ATR Health and Lifestyle Questionnaire
 - » 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
 - » Continuing to lead the International Network of Twin Registries.

2014 has been a very successful year for the ATR in research, funding and Registry growth. The outlook for 2015 and beyond is promising, with expansion and development in all these areas.

Keynote speaker, Nancy Segal and John Hopper, ATR Director with twin delegates at the Healthier Kids Conference, December 2014



Outlook for 2015

Moving forward The ATR will be supported by the new funding structure of the Australian Centre of Excellence in Twin Research (ACETR).

This new phase will ensure that we are the drivers of new and innovative twin research by collaborating with leading researchers within Australia and internationally. We will 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:

- » Generating knowledge in new initiatives such as establishing a twin perinatal study
- » Strengthen our collaborations with key institutes and researcher teams such as the Charles Perkins Institute at the University of Sydney
- » Creating a twins and families data repository by implementing the collection, harmonisation and archiving of research data from within the ATR and from external twin researchers
- » Supporting the establishment of the Brazilian Twin Registry
- » Publishing on data from the Health and

Lifestyle Questionnaire

- » Ensuring our experience and expertise in data and statistical knowledge is promoted
- » Supporting and supervising PhD students to conduct twin research

This will be achieved in the **twin community** by:

- » Building on our representation of the twin community's needs at conferences and seminars
- » Establishing a formal twin community forum, to seek input on research topics, research outcomes and ATR activities
- » Providing discounted zygosity testing to ATR members
- » Providing the opportunity to engage with researchers to conduct studies at the Twins Plus Festival in March 2015

This new phase will ensure, through the ACETR, that the Australian research community has efficient and reliable access to the benefits available from the utilisation of twins in health and medical research.



About the Australian Twin Registry

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 2014, the ATR maintained information on 40,704 sets of twins and triplets, and supported almost 60 research projects in application, active recruiting, data collection or writing phase, covering a broad spectrum of health-related themes (see **Researcher Reports** in Appendix 1).

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 2014 the ATR made significant progress towards achieving this vision.

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 newly 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.

Values

The following values guide the ATR in achieving its core functions:

Respect: The ATR conducts its operations with the fullest respect for the volunteerism of the twins and their relatives in their registration and participation; for the ATR staff in monitoring and maintaining the use of this resource; and, for the researchers in their efforts to conduct timely and relevant studies in accordance with their commitments to their funding bodies, made with the agreement of the ATR.

Leadership: The ATR will maintain and expand its role as an independent facilitator of twin studies, in training and informing researchers about the potential, design, conduct and analysis of twin studies, and in providing information about issues of relevance to twins and their relatives.

Equity of Access: The ATR undertakes its functions under the principles of equity of access by researchers irrespective of factors, such as institution, discipline, and relationship to ATR, and equity of participation of twins eligible for particular studies and activities.

Privacy and Confidentiality: The ATR holds information on registered twins in the strictest confidence and in accordance with Australian legislative requirements.

Consumer Participation: The ATR engages in and conducts activities with twins and parents of twins whenever appropriate, whether or not they are members of the ATR.

Excellence in Research: The ATR strives to enable researchers to achieve excellence in their research.

The ATR's core functions are:

■ Core function 1: 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.

■ Core function 2: Scientific Merit

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 3: Participation

Use judicious management and administration of approach to 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 4: Value Add

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

■ Core function 5: Governance

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





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.



Sophie and Amanda Fontaine are registered twins who spent 5 days at the ATR for student work experience. During their time here they wrote the following piece:

"Our mum and dad registered us in the Australian Twin Registry when we were just 10-months-old. Mum discovered the ATR through AMBA (the Australian Multiple Birth Association). As we have grown older, we have enjoyed being involved in various ATR studies.

We feel it is important for twins to be acknowledged as unique individuals, as well as being able to celebrate their extraordinary relationship.

The ATR has given us many opportunities to commemorate our twinship and provided material which is both thought-provoking and fascinating. The Registry is a well organised team of dedicated people who share their passion for working with twins and their families. Not only can we give back to our community by participating in studies, but we can also develop and grow as individuals whilst gaining more of an understanding of how science plays a huge part in the lives of twins."

Thanks to Sophie and Amanda Fontaine, 16, from Gippsland, Victoria who have so far participated in eight studies since joining the ATR as babies.

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.

In 2014, the database held data on 88,452 individuals representing 43,797 twin pairs and 429 HOM sets.

In 2014, 11,410 twin and other contacts (i.e. parent or secondary carer) records were updated. There were 27,924 study approaches to ATR members to participate in research. The ATR maintained accurate information on 92% of its members.

Members of the ATR are recorded under a specific status, depending on the currency of their contact details and individual preference for involvement in research activities. The members enrolled in the period covered by this report (73%) are categorised as Active/Active, Active/Questionnaire and Questionnaire/Questionnaire pairs, indicating that they are willing to consider participating in research. The current status of members of the ATR is summarised in Table 1.

Twin Pair Registrations: A total of 72% twin pairs have both members active, and an additional 9% of members' contact details require updating (recorded as Pending). Junior members represent approximately (32%) of the entire Registry, the remaining (68%) being senior members.

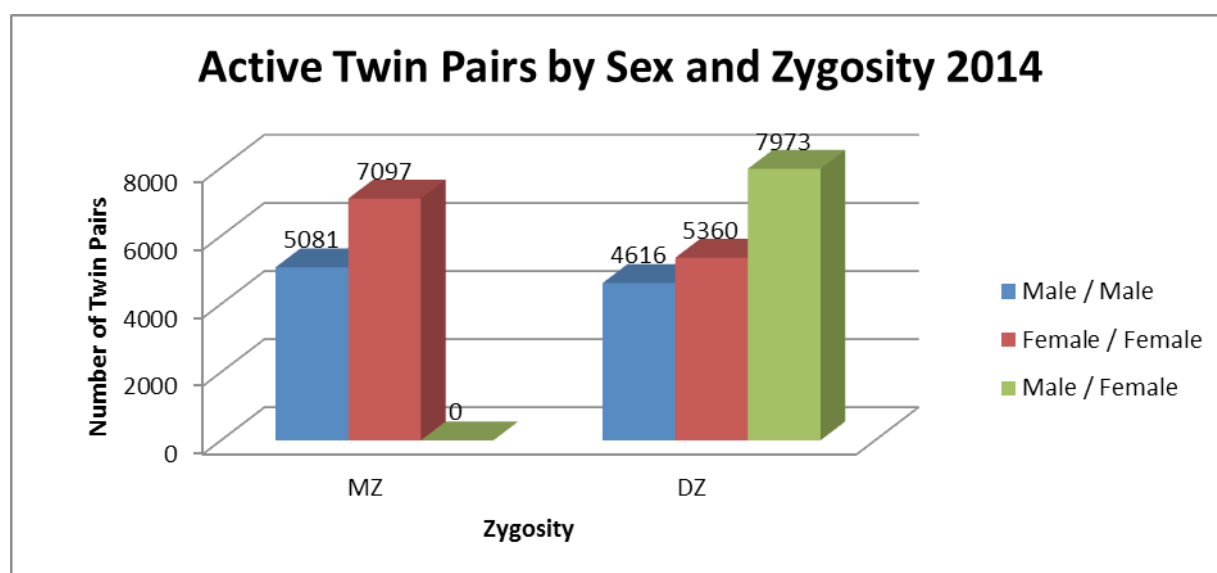
*Top row details T1 (twin one) status, and left most column details T2 (twin two) status. **O/S** identifies twin members who have moved overseas, but are still available for electronic surveys; **Lost O/S** refers to members for whom the Registry has an unconfirmed overseas address, and exhausted all avenues for obtaining up-to-date contact details. **Hold** denotes twins who cannot receive communications from us for a period of time due to illness or holiday etc. **Pending** denotes members for whom we do not have current details; so they are 'pending' an update.*

Table 1.

T1/T2 Status	Active	Deceased	Lost	Lost (O/S)	Inactive	Questionnaire	Newsletter	O/S Temp	Pending	Total
Active	31,349	420	78	3	479	341	100	88	1,387	34,245
Deceased	385	592	9	0	230	7	24	1	54	1,302
Lost	82	7	755	1	14	1	1	0	21	882
Lost (O/S)	4	0	1	12	2	0	0	0	2	21
Newsletter	83	27	4	1	22	1	104	1	16	259
Inactive	429	266	15	0	1,573	3	12	4	88	2,390
Hold	90	1	0	0	2	1	1	60	11	166
Questionnaire	307	5	0	0	4	144	2	2	25	489
Pending	1,392	63	31	0	81	35	17	10	2,412	4,041
	34,121	1,381	893	17	2,407	533	261	166	4,016	43,795

The current numbers of active twin pairs by sex and zygosity are shown in **Figure 1**. It does not show the 'lost' or 'unknown' twin pairs of which there are 1,222 pairs.

Figure 1. Active Twin Pairs by Sex and Zygosity 2014.



The distribution of active twin pairs by location is shown in **Figure 2**, together with the overall distribution of Australian population by State and Territory (as reported by the Australian Bureau of Statistics in 2014). Comparison of the two graphs shows that most populated states, New South Wales, Victoria, Queensland and Western Australia are the same ones where most active ATR members reside.

Figure 2. Australian Population by state 2014

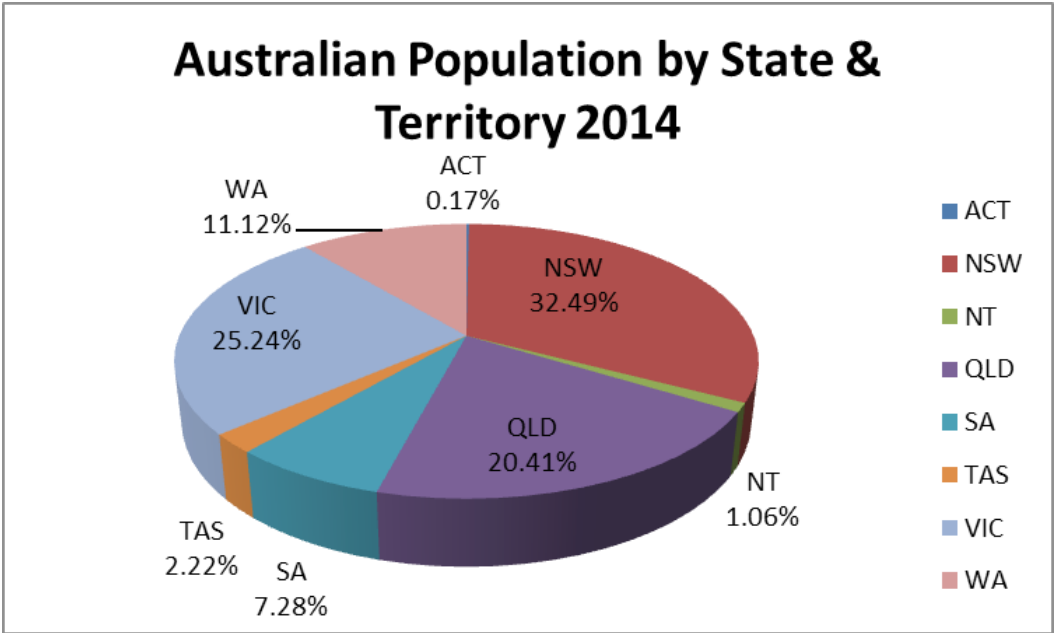
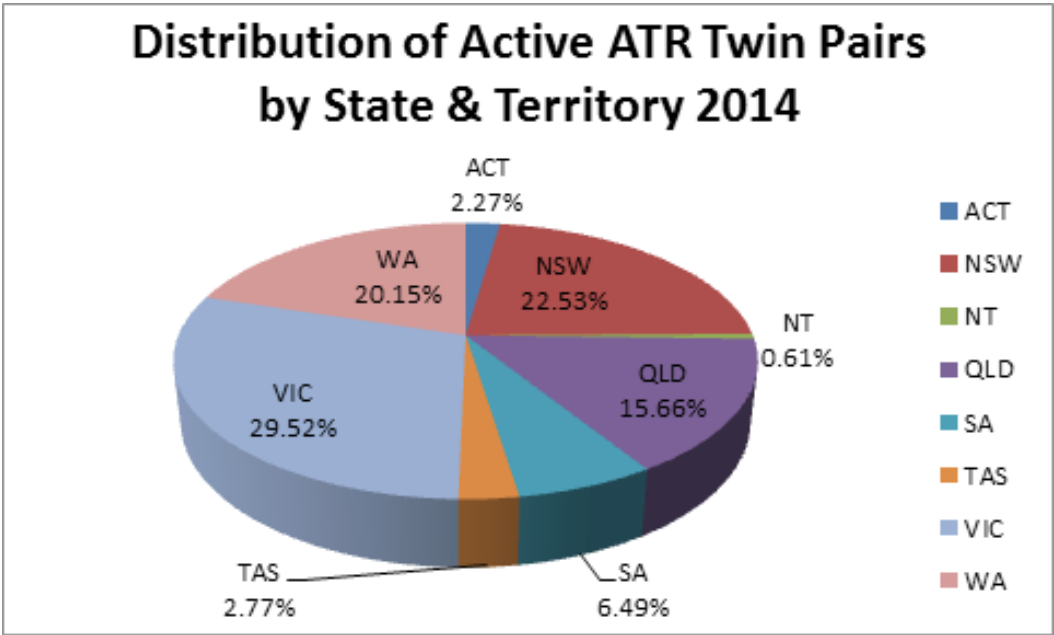


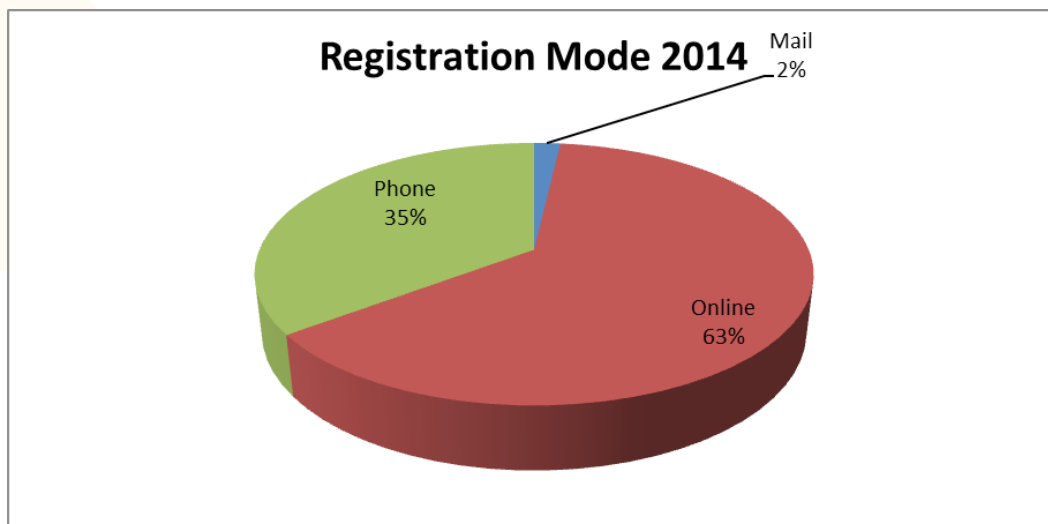
Figure 3. Active Twin Pairs by State 2014. *Note: because some twins live in separate states or one twin member in a pair lives overseas, this excludes twins overseas, or with no address available.*



Recruitment

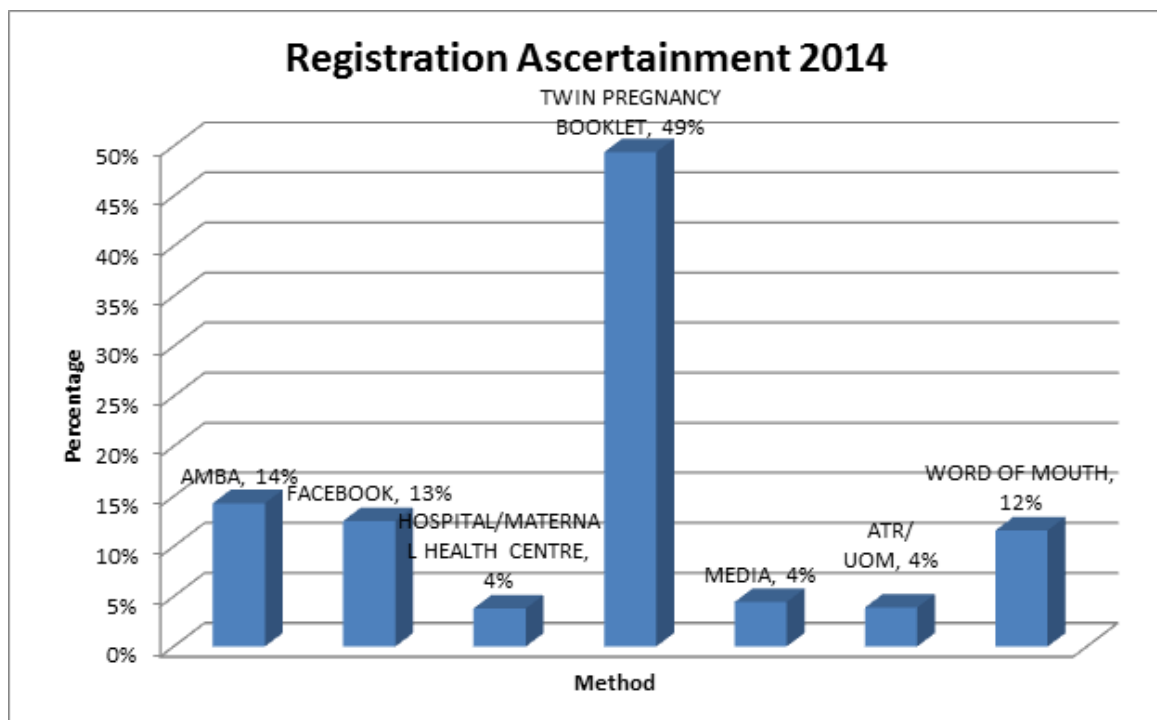
Continuous recruitment of new members is vital to ensure the future viability of the ATR. The ATR's goal is to increase membership by more than 2,500 twin pairs over 5 years. In the reporting period 1 January 2010 to 31 December 2014 this has been exceeded with 9,217 sets (twin pairs and higher order multiples (homs)) being added. In 2014, the ATR recruited a total of 1,103 twin and hom sets. The internet is by far the dominant means of registration (63%), (Figure 4), 35% by phone and 2% via mail. The internet is an ever-increasing influence, with online registrations having increased by 20% compared to last year.

Figure 4. Mode of new registrations (twin and triplet sets) in 2014.



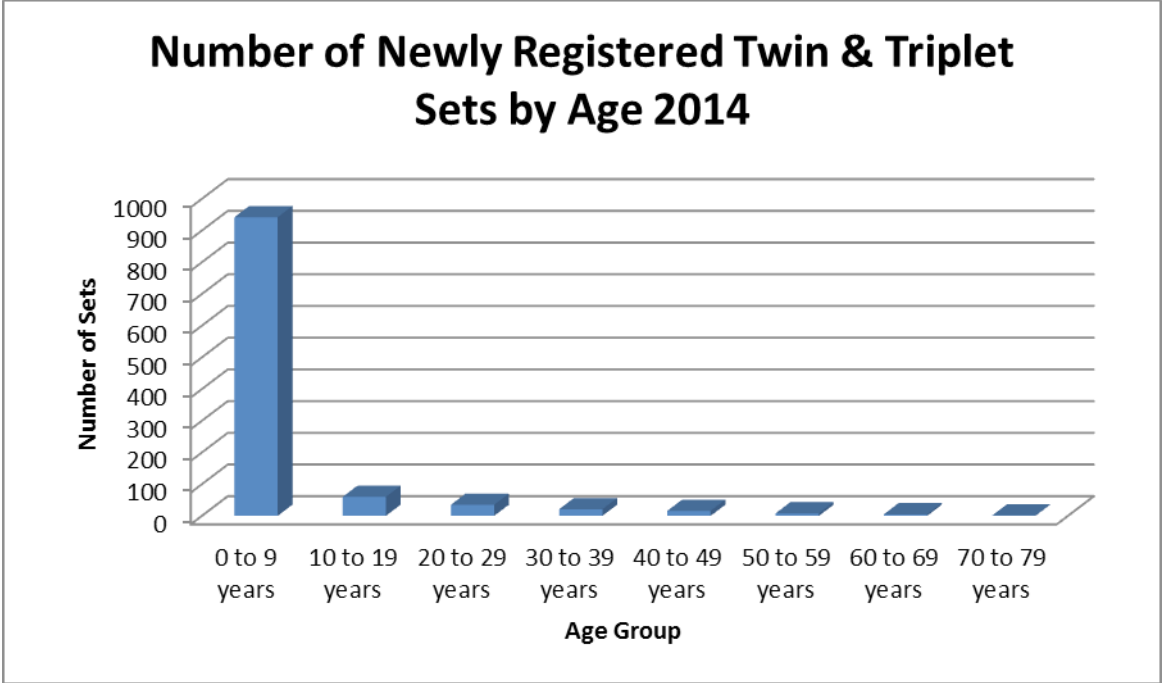
New registrations come through a range of avenues. The most effective means is the request for the free Twin Pregnancy Booklet which enables the ATR to approach parents of multiples to join the ATR. The second main recruitment channel is through promotion within AMBA (14%).

Figure 5. New registrations by Ascertainment (twin and triplet sets) in 2014.



Every effort is made to ascertain where new registrations to the ATR are from, as this informs where the ATR should focus future promotion. Other registrations come through the Australian Multiple Birth Association (AMBA) (14%), word of mouth (12%), internet/ Facebook (13%), hospital and maternal health centres (4%), media (4%) and ATR/University of Melbourne (4%).

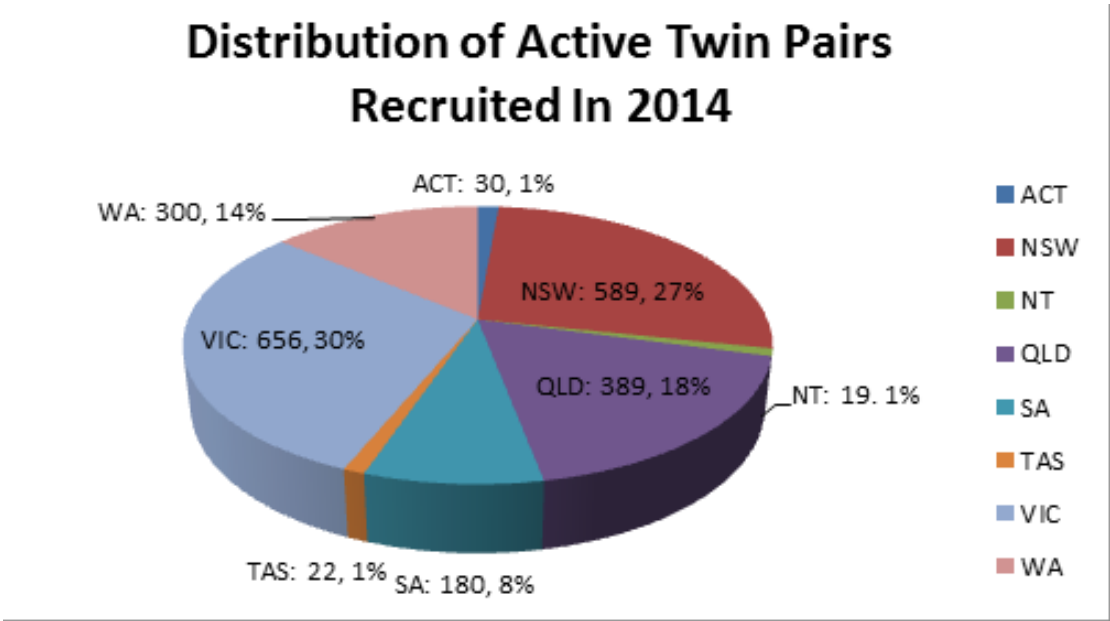
Figure 6. Number of new active twin and triplet sets registered in 2014 shown by age range.



The majority of new members (85%) enrolled with the ATR during the reporting period were aged 0-9 years (**Figure 6**). This is consistent with the previous 20 years.

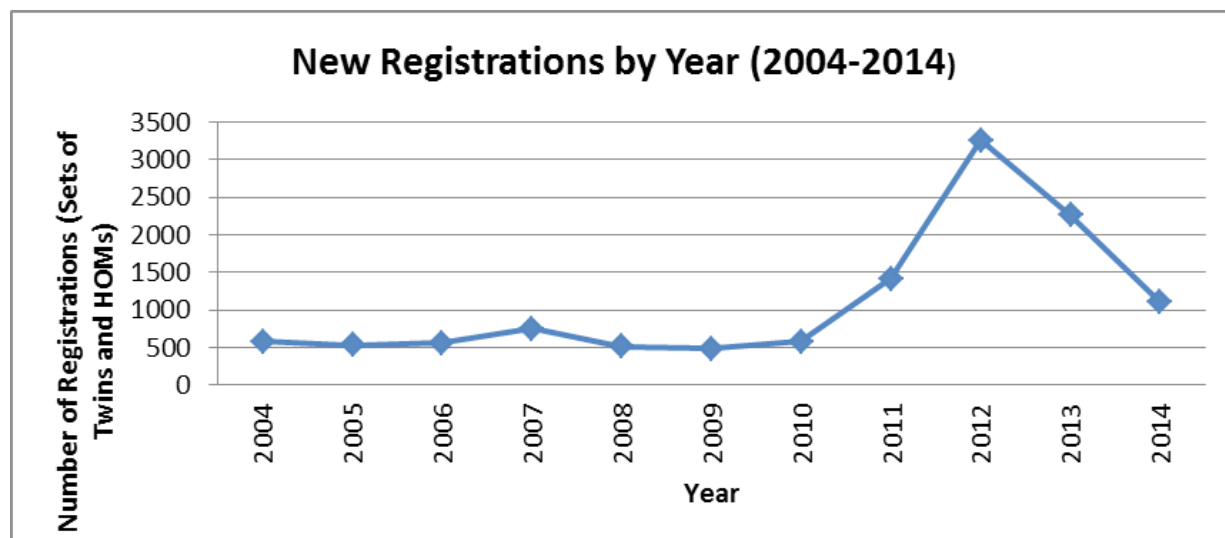
Most new members came from Victoria (30%) followed by New South Wales (27%), Queensland (18%), and Western Australia (14%) (**Figure 7**).

Figure 7. Distribution of active twin pairs, by State or Territory, recruited during the reporting period in 2014. *Note: numbers of triplet sets are not included in the graph.*



The numbers of new registrations by year since 2004 are represented in **Figure 8**. The peak in registrations in 2012 and 2013 was due to a larger proportion of new registrations from the Western Australian Twin Registry in 2012.

Figure 8. Number of new twin and HOM sets registered with ATR by year since 2004.

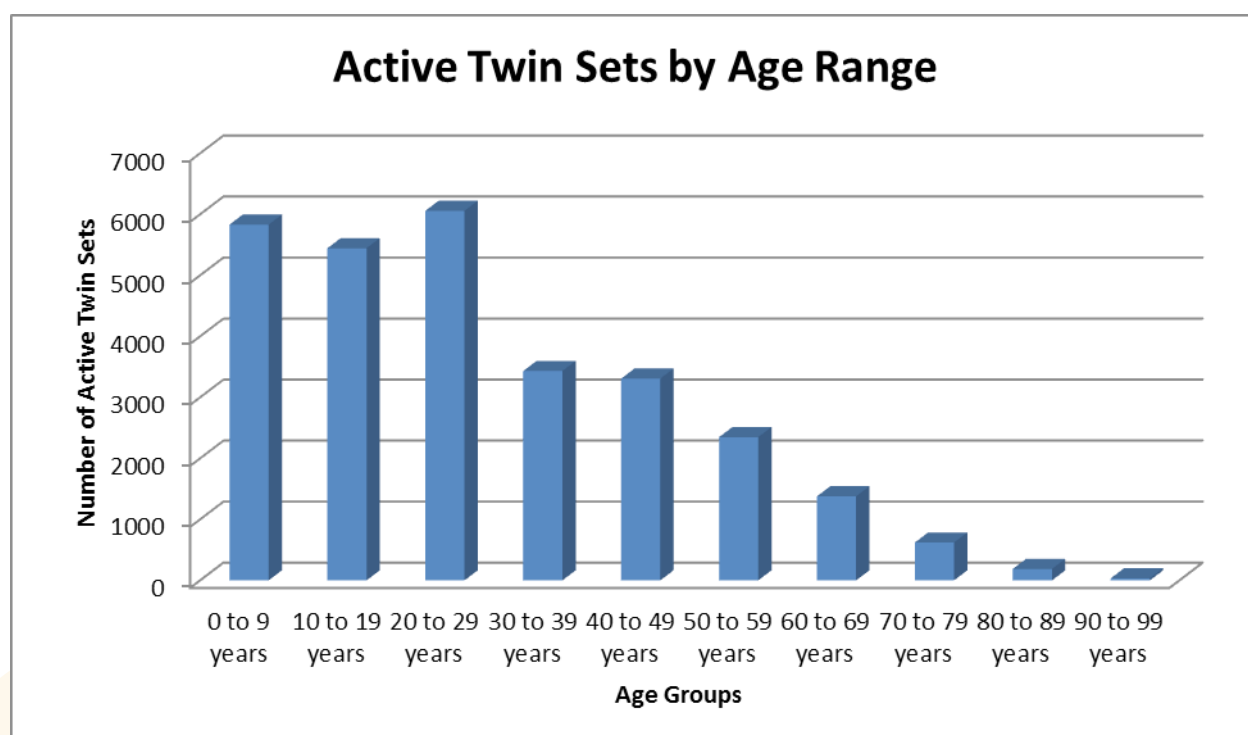


**2012 peak: Merge of Western Australian Twin Registry with Australian Twin Registry*

The distribution of active pairs of twins, across the Registry, is shown relative to the age groups of members in **Figure 9**. The addition of WATR members to the Registry is responsible for the 20-29 year group spike. Future years see the first three categories 0-9 years; 10-19 years and 20-29 years, as the three most consistent age groupings.

Figure 9. Active twin pairs shown by age group, as at publication.

Note: Data on triplets are not included in the graph.



Currency and Accuracy of Membership Contact Details

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-Newsletter; following up return to sender mailed communication through study approaches, the printed newsletter and via phone calls to the twins' or 2nd and 3rd contacts.

Media Activities

The ATR continued initiatives online such as the quarterly E-news to provide feedback on study findings and activities to ATR members and to researchers. In 2014, the ATR aimed to raise awareness of twin's role in research and translating this to the wider community.

In 2014, 3 E-newsletters were sent to ATR members, 4 E-newsletters were sent to researchers.

In Press – articles in Newspapers, Magazines, TV, Radio and other media including online

In 2014 media coverage was achieved for ATR and other substantial events: 31 press articles in newspapers, magazines, TV, radio and other media including online. The media coverage relates to the Singing Ability study, the Fat Taste Sensitivity study, the Healthier Kids Conference, the experience of being a twin and the Registry itself. Refer to [Appendix 2](#) – In Press Articles for all of the ATR media coverage during 2014.

Research and the Media

Healthier Kids Conference: Insights from Twin Research

The Healthier Kids Conference was held on Friday, December 5th at the new Royal Children's Hospital building. The first of its kind in Australia, this conference brought top international and national twin researchers together and was a great success. The conference had a full itinerary of 23 presenters and 95 delegates (including 30 twins) and a total of 250 attending.

A primary goal of the conference was to highlight the extraordinary power of twin research designs for understanding the origins and development of human physical, behavioural and medical traits. However, the conference was unique in also inviting twins and parents of twins to provide input into the research process.

Key themes were: (1) the significance of twin research findings for the general public, (2) the importance of involving clinicians and other medical specialists in twin research, and (3) the need to solidify ties between researchers and twins. Throughout the conference, the goal of more closely addressing challenges facing families with twins and other multiples was emphasised.

The mid-day keynote speaker was Nancy Segal, The Minnesota Study of Twins Reared Apart (MISTRA). Her presentation generated a lot of interest which was followed by an open forum between the researcher and twin community, finishing with a concurrent last session for researchers and twins.

See next page for a list of topics covered included.



The ATR received extensive media coverage in the lead-up to and on the day of the conference

"Nancy Segal is professor of Psychology at the California State University, Fullerton and who incidentally is a twin herself has made the study of twins her life's work; she studies both identical and fraternal twins who have been raised separately and together. These twins have been able to provide scientists and social researchers insight into the extent of genetic and environment causes of a range of traits."

Twins, what these genetic miracles can teach us

Penny Johnston, 774 ABC
Melbourne

Topics covered included:

Conception to Birth

- The Role of the Obstetrician
- The Importance of the Intrauterine Environment in Shaping the Neonatal Genome
- Twin Placentas – Reading the 'Diary of Pregnancy'
- The Life and Death of Chang and Eng
- Fetal Therapy in Twins

Childhood

- Twins Modelling
- The Genetics of Epilepsy
- Language and Literacy
- Bone Health
- Tooth Emergence and Dental Caries
- Eye Disease

Forum

- What Twins Want from Researchers
- What Researchers Need from Twins

Adolescence

- Brain Imaging and Genetics
- Disordered Eating Over Adolescence
- DNA Methylation
- Genomic Studies with Twins
- Including Twins in Clinical Trials

Twins and Families

- Raising Twin Children
- Health of IVF Twins and Triplets
- Twins Singing Study
- The Genetic Basis of Speech and Language in Twins
- Individuality and Twinship
- Challenges Facing Parents of Multiples

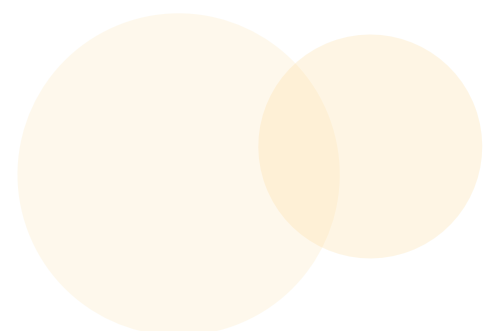
The ATR received extensive media coverage in the lead-up to the conference and on the day.

Twin Pregnancy Booklet

The ATR, in conjunction with health professionals and twin parents, has developed a comprehensive *Twin Pregnancy Booklet*, available free of charge to families expecting twins. The booklet contains helpful information about twins, what to expect during pregnancy and when the babies come home. Currently the ATR is sending out over a thousand booklets annually to expectant and new parents all over Australia, and demand for this comprehensive resource is growing significantly each year.

The booklet has received great feedback from parents who tell us there are few quality and easily accessible resources about raising twins. As the foremost authority in Australia on twin health and research, the ATR provides a trusted source of information to families.

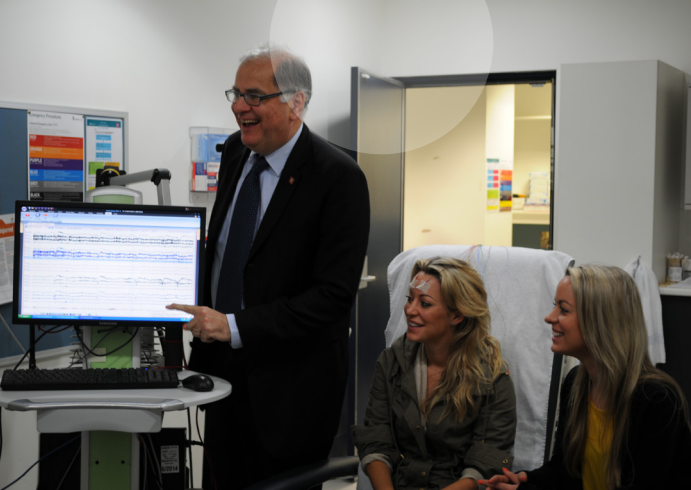
2014 has been spent on updating the booklet for re-print in 2015.





Scientific Merit

- 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.



High Profile twin researcher awarded collaboration with the ATR

A simple flyer caught the attention of a young researcher, Professor Sam Berkovic, nearly 30 years ago and inspired him to begin a life-time's work with twins.

Prof Berkovic returned to Australia in 1987 after studying overseas, eager to conduct further research in his main area of interest — epilepsy. As he read a booklet about how to apply for grants, he noticed a bright yellow one-page flyer about the Australian Twin Registry, and the rest became history!

Sam went on to involve hundreds of ATR twin members in his research, contributing to a deeper understanding of the genetic and environmental causes of epilepsy.

His research has led to improved epilepsy diagnosis and treatment worldwide. Today, he is a world-leading clinical neurologist, a Laureate Professor in the Department of Medicine, University of Melbourne and Director of the Epilepsy Research Centre of Austin Health.

ATR Director, Professor John Hopper, said the award – which recognises eminent achievement and merit of the highest degree in service to Australia or humanity at large – could not have gone to a more worthy recipient.

“All of us at the Australian Twin Registry and our twin members congratulate Sam,” he said. “He has long been a great ambassador for twin research and we look forward to supporting more ongoing vital work which has made such a difference to millions of people living with epilepsy.”

Photo (above): Sam takes identical twins and ATR members, Amanda and Nicole Campbell, on a fascinating journey through the Melbourne Brain Centre

Current Research Studies

The ATR continues to enable researchers through the recruitment of twins for health-related studies.

Studies are classified by 3 areas:

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

The total number of active and ongoing studies utilising ATR services and/or involving ATR members were 12 studies in 2014. This includes the active processing of 3 Expressions of Interests (EOIs) for new research, 1 new research application (as a result of an approved EOI) were processed throughout the year. In total, there were 9 active recruiting studies with participants in 2014, 3 of which were completely new, and the provision of ad hoc support to a further 38 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 2014, is shown in **Table 2** and a count of active recruiting studies in the past seven years is shown in **Figure 10**.

Table 2: Studies by Status 2014.

Study Status reached by end of 2014	Number
Application (EOIs, Full Application, Protocol Change)	11
Recruiting	9
Data Collection	5
Data Analysis	19
Writing Up/Publishing	14
TOTAL	58



Twin research shows us that our teachers may be doing a better job than we think

It is often assumed that differences in teacher characteristics are the main source of variability in children's educational achievements. Estimating and understanding teacher effects is important not only for gaining insight into individual student performance but for guiding investments in teacher training, career advancement and remuneration.

But ongoing twin research by Professor Brian Byrne and Dr Will Coventry at the University of New England in NSW is challenging some of these long held assumptions.

The researchers first began looking at student reading ability in 1999 with the help of identical and non-identical twin pairs from the Australian Twin Registry. They have since extended their research to include NAPLAN results (National Assessment Program: Literacy and Numeracy), with the cooperation of over 2,500 families of multiples registered with the ATR.

"To follow the public debate on why some children prosper in school and others falter, you'd think it was all down to teachers," says Professor Byrne. "The media as well as public figures and politicians are quick to blame any student failures on inadequate teaching."

However, his research has found there is a complex interplay of genetics and environmental factors happening in the classroom. Some findings so far include:

- Many factors affect a child's progress including heritable or genetic factors, classroom environment, classroom peers, as well as individual teachers.
- Previous education researchers held that teacher quality will show up most clearly when a child has a "bad" teacher several years in a row. In the UNE research, there was no support for that idea.
- Teachers are doing a much more equal job in fostering early literacy and they have less to do with differences between students' reading achievement than often supposed.
- Interestingly for parents of twins, the researchers found twin pairs who were in separate classes from each other were almost as similar to each other in literacy development as twins in the same class.

Professor Byrne says twins are invaluable in these studies and he thanks the many ATR families for their support. The research is continuing and more twins and their families are welcome to join in. Learn more at UNE's website.

Figure 10. Number of active recruiting studies, reported as at Annual Report publication.



Health & Lifestyle Questionnaire

The ATR Health and Lifestyle questionnaire commenced in January 2014, with versions developed for Adults (18+) and Juniors (below 18). This online questionnaire asks about a range of topics including members' backgrounds, health conditions, families and lifestyles. All members of the Australian Twin Registry, with email addresses, are invited to participate in this important questionnaire.

The questionnaire aims to:

- Provide a more detailed description of ATR members to our researchers when planning studies
- Help the ATR invite twins to participate in research studies most relevant to them
- Provide summary findings to members and journals

To date:

- The questionnaire has been sent to 15,431 adult and 7,492 parents of junior twins
- 5,666 adult twins (1,393 pairs and 2,880 individuals) and 2,441 parents of junior twin pairs have completed the questionnaire.

All adult data below is based in individual responses except for *Zygosity* and *Gender of Responding Pairs* which presents paired data. All junior data presented here is paired data.

Figure 11. Adult responders by age group.

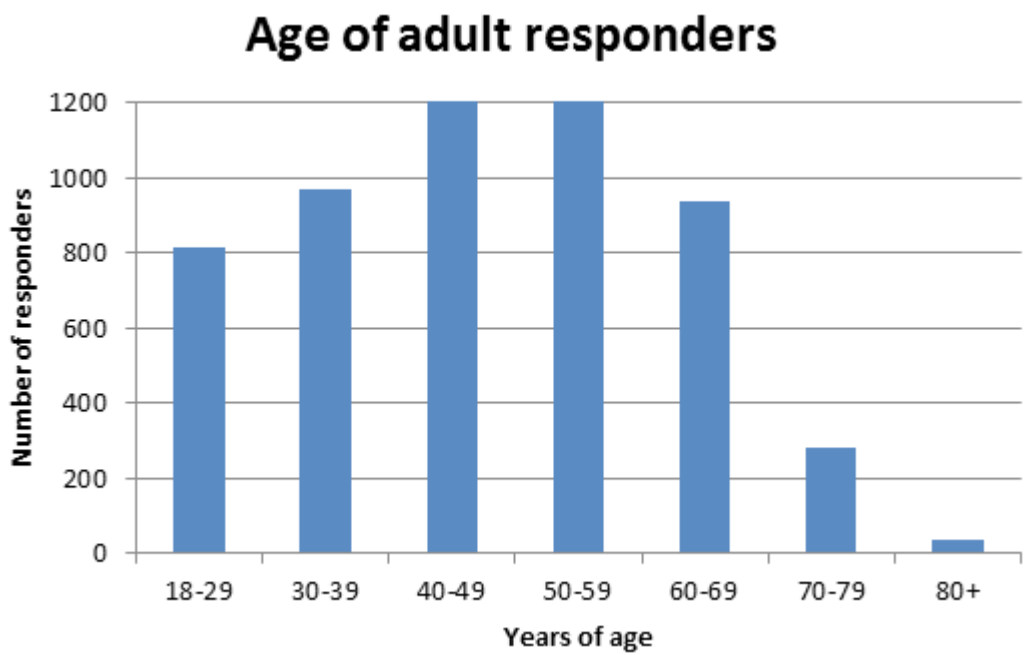


Figure 12. Junior responders by age.

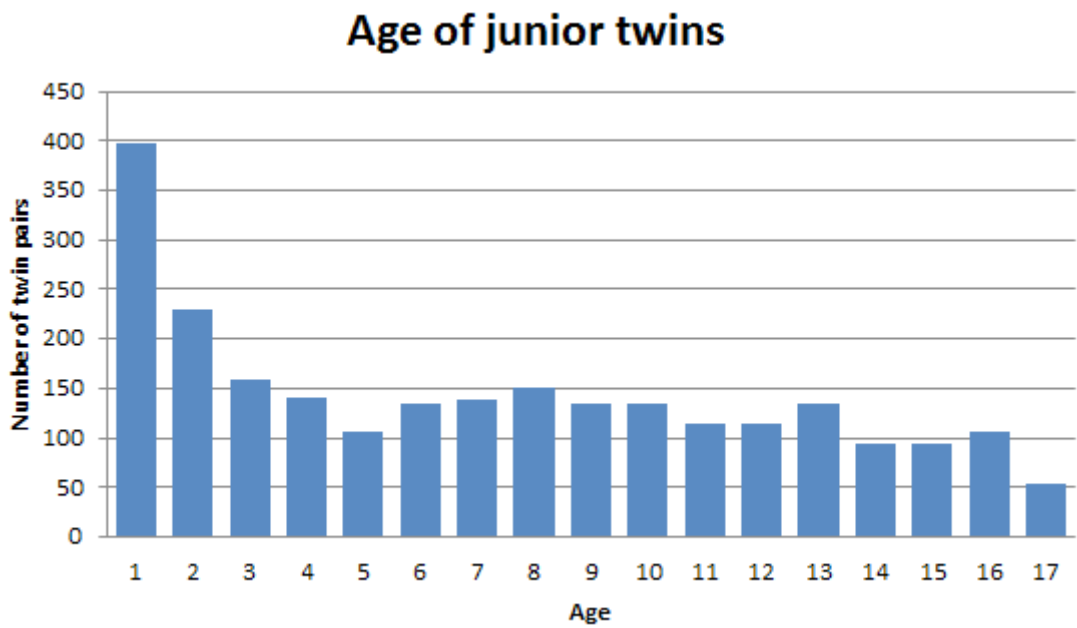


Figure 13. Adult responders by zygosity and gender.

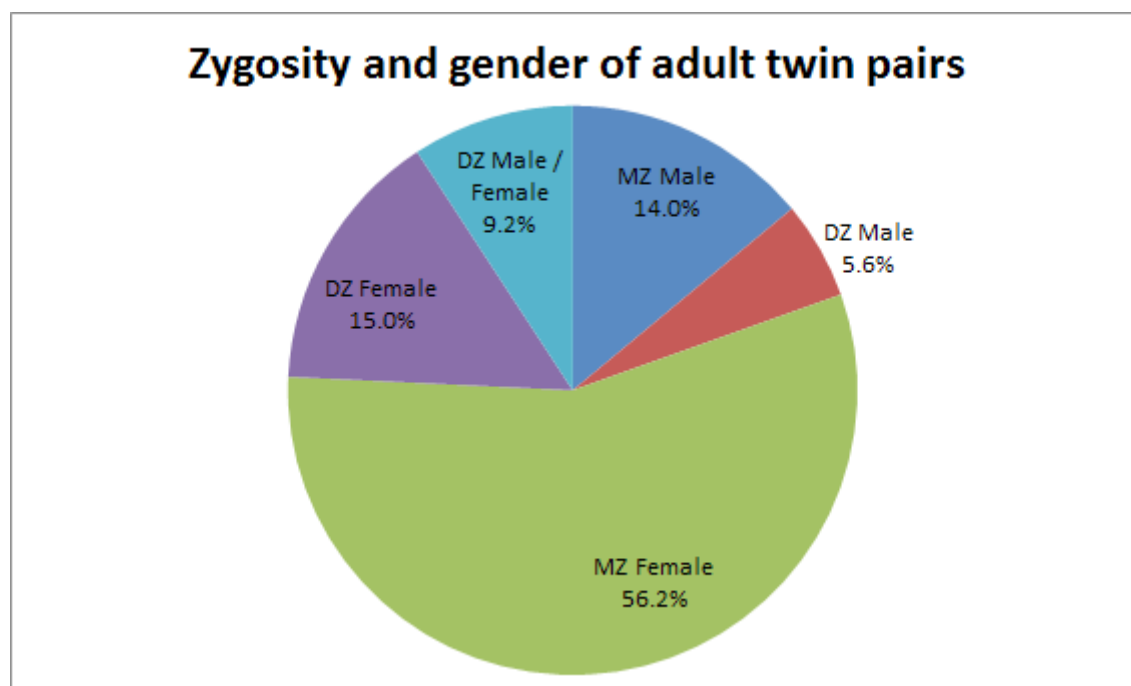
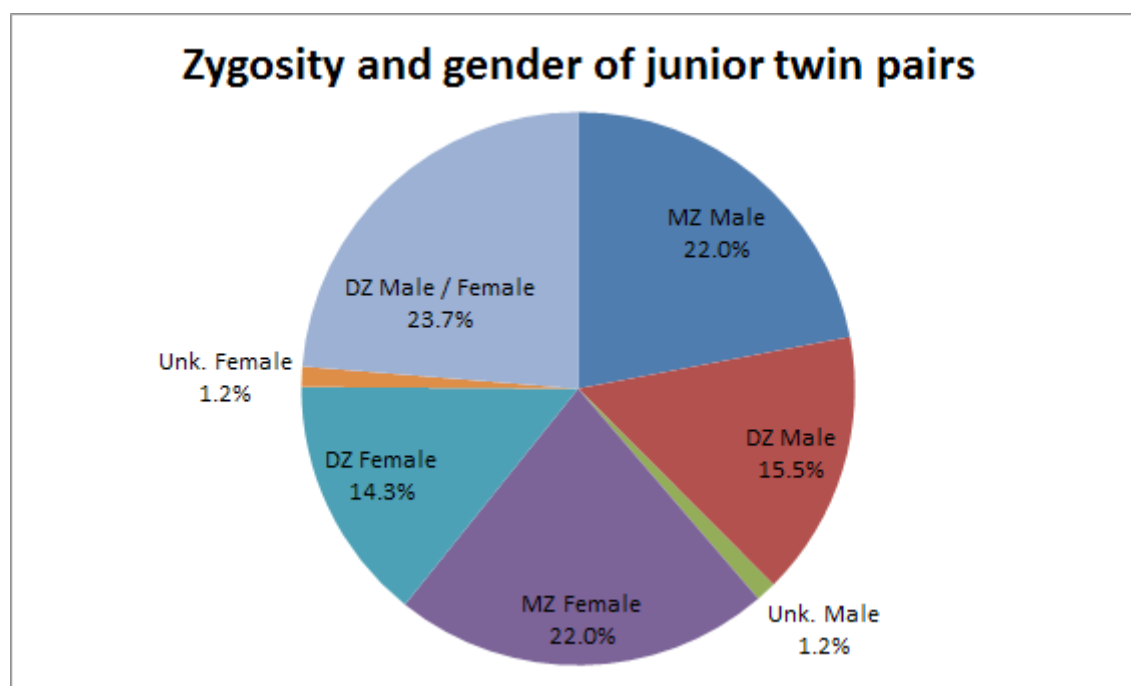


Figure 14. Junior responders by zygosity and gender.



In 2014, one study requested data from the Adult Health and Lifestyle questionnaire (*The causal effect of education on health & health related behaviours, DATA-1403*).

Researcher Reports

The Researcher Annual Progress Reports provided by the researchers and associated staff summarise the current activities undertaken by each study active in the current reporting period

(1 January to 31 December 2014), major achievements for this period and future plans.

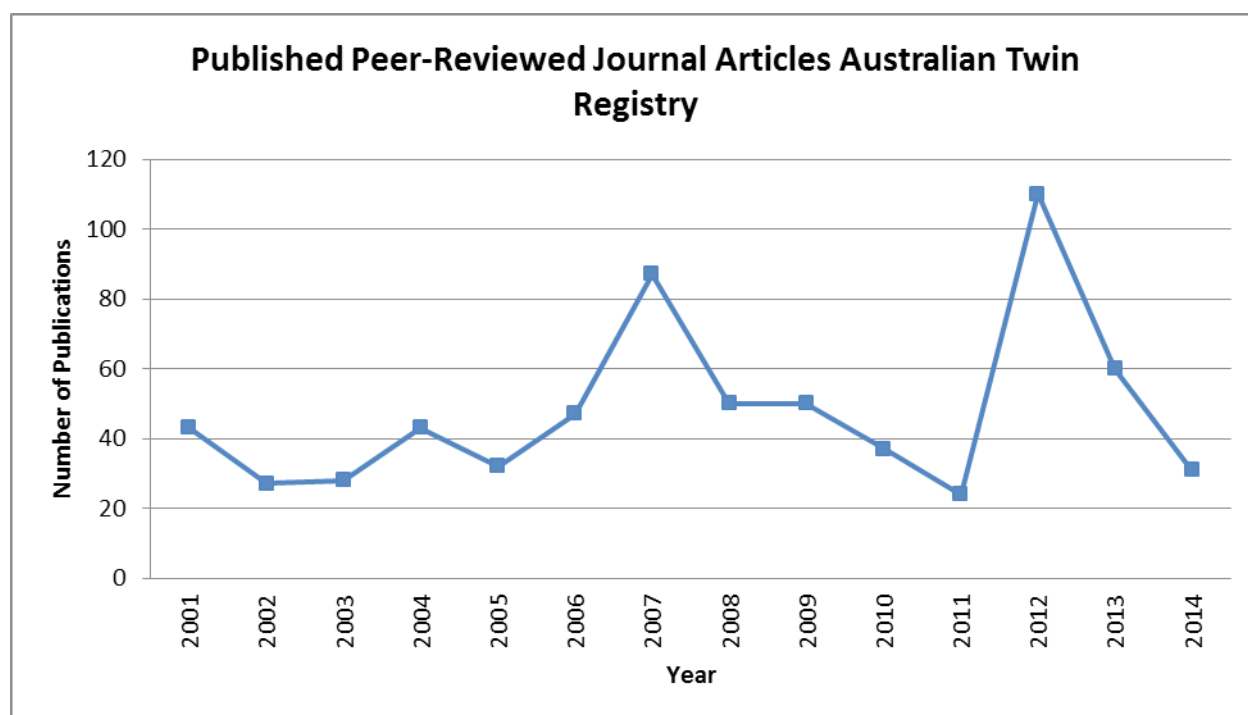
The Annual Researcher Satisfaction Survey has been fully integrated with the ATR's Annual Progress Report. Refer to Value Add section.

Publications

An important measure of the output of the ATR is the number of publications arising from studies supported by the facility. An important goal set by the ATR in the Enabling Grant was to increase the number of peer-reviewed articles to 50 per year. On average the ATR has facilitated 52 publications per year over the last 5 years.

In 2014, the ATR has recorded 31 peer-reviewed journal articles (**Figure 15**), 2 book chapters, and 17 conference proceedings, for a total of 50 publications. The goal of having 50 peer-reviewed articles yearly was reached, although 6 additional publications were reported to be in press or in preparation in 2014. The list of all 2014 publications can be found in [Appendix 3](#).

Figure 15. Number of articles arising from ATR studies that were published in peer-reviewed journals every year since 2001.



Meetings and Conferences

- Healthier Kids Conference: Insights from Twin Research (refer **Media Activities**).
- Jeff Craig, Tess Cutler, and John Hopper attended the Joint 3rd World Congress on Twin Pregnancy & 15th International Congress of the International Society of Twin Studies (ISTS) in Budapest. John presented one talk and two posters and Tess presented two posters. Jeff presented two talks. INTR had a separate session on biospecimens and a paper by Twin Research and Human Genetics was included.
- The ATR held a stand at the Maternal and Child Health Nurses Conference. 900 people registered for the event and it provided the ATR an opportunity to give information back to the health community and raise awareness of the ATR.

Research Travel Grant Scheme

The Travel Grant Scheme supported a total of 19 early-career researchers in Round 12 and 13 to attend national and international research workshops and conferences relating to twin research.

Grants for Round 12 of the ATR Research Travel Grant Scheme were awarded in March 2014 and Round 13 in October 2014.

Congratulations to the following successful recipients:

Round 12

- Ashild Bjornerem – Department of Health and Care Sciences, Faculty of Health Sciences, UiT – The Arctic University of Norway
- Karen Mather – Centre for Healthy Brain Ageing, School of Psychiatry, UNSW Medicine, University of New South Wales
- Jane Ebejer – Genetic Epidemiology, QIMR Berghofer Medical Research Institute
- Benjamin Harrison – Melbourne Neuropsychiatry Centre, Department of Psychiatry, The University of Melbourne
- Michelle Lupton – Neuroimaging Genetics, QIMR Berghofer Medical Research Institute
- Yi Lu – Statistical Genetics, QIMR Berghofer Medical Research Institute
- James Sherlock – School of Psychology, University of Queensland
- Baptiste Couvy-Duchesne – Genetic Epidemiology group (Neuroimaging Genetics), QIMR Berghofer Medical Research Institute
- Jessica Miller – Susceptibility to Paediatric Infection, Infection and Immunity Group, Murdoch Childrens Research Institute
- Ruba Odeh – Craniofacial Biology Research group, Faculty of Health Sciences, The University of Adelaide
- Negar Shahmoradi – Endocrine Centre for Excellence, Austin Health
- Katrina Scurrah – Department of Physiology, University of Melbourne



“Overall, the TWINS2014 conference was an amazing opportunity and provided a thorough insight into international twin research. I am extremely grateful to the Australian Twin Registry for providing me with a Travel Grant to support my attendance at this event.”

Aneeka Bott,
Department of
Anaesthesia and Pain
Medicine, Sydney
Children’s Hospital

Round 13

- Aneeka Bott – Department of Anaesthesia and Pain Medicine, Sydney Children’s Hospital
- Tessa Cutler – Australian Twin Registry
- Justine Gatt – School of Psychology, UNSW and Neuroscience Research Australia
- Anita Goldinger – Quantitative Genetics Group, Diamantina Institute, The Translation Research Institute

- Yuk Jing Loke – Early Life Epigenetics, Murdoch Childrens Research Institute
- Joseph Powell – Centre for Neurogenetics and Statistical Genomics, Queensland Brain Institute, University of Queensland
- Richard Saffery – Cancer and Disease Epigenetics, Murdoch Childrens Research Institute; Department Paediatrics, University of Melbourne

Grants provided opportunities for new researchers to attend diverse meetings and conferences. To date \$44,214 has been allocated to 80 young researchers since the beginning of this Enabling Grant.

International Research and Research Training Fund (IRRTF)

This will consist of \$50,000 per year for three years to work with Dr Paul Ferreira, University of New South Wales and his collaborators in Brazil to conduct a twin study with lower back pain and to enable the establishment of a Brazilian Twin Registry. This also includes partial funding for a Brazilian PhD student, Lucas Ferreira to be based at the ATR for three years.

The ATR will assist in the:

- 1) Design of the Back Pain and Health and Life Style Questionnaire;
- 2) Twin recruitment protocol;
- 3) Data collection, management and archiving; and
- 4) Establishment of the Brazilian Twin Registry systems in governance, database, researcher collaborations and applications and data management of twin members.



■ *Participation*

- Use judicious management and administration of approach to 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.

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 2014, 42 mail outs were conducted and a total of 27,924 letters or emails were sent (**Table 3**).

Table 3: Total number of mail outs and total letters by email and mail in 2014.

Study ID	Study Title	Num Mail Outs	Total Letters/ Emails	RTS Rec'd	RP Mail Rec'd
2013-005	Genetics of speech and language disorders.	1	43	0	0
2012-003	A twin study of the NAPLAN	1	289	10	98
2012-014	A study of breast cancer and epigenetics	1	126	3	21
2013-001	Why do we get addicted? A population based twin study of the relationships between impulsive and compulsive behaviours	2	958	0	0
2012-012	Why do some people overconsume fatty foods	4	1724	4	108
2012-010	Investigating the genetic basis of singing ability: a twin study	3	1755	0	0
2008-002-3	Type 2 diabetes and brain function – a co-twin study of brain metabolism	4	106	57	1
ATR 002	ATR junior health and lifestyle questionnaire	9	7492	0	0
ATR 001	ATR senior health and lifestyle questionnaire	21	15431	0	0
DATA-1403	The causal effect of education on health & health related behaviours	0			
	TOTALS	42	27,924	770	120

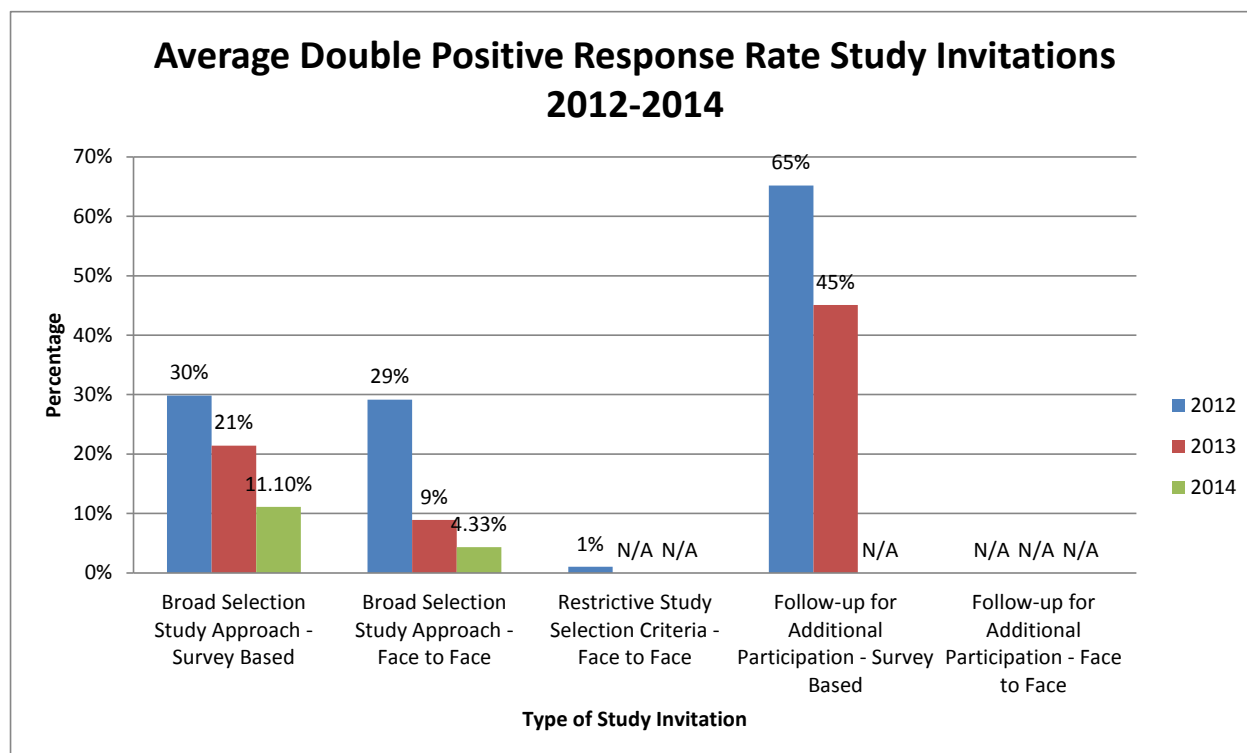
Table 3 shows these numbers by study and summarises approaches for both Junior members (1 approach per family) and Senior members (1 approach per twin). The number quoted for each study also includes reminder mail outs.

Responses to Invitations to Participate in Research Studies

The overall response for a study is defined as the number of 'Positive ("Yes")' and 'Negative ("No")' responses over the total number of twin members approached. The 'Response Rate' (RR) is an important statistical element in the interpretation of research results and as such, the ATR aims to obtain a response from as many members approached as possible.

Figure 16 includes positive responses from both individuals in a twin pair, or an individual response from a parent of a junior twin pair. It shows these double response rates, or double "yes," for survey-based studies in 2014, and for those studies that have been ongoing since 2012 in three categories – Broad Selection Study Approach – Survey Based, Restrictive Study Selection Criteria – Face to Face, and Follow-Up for Additional Participation. Some study response rates will show responses for each year between 2012 – 2014, whilst other studies will only show for one or two years depending on the format of the study. *Please refer to Appendix 4 for further break-down of Broad Selection Study Approach and Follow-Up figures.*

Figure 16: Study response statistics to date for most active studies 2012-2014 (ongoing and recruiting)



Response rates to Registry mail outs will depend on the study. For example, standard open studies which have few criteria on participants often receive more positive study responses, such as the emotional well-being and lower back pain studies, whereas a study requiring a specific criteria or re-approach to participants is often due to 'hard to get' participants, such as the anti-epileptic and bone health study.

Response rates to Registry mail outs appear to be higher for those studies approaching either families with young twins (under 18 years old) or older, adult twins (40 years and older). Twins between the ages of 18 and 40 years have the highest 'NR (*Nil Response obtained*)' and 'Negative' RR and are the most difficult group for which to maintain current contact information. 'Negative' RR also include twins who are ineligible to participate in a study based on the criteria set by researchers, for example, where the member does not display a particular trait or does/does not suffer from a particular disease.

Telephone Follow Up

As part of its services, the ATR offers researchers the option of telephone follow-up, which can be used in conjunction with reminder letters or as a stand-alone, follow-up mechanism. This form of follow-up was used by half of the ATR actively recruiting studies during the reported period.

The number of hours and resulting phone calls for study phone follow-up are outlined in **Table 4**. Please note, these figures do not include telephone calls and hours spent tracing twins who have changed address.

Table 4: Number of calls and hours spend on the calls by the ATR staff in 2014.

Study ID	Study Title		Total Hrs
2013-005	Genetics of speech and language disorders.	35	3
2012-014	A study of breast cancer and epigenetics	139	12.5
2012-012	Why do some people overconsume fatty foods	139	13.5
2013-001	Why do we get addicted? A population based twin study of the relationships between impulsive and compulsive behaviours	232	15.5
2008-002-3	Type 2 diabetes and brain function – a co-twin study of brain metabolism	128	16
	TOTALS	673	60.5

A decline in researchers requesting Telephone Follow Up in favour of email follow up continues in 2014. (Refer to **Figure 19** to see the changing response modes to studies). The decrease in Telephone Follow Up, from 7,263 in 2013 to 673 in 2014 can be attributed to labour costs.

Record Updates

The Registry also undertakes proactive tracing of its members, this is an ongoing and important maintenance activity and ensures that the Registry remains viable. All prior addresses and any actions taken to trace members are recorded on the ATR database.

In 2014, a total of 11,410 individual twin and other contacts (i.e parent or secondary carer) member records were updated in the ATR database. This number includes an individual where their individual record, address information or phone number/email address has been edited, and those records followed-up due to receipt of a Return to Sender (RTS), and routine tracing or contact after a study approach has been sent. A count of all individual records updated yearly since 1994 is shown in **Figure 17**.

Figure 17. Numbers of individual records updated per year (2004-2014).

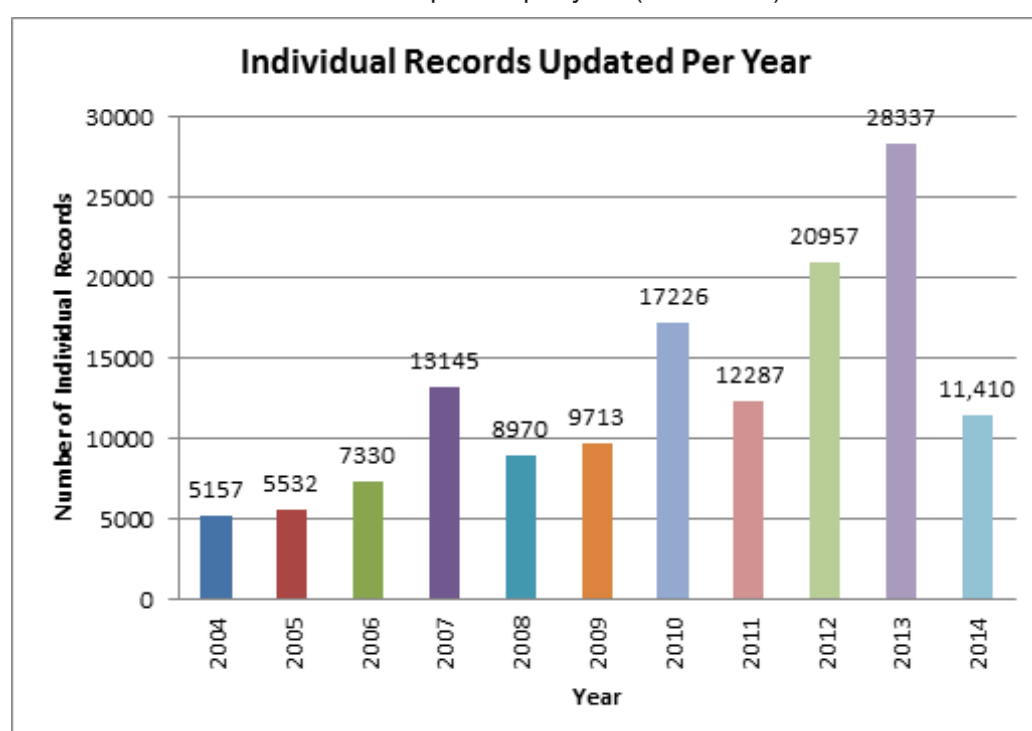
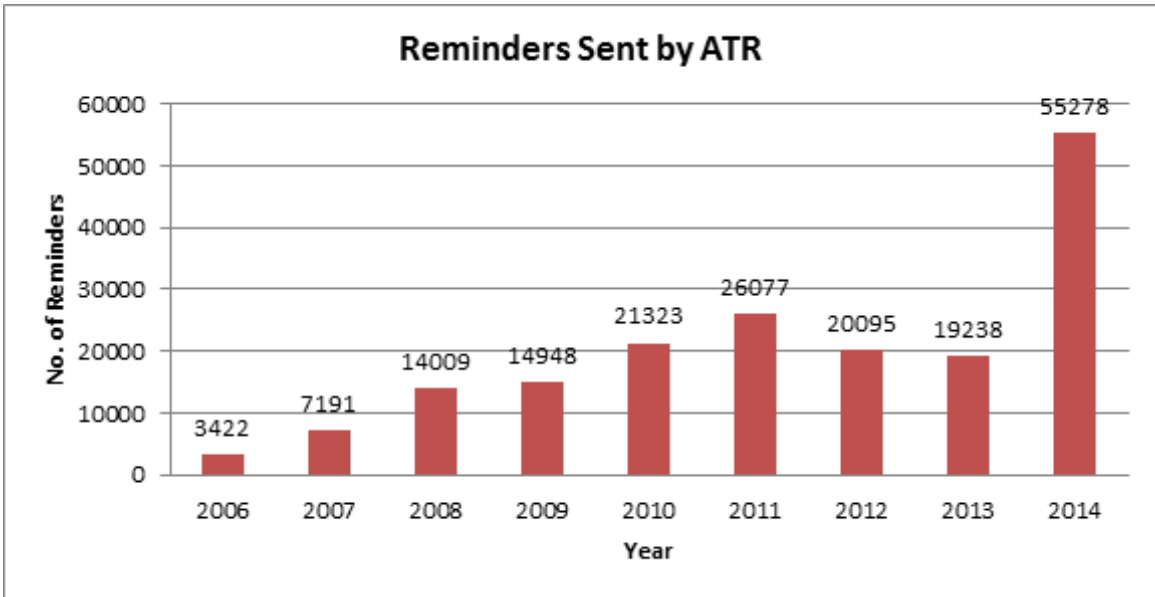


Figure 18 illustrates the number of reminders and follow-ups generated by ATR staff in the past nine years. The 2014 spike (55,278) was due to the Health & Lifestyle Questionnaire being sent to all active twin members of the Registry.

Figure 18. Numbers of reminders sent by the ATR staff (2006-2014).



The majority of the 55,278 reminders were by email (**Figure 19**). The large number of reminders sent in 2014 are due to the ATR Health & Lifestyle Questionnaire being sent to all members with email addresses.

Figure 19. Number of reminders by category (phone, mail and email) 2010-2014.

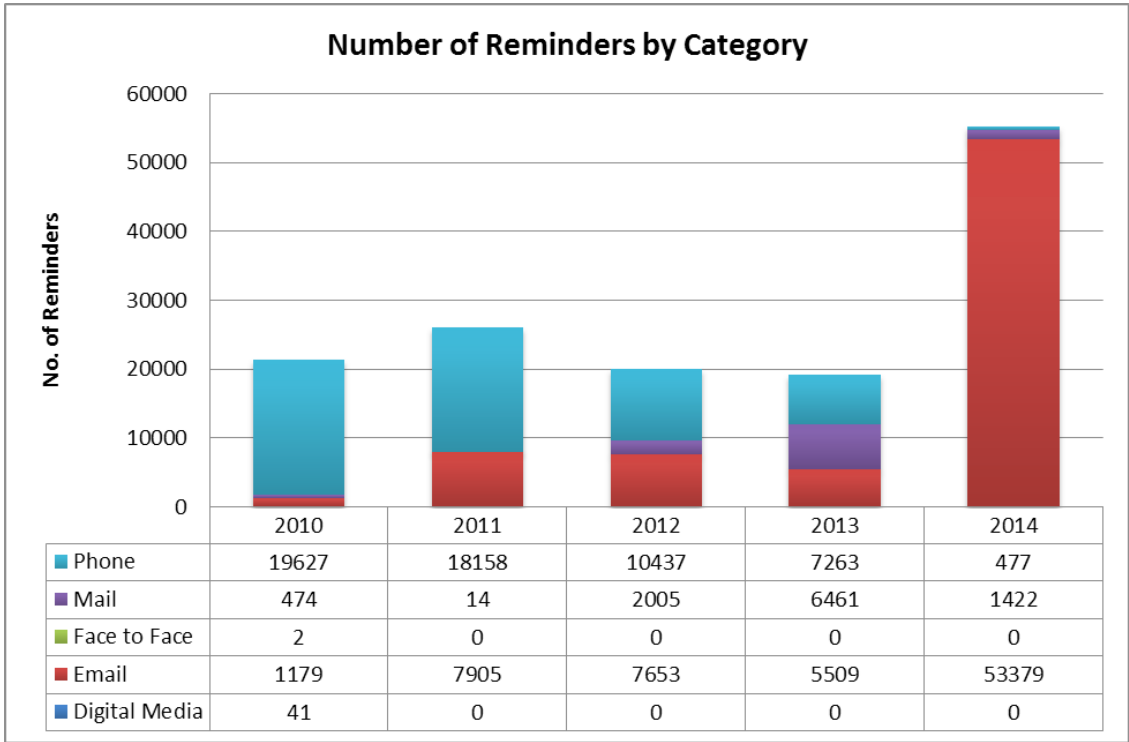


Figure 19 highlights how in 2014, there was a dramatic increase in email as the dominant tool for sending reminders to ATR members, while the number of phone and mail reminders have been reduced to a minimum. Although email is potentially far more efficient and economical, it also has a reduced response and tends to select for members who respond more favourably to emails.

Adverse Effects and Complaints

The ATR takes any complaint from members seriously and endeavours to promptly resolve the issue presented. The ATR requires all adverse effects and complaints to be communicated to ATR Management.

During the reporting period, one ATR member filed a complaint to the Registry about a study facilitated by the ATR. ATR Management consulted the Advisory Board and sought input to resolve the issue satisfactorily for both parties.



■ *Value Add*

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



The ATR received some great feedback from the research community for 2014:

“Exemplary assistance and guidance,”

David Champion (pictured above), Department of Anaesthetics and Pain Medicine, Sydney Children’s Hospital

“Without the ATR network of twins a study of this kind would be almost impossible to organise and execute,”

Caryll Waugh, HIV and emerging infectious viruses, Deakin University Medical School and CSIRO AAHL

“We have always found that the communication with the ATR has been efficient, friendly and informative,”

Prof Daniel Green, School of Sport Science, Exercise and Health, The University of Western Australia

In response to the NHMRC’s requirement for stakeholder feedback, the ATR implemented a Quality Assurance (QA) Program in 2006 that incorporates stakeholder satisfaction feedback, monitors the quality of service delivery to twin members and researchers and identifies critical points during the implementation and roll out of a research project where reflection and forward planning are important to maintain quality. The ATR has two major stakeholders: twin members and researchers working with the ATR.

The 2014 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 researchers’ overall satisfaction with communication with the ATR;
- (2) the researchers’ 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

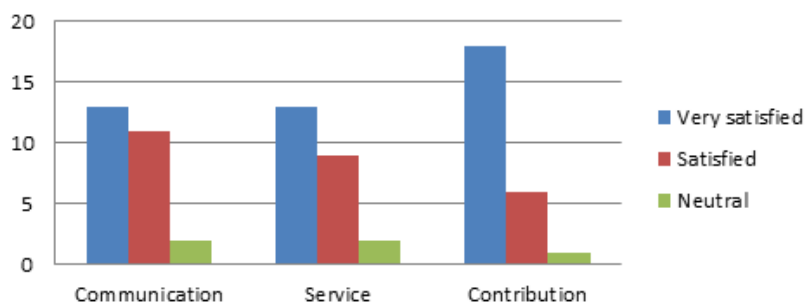
Responses were recorded as

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

We received feedback from 32 research groups. Overall, researchers were very satisfied with the contribution overall provided by the ATR and also the communication and service the ATR has provided (**Figure 20**).

Figure 20. Overall satisfaction scores from researchers, evaluating communication with ATR, and service and contribution provided by the Registry.

Scores for Overall Satisfaction from Researchers who have interacted with the ATR in 2014



Communication:

"The communication with the ATR staff has been excellent, as emails have been promptly responded to, and face to face meetings or telephone conversations discussing aspects of the project have been extremely helpful. The ATR staff have also informed us promptly of any technical problems or other complaints from the participants so that we can address their concerns in a timely manner."

Service:

"It has always been a true pleasure to work with you all! I am grateful for the opportunity and I thank you for all that you do to make good science happen and to facilitate such a great sense of community!"

Contribution:

"It would not have been possible to conduct our twin study without the twin database maintained by the ATR. Being invited to present or set up a booth at ATR-run events such as the Healthier Kids Conference and Twins Plus Festival this year also provide valuable opportunities for us to reach out to a large pool of potential participants as well as network with other twin researchers, both of which we are grateful for."

"Without the ATR, the scale of this project would not have been possible."

"Without the ATR, it would have made it very difficult to recruit twin participants. The 2012 Twins Plus Festival provided an excellent opportunity for data collection."

ATR Data Index Project

To accomplish the ATR's goal of fostering new research collaborations, the Registry created a web-based, searchable index of questions and topics covered by previous studies conducted via the ATR. The index was launched in 2009 and it is accessible at:

http://www.twins.org.au/study_index/BasicSearch.php.

On this webpage, users can conduct searches based on keywords and specific criteria. A search generates a list of all previous studies matching the search items, including ATR study ID number, study title, investigator/s, host institution/s, year/s study conducted, and study status (completed, in progress, etc). Advanced searches return the types of twins approached for each study (e.g. MZ/DZ; male/female; adult/junior combinations); any questionnaires or tests administered and any other measures or samples taken. Records can be systematically reviewed and checked against existing hard copy files to ensure listings are complete.

Researchers interested in utilising the existing data itself can contact the Registry initially, who then fosters a link with the originating research group.

The Ark

In 2010, the ATR became a collaborative development partner for The Ark, an international informatics project based at the University of Western Australia. The Ark suite of web-based tools will assist the ATR's day-to-day operations and will also be available to researchers conducting twin research. Ultimately, access to the ATR hosted instance of The Ark software will minimise the need for researchers to develop and host systems to manage their studies. Ark modules are currently in place to support recruitment, subject management, electronic questionnaires, modelling and visualisation of pedigree structures (with multiple birth support) and management of phenotypic, disease and biospecimen data.

During 2014 there have been seen several major advances in Ark development work. The data extraction module has been optimised and now produces exports much more quickly than previous releases. A "global search" function has been added to enable dynamic searches for Registry participants across multiple studies and sub-studies based on demographic and biospecimen criteria. A

configurable “Disease” module has been introduced to capture the affected status and related disease information for participants. In addition to these changes, over 70 miscellaneous improvements have been implemented, along with resolutions for small issues and bugs.

The remainder of 2015 will be spent polishing the system towards a milestone Ark 1.3 release, with a strong focus on usability and stability. This includes significant improvements to the user-interface of the phenotype module, and integration of the disease module with the pedigree visualisation component.





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ATR Management

As of 31 December 2014, the ATR Management comprised:

- Prof John Hopper, AM, Director, ATR; NHMRC Senior Principal Research Fellow; Director (Research), Centre for MEGA Epidemiology, University of Melbourne
- A/Prof Jeff Craig (Deputy Director ATR, *appointed as Deputy Director in October 2013*), Group Leader, Early Life Epigenetics at the Murdoch Children's Research Institute)
- Mr Vincent Pollaers, Chair, Advisory Board
- Kate Murphy, ATR Manager
- Jenny Boadle, ATR Study Coordinator
- 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.

Advisory Board and Charter

In 2014, the Advisory Board members were:

- Mr Vincent Pollaers (Chair, Twin Representative, New South Wales)
- Mrs Karen Willetts (AMBA Representative, New South Wales, *appointed in November 2013*)
- Dr Paul Jelfs (Australian Bureau of Statistics, Australian Capital Territory)
- A/Professor Paul Lancaster (University of Sydney, Retired)
- Ms Sue Carrick (Twin Representative, New South Wales)
- Prof Margaret Otlowski (University of Tasmania, Tasmania)
- A/Prof David Whiteman (Queensland Institute of Medical Research, Queensland)
- Dr Keith Horsley (Australian Institute of Health and Welfare, Retired)

Ex-officio:

- Prof John Hopper (Director, ATR, University of Melbourne)
- A/Prof Jeff Craig (Deputy Director ATR, Group Leader, Early Life Epigenetics at the Murdoch Children's Research Institute), was appointed as Deputy Director in October 2013



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 Health at the University of Melbourne for his contribution to medical and scientific research. He is a Special Adviser to the Collaborative Grant Program at the National Breast Cancer Foundation.

He currently holds the position of General Counsel & Company Secretary of Greencross Limited, a leading ASX200 pet care company. Prior to this, he was Group Human Resources Manager, General Counsel and Business Development Executive – Services, for Mammoth Pet Holdings Pty Ltd, parent company of the Petbarn and Animates pet specialty retail chains in Australia and New Zealand. Past career highlights include: a corporate lawyer with Freshfields, London; General Counsel, IBM Australia/NZ; Managing Director, McKinney Rogers Asia Pacific.

Mr Pollaers holds Bachelors' degrees in Electrical Engineering and in Computing Science. He also holds a Post Graduate Diploma in Applied Psychotherapy and Counselling. He completed the Common Professional Examinations and the Law Society Finals at the College of Law in London and is a qualified solicitor in New South Wales, and in England and Wales.



Karen Willetts

Karen's role as a mother of (now 13 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.

Following the arrival of her twins she joined her local club in the Lower North Shore of Sydney in 2002, where she has held many roles including President. In 2006 she became part of the AMBA State Committee, and the following year, undertook her first role at national level with AMBA – State Liaison Director. In 2008, she was elected at national level as AMBA Chairperson; a position she held for 3 years. Having completed the maximum tenure, Karen stepped down from the AMBA Board of Directors in 2008. She continues to play an active role within the organisation and brings a wealth of experience and knowledge to the ATR.



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 seeks to drive new and innovative information developments across a range of social issues, working collaboratively with a range of stakeholders and partners.

Paul has extensive experience in Commonwealth and State Government agencies in both information management and service delivery. He has worked across these agencies to promote the best use of information to support government, non government agencies and the community to plan, make evidence based policy decisions and conduct quality evaluations. Paul 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

Professor Paul Lancaster was the founding Director of the National Perinatal Statistics Unit in 1979 (now part of the University of NSW) and, in 1983, established the world's first national register of IVF pregnancies.

In 1991, he initiated an international program to monitor IVF and steered the establishment of the International Centre for Birth Defects in Bergen, Norway. In 1994, he instigated the Master of Medicine (Reproductive Health Sciences and Human Genetics) at the University of Sydney.

Other career highlights include Chairing the Executive Committee of the International Clearinghouse for Birth Defects Monitoring Systems; a member of the WHO Expert Advisory Panel on Human Genetics; and President of the Australian Birth Defects Society.

He is an Honorary Associate Professor at the Northern Clinical School, University of Sydney; and a member of the International Committee for Monitoring Assisted Reproductive Technologies, and the Medical and Scientific Panel of the Infertility Treatment Authority of Victoria.



Sue Carrick

Sue Carrick leads Susan Carrick and Associates as a Special Adviser, Research Strategy and Partnerships to leading Australian research foundations. She has held senior executive and director roles with experience in growing national investment in medical research; forming and maintaining key government, corporate and not-for-profit relationships; and building a nationally coordinated approach to the way breast cancer research is conducted in Australia.

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 has a Master of Health Science and qualifications in qualitative research methods, family planning, midwifery, and general nursing. She is one of a male/female twin pair.



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 in this area has been reinforced through a number of her professional involvements including 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. Professor Otlowski provides research supervision of PhD and research masters degrees in the areas of Ethical Legal and Social Implications of Human Genetics, Family Law and Law and Ethics of Health Care/Medical Law.



Professor David Whiteman

Professor David Whiteman is a medical epidemiologist with a special interest in the causes, diagnosis, prevention and treatment of cancer.

Following a PhD in cancer epidemiology at the Queensland Institute of Medical Research (1997) and specialist training in Public Health Medicine, David was invited to the University of Oxford as a Nuffield Medical Research Fellow to work in several areas of cancer research. He returned to Queensland in 2000, where he is currently studying the causes of melanoma and diseases of the oesophagus, ovary and skin.

In addition to his research activities, he is a member of the Academy of the National Health and Medical Research Council of Australia (NHMRC), the National Research Advisory Committee for Cancer Australia, the Research Committees of Cure Cancer Australia Foundation and Wesley Research Institute, and the Fellowships Committee of the International Agency for Research on Cancer. He is Chair of the Barrett's Neoplasia Guidelines Committee for the Cancer Council Australia.

He is the father of twin girls who are now 20 years old.

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, Designer, two Administration Assistants, two phonestaff, research assistant, one PHD student and a part time Database Manager.

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

Dispute Resolution Process

The Dispute Resolution Process approved by the Advisory Board enables impartial and transparent management of any dispute arising between the ATR and stakeholders. No action was necessary under this process in 2014.

Budget

The NHMRC Enabling Grant Special Facilities Scheme provided the ATR with \$500,000 per annum between 2010 and 2014. The ATR also recovers costs associated with approaching twins for studies from researchers.

Donations

The ATR welcomes donations towards the administration and management of the Registry. Donors are provided with a receipt. Donations may be earmarked for specific activity.

We are very grateful for all the support we receive from Registry members and the wider community who have donated in 2014 towards a total of \$20,355

Appendix 1: Researcher Reports 2014

2012-003 A BEHAVIOUR GENETIC STUDY OF THE NAPLAN RESULTS OF AUSTRALIAN TWINS AT GRADES 3, 5, 7 AND 9 (TWINS AND NAPLAN STUDY)

Principal Investigator
Dr William Coventry
University of New England
Armidale, NSW

The National Assessment Program: Literacy and Numeracy (NAPLAN) tests are designed by educational authorities, are objective, and have been administered Australia-wide since 2008 so are unquestionably the most valuable national database on school achievement available. The twin studies of this data will provide a more solid base for public policy debates on educational policy and practice. The NAPLAN tests are administered in Grades 3, 5, 7, and 9 each calendar year, and over the course of a **5-year project**, 2012-2016, this study will collect data on approximately 1818 twin pairs in total, with 376 of these assessed across all 4 test occasions and the remainder contributing test scores ranging from 1 to 3 occasions.

The longitudinal behaviour-genetic study of the NAPLAN results at grades 3, 5, 7 and 9 will identify the influence of genes and the family and unique environment in explaining 1) individual differences in school performance and 2) stability and change across grades within Australia. The range of environmental measures collected will allow scrutiny of specific environmental factors and how they interact with genes to predict NAPLAN results for reading, writing, language conventions and numeracy. Finally, the study will further progress knowledge on the differential effectiveness of teachers in producing student outcomes.

During 2013, 2,200 families were recruited to the study. Questionnaire data concerning home demographics, parent attitudes and twin zygosity were collected from all families; child-specific questionnaire data was collected for approximately 80% of participants. NAPLAN results have been collected from education departments in NSW, QLD, WA, VIC and SA. Feedback to participants was included in the ATR Magazine, e-News and on the website and results from the study were sent via email newsletter to all participants with an email address (approximately 2,095 families out of 2,200 recruited) in December 2013 and a copy of this newsletter was also provided to the ATR.

Achievements in the period Jan 2014 – Dec 2014:

A second Australian Research Council grant was awarded in November 2014, allowing the project to continue for another 3 years.

Plans for Jan 2015 – Dec 2015:

Recruit new participants to the study. Continue collecting NAPLAN data from states and territories. Send the child-specific questionnaire to families of students sitting NAPLAN tests in 2015. Analyse data and write up results. Submit papers for publication

Recruitment to date:

Number of families recruited: 48

Zygosity: 6 x female DZ, 4 x male /female DZ, 9 x male DZ, 20 x female MZ and 9 x male MZ

States: 5 x NSW, 6 x QLD, 4 x SA, 16 x VIC and 17 x WA

What was sent:

Approach letter, info sheet for parents and child, family questionnaire, response form, consent forms

What they did with the twins: questionnaire and naplan results access

Feedback provided: n/a

Current Status: ACTIVE – RECRUITING

2012-010 INVESTIGATING THE GENETICS OF SINGING ABILITY: A TWINS STUDY

Principal Investigator
A/Prof Sarah Wilson
The University of Melbourne
Melbourne, VIC

Our study will be the world's first twin study to investigate the genetic basis of singing ability through an interactive online assessment. It aims to explore the relative contributions of genetic and environmental components to singing ability.

Feedback will be provided to the participants through the ATR in the form of a brief report outlining the findings of the study. Additionally, the research findings will be broadly disseminated through the media and at local, national and international scholarly and community Forums. The results will also be published in high impact, peer-reviewed journals and as a PhD dissertation for Yi Tang Tin. Links to publications arising from this study will be made available to participants through the ATR.

During 2013 the online software for the twin study was developed and pilot-tested in January -March. Recruitment of twins officially commenced in mid May 2013 following a successful media launch organised by the Australian Twin Registry (ATR). 1,077 twin pairs received email invitations from the ATR to participate in the online study. 106 twins participated in the study, of which there were 34 complete twin pairs.

2014:

Funding: John Hodgson Scholarship \$5,700

From the beginning of August onwards, we have been putting up promotional posters around the University of Melbourne Parkville and Victorian College of the Arts campuses. Online notices promoting the study were also posted on the University of Melbourne online student portal throughout Semester two. We have also posted advertisements on Gumtree and Facebook to promote the study. In addition, Ms Yi Ting Tan was interviewed on 3RRR radio station on 27 Aug 2014 and had the opportunity to promote the study on the Hullabaloo show.

Publications

Tan Yi Ting. (2014) Investigating the environmental influences underpinning singing ability using a twin survey. Conference presentation at 37th National Conference of the Musicological Society of Australia.

Tan Yi Ting. (2014) Let's Hear Twins Sing. Conference presentation at the Australian Twin Registry's Healthier Kids Conference.

Tan YT, McPherson GE, Peretz I, Berkovic SF and Wilson SJ (2014). The genetic basis of music ability. *Frontiers in Psychology* 5:658. doi: 10.3389/fpsyg.2014.00658

Plans for 2015:

We will be promoting our study during the Twins Plus Festival in March 2015 to recruit more twins for our study. We have plans to publish a methodology article on techniques to facilitate the extraction of pitch data from the voice recordings of the participants. We expect to finish our recruitment and data analysis within this year, Ms Yi Ting Tan also aims to submit her PhD thesis by end of this year.

Once the study arising from this research has been completed, a brief summary of the findings will be available to the participants via email or disseminated through the ATR. The research findings will be broadly disseminated through the media and at local, national and international

scholarly and community forums. The results will also be published in high impact, peer-reviewed journals and as a PhD dissertation for Yi Ting Tan.

Recruitment to date:

We have recruited in total 100 complete twin pairs so far:

63 pairs of MZ (9 pairs male, 54 pairs female) and 30 pairs of DZ (6 pairs male, 24 pairs female and 7 pairs of male/female)

Age: 15-65yrs

States: all

What was sent: 6 online singing tasks and questionnaire

What they did with the twins:n/a

Feedback provided: pending

Current Status: ACTIVE – RECRUITING

2008-002-03 TYPE 2 DIABETES AND BRAIN FUNCTION: A CO-TWIN STUDY OF BRAIN METABOLISM

Principal Investigator

A/Prof Velandai Srikanth

Monash University

Melbourne, VIC

The original study (2008-002-1) aimed to establish if diabetes is associated with atrophy (neural loss and shrinkage) in the cerebral cortex, particularly the hippocampus, and thereby contributes to the risk of dementia. The study aimed to test this hypothesis in the setting of twin research by comparing the brain volumes and functional MRI during a memory task between twins who are discordant for Diabetes. By clarifying the effect of diabetes on the brain, such a study has the potential to open new avenues in dementia therapy. For example, drugs used in the control of Diabetes or its complications may become useful either to prevent or delay onset of dementia. This research directly addresses the National Research Priorities of Ageing Well, Ageing Productively, and Preventative Healthcare.

This study aims to understand how Type 2 Diabetes (T2DM) contributes to brain atrophy (neural loss and shrinkage) and thereby the risk of dementia. This study aims to test this hypothesis in the setting of twin research by comparing brain glucose use between twins who are discordant for T2DM. Greater understanding of the mechanisms in T2DM that contribute to dementia has the potential to guide measures to prevent and manage dementia. For example drugs used to control Diabetes or its complications may become useful to prevent or delay onset of dementia.

Participants receive feedback about their weight and blood glucose at the time of measurement. When data analysis and publication is complete, participants will receive written, de-identified feedback regarding the results.

In 2013 the ATR mailed out over 100 letters to discordant pairs. Measurements have been obtained for 4 twin pairs with another 4 twin pairs agreeable and to be measured. The study received recognition from the NHMRC receiving an \$860,000 grant to further research over the next five years - 2014–2019.

During 2014 the study continued with recruitment and has been exploring other avenues such as international collaboration to optimise participant numbers.

Number of pairs recruited: 21 new pairs (see below). Re-approached 14 previous pairs for more testing.

Zygosity: 7 x female DZ, 2 x male MZ, 5 x male/female, 5 x female MZ and 2 x male DZ

Male /Female: 5

Age: 65+

States: Individual stats: QLD X 7, NSW X 10, TAS X 4, WA X 12, VIC X 5, ACT X 3, SA X 2

What was sent: Approach letter, PLS and response form

What they did with the twins: MRI, cognition test, measurements, retinal scan and photo, skin ageing measurement, Health and Lifestyle Questionnaire, 24hr blood pressure measurement, 7 day physical activity measurement, PET scan

Feedback provided: Blood pressure and weight feedback

Plans for Jan 2015 – Dec 2015

Ongoing recruitment. Analyses of data collected to date

Feedback to be provided regarding health indicators as described in information and consent statement to be posted to individual participants when review of other data by relevant professionals is completed (i.e., check for any incidental findings that require follow up before providing general feedback).

Manuscript under review.

Current Status: ACTIVE – RECRUITING

2008-001

2008-001-2

2008-001-3

EFFECT OF MENOPAUSE ON THE STRUCTURE OF BONE BONE STRUCTURE AND STRENGTH DURING MENOPAUSE: A CO-TWIN CONTROL STUDY

Principal Investigator

Prof Ego Seeman

Austin & Repatriation Medical Centre

Melbourne, VIC

The heterogenous structural changes and biomechanical consequences of menopausal bone loss will be studied prospectively by measuring bone macro- and micro-structure using new technique of high-resolution pQCT, remodelling, and bone strength during 3 years in premenopausal, peri-menopausal and postmenopausal twins and in women with forearm fractures. The aims were completed, we have shown that structure of bone does predict bone re-modelling as stated in our hypothesis. The work was presented at the plenary oral ASBMR in 2009. The contribution of nutritional factors to bone loss and fragility will be analysed for the follow up visit.

Prior to 2013 the 2008-001 study recruited F/F DZ & MZ twins between 40-60yrs living in Melbourne to engage in a 10 minute phone questionnaire and have leg, wrist, hip and spine scans at Heidelberg Hospital taking approximately 45 minutes. Blood samples were also sought.

During 2013 the 2008-001-2 component of the study re-approached existing participants and approached new participants F/F DZ & MZ twins who are aged between 25-75 years of age

and living in Melbourne. The participants will be asked to fill in a 10 minute phone questionnaire and have leg, wrist, hip and spine scans at Heidelberg taking approximately 45mins as well as blood samples.

In 2013 the study recruited and scanned 36 singletons for their follow up visit (2008-001) and 108 singletons for their study visit (2008-001-2).

Bone density results are sent out to participants within 2-4 weeks after their visit. A letter is sent to further explain how to read the results.

2014

In 2014 the 2008-001-3 study will recruit and follow up with 120 singletons for their follow up visit. The researchers will conduct the mail out as the planned follow-up which the twins were notified about in the original invitation.

Number of pairs recruited: 42 individuals were re-approached

Zygosity: female MZ and DZ

Age:65+

States: VIC only

What was sent: Direct approach by researchers.

What they did with the twins: phone questionnaire, hospital visit; bone density test and measurements, blood sample

Feedback provided: Detailed measurements of bone structure and density; results explained by researchers.

Current Status: ACTIVE – RECRUITING

2004-001 TOOTH EMERGENCE AND ORAL HEALTH IN TWINS AND THEIR FAMILIES

Prof Grant Townsend

Dental School

The University Of Adelaide

Study Aims:

There are two main aims of this study. The first is to use multivariate modelling approaches to determine the relative contributions of genetic and environmental factors to variation in emergence of primary teeth in a large sample of twins. The second aim is to find out what the relationship is between the timing of emergence of the primary teeth and the colonisation of the oral cavity with mutans streptococci (MS), key organisms in the initiation of dental caries.

Significance to wider researcher area:

At present, little is known about how important genetic factors might be in human tooth eruption. Our study is the first large scale study in humans that specifically addresses this issue which is of fundamental biological and clinical importance. We are also studying the possible association between tooth emergence and oral colonisation with decay-producing bacteria, and also how these bacteria are transmitted between family members. The outcomes of these studies should have direct implications for oral health strategies. Knowledge of the timing of initial colonisation with mutans streptococci will provide important guidelines about the most effective period for intervention to reduce cariogenic bacteria and prevent early childhood caries. By studying transmission of MS between all family members, we will clarify the importance of paternal/ sibling transfer, in addition to the recognised role of maternal transfer. This may lead to a broadening of the preventive strategy for reducing caries risk in children.

Additional information: By studying the composition and abundance of bacteria present in the mouth and linking these together with dental decay status, the epigenetic profile of the individual and environmental information on health and diet, we are aiming to obtain a better understanding of how and why oral health and oral disease develops.

Achievements in the period Jan 2014 – Dec 2014:

As part of the new sub-contract for the JCVI with the Early Life Epigenetics Group (PETS cohort) at the MCRI, and also as part of the new NHMRC project grant awarded to Adler et al, we have conducted clinical sessions during 2014 in Adelaide and Sydney focusing on assessment of dental decay and the collection of oral microbiological samples as a means of assessing caries development. During these sessions we have also taken various anthropometric measurements, dermatoglyphs and tested functional laterality. We have continued to collect data on tooth exfoliation timing and the exfoliated baby teeth. Our book about our Twin Studies has undergone peer review and is now awaiting publication.

2014

Direct researcher approach

Plans for Jan 2015 – Dec 2015:

We are continuing clinical assessment of dental decay and collection of microbiological samples in twins from South Australia, New South Wales, Australian Capital Territory and Victoria. We will also take various anthropometric measurements, dermatoglyphs and test functional laterality at these sessions. We intend to distribute questionnaires on dental health, general health and development, and diet which will supplement the clinical data. We will continue collection of tooth exfoliation dates and the exfoliated teeth. Our book currently titled “Twin Studies: Research in Genes, Teeth and Faces” will be published in mid-2015.

2007-004 ROLE OF GENETIC AND ENVIRONMENTAL FACTORS IN ATRIAL FIBRILLATION

Prof Diane Fatkin

Sr Bernice Research Programme

Molecular Cardiology Division

Victor Chang Cardiac Research Institute

1. To collect clinical details and blood samples from a population of monozygotic (MZ) and dizygotic (DZ) twin pairs in which one or both twins has AF.
2. To determine the heritability of AF using a population of MZ and DZ twins
3. To identify and characterize disease-susceptibility polymorphisms

The aim of our research study is to better define the genetic changes that can predispose to AF, a disorder associated with irregular electrical activity of the atria (upper chambers of the heart) and to look at interactions between genes and acquired (environmental) factors that result in dilatation of the atria and increased AF risk.

Achievements in the period Jan 2014 – Dec 2014:

No new twin pairs have been recruited.

We are continuing to collect outstanding clinical data as well as blood samples from all participants.

Our collaborator, Dr Katrina Scurrah, is currently investigating appropriate methods to undertake the heritability analysis.

93-011 LONGITUDINAL STUDY OF MELANOCYTIC NAEVI IN TWINS (Mole Development in Pubescent Twins)

Nick Martin

Epidemiology Unit

Queensland Institute Of Medical Research

Royal Brisbane Hospital

Investigating mole development in pubescent twins

Achievements in the period Jan 2014 – Dec 2014:

Participants tested for the period 01/01/14 – 31/12/14:

ZYGTEXT	FST_VIS	S	SEC_VIS
DZF	23	20	
DZFM	24	4	
DZM	22	12	
DZMF	14	12	
MZF	37	24	
MZM	20	16	
SIBLINGS	21	0	
TOTAL	163	88	

Direct researcher approach

We do send a newsletter (biannually) giving general updates and feedback on the project as a whole. The latest newsletter was distributed in December 2014/January 2015. There is also a website available to twins where they can access updates and publications etc. <http://www.genepi/qimr.edu.au/>

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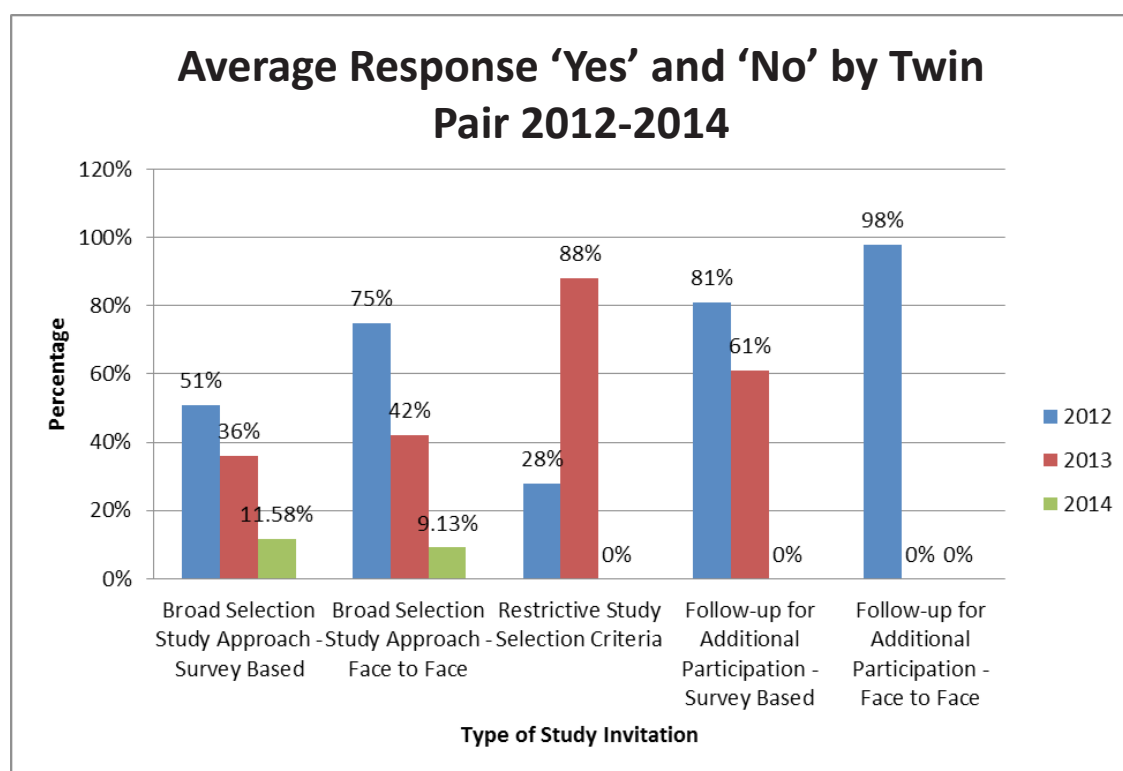
Appendix 4: ATR Response Rates in Active Studies 2012 – 2014

Appendix 4 provides further breakdown of the response rates for various modes of response, sex and age between 2012 and 2014. Response rates to Registry mailouts appear to be higher for those studies approaching either families with young twins (under 18 years old) or older, adult twins (40 years and older). Twins between the ages of 18 and 40 years have the highest 'NR (*Nil Response obtained*)' and 'Negative' RR and are the most difficult group for which to maintain current contact information. 'Negative' RR also include twins who are ineligible to participate in a study based on the criteria set by researchers, for example, where the member does not display a particular trait or does/does not suffer from a particular disease.

1. Study Response Rate - Yes/No Response Rate by Twin Pair

Figure 1 shows an average pair response rate for each year for studies with mailouts from 2012-2014. The twins must have responded Yes or a Hard No. The study response rates illustrate the average response rates for each year, 2012 – 2014. The positive strong 'yes'/'no' response reflects the engagement of the ATR membership.

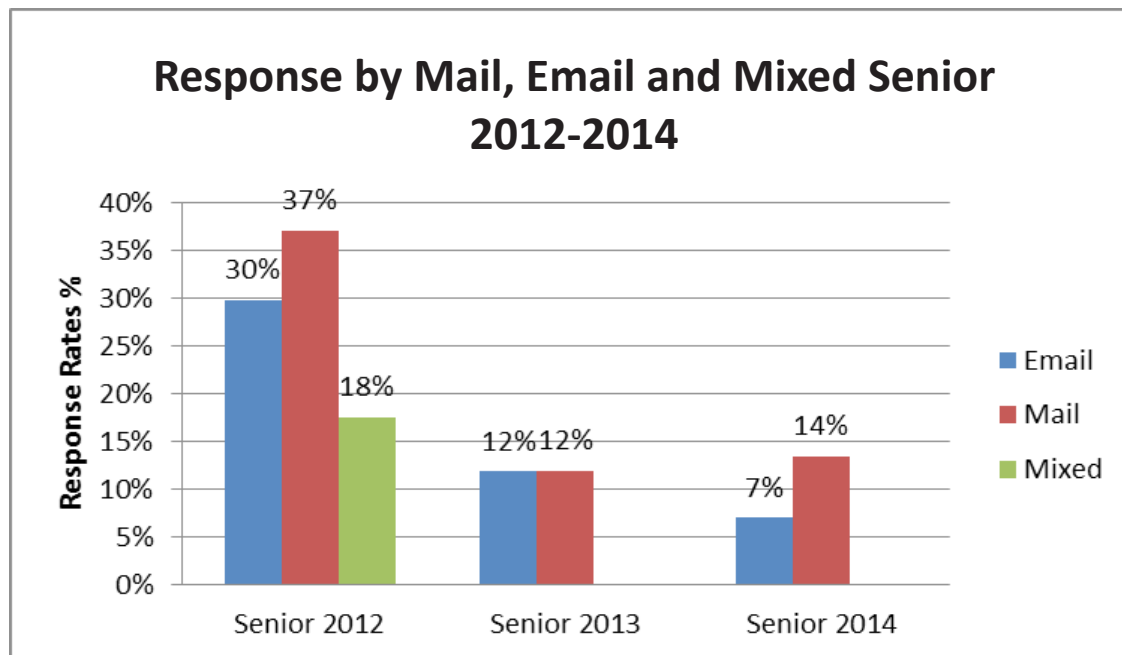
Figure 1: Study Response Rate – Yes/No Response by Twin Pair 2012-2014



2. Study Response – Twins who Responded Positively – Age Group, Mail and Email

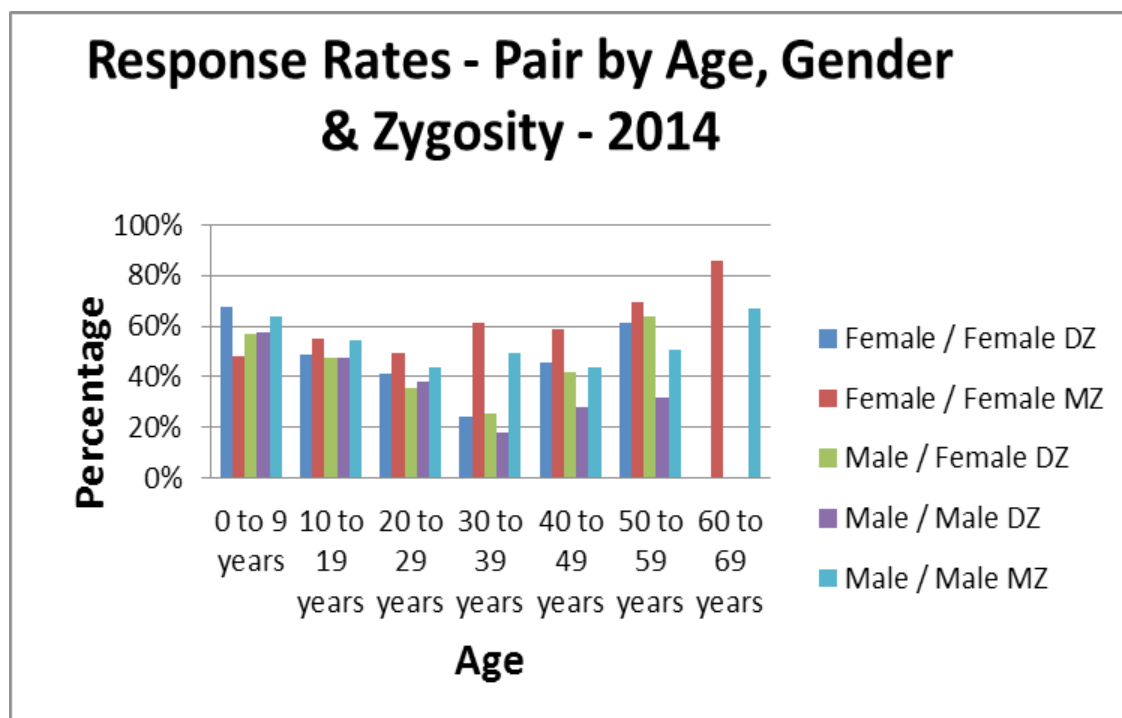
This figure shows a positive pair response rate by year with a breakdown by age and method of approach. There were no figures for junior pairs in email or mail response modes as parents will often respond for the twin pair, although it should be noted that the average for mail out responses from the junior group is 30-40%, comparable to the senior response rate. **Figure 2** below takes into account senior twin pairs who responded via mail or email during 2012-2014. Email response rates for senior twin pairs appear to have declined which may be due to the ageing population of senior twins. Phone is a preferred method of contact for seniors.

Figure 2: Study Response Rate who Responded Positively by Age, Mail, Email and Mixed



3. Study Response – Pairwise – Responded – Age Group Gender Type

This figure shows a pair response rate broken down by age groups, gender type and zygosity for studies with mailouts 2012-2014. The twins must have responded Yes or a Hard No. **Figure 4** only includes studies which had general selection criteria and were first time approaches, in that, studies were excluded that re-approached already participating twins for further involvement. Generally, the 0-9 and 10-19 year age groups are most responsive, across zygosity/ sex groups; response rates then typically increase again from 40 years upwards.



Response Rates - Pair by Age, Gender & Zygosity - 2014

