



# 2013 ANNUAL REPORT



Bringing **twins** and **researchers** together for vital health research that benefits everyone.



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# 2013 Highlights

Bringing **twins** and **researchers** together for vital health research that benefits everyone.



## INTERNATIONAL PATRON

January HRH Crown Princess of Mary of Denmark becomes ATR International Patron

In October our Patron met with ATR researchers and members at Sydney Children's Hospital

## Communications



134+ hits in the media - nationally and internationally - covering diverse topics such as the Patron announcement, singing study, Patron visit to Sydney and Tim Spector forum



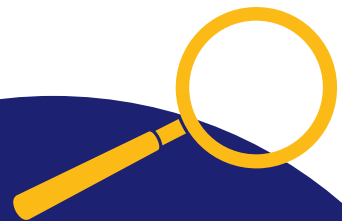
## Membership in 2013

37,669 twin pairs to date

2262 new twin and triplet sets

28,377 member records were updated

10,775 members approached to participate in studies



## RESEARCH

Our research covers a broad spectrum of health issues studied - growing pains, bone health, epilepsy, singing ability:

13 studies required twin participants

19,238 reminders were sent to study participants

60 publications, 4 book chapters and 25 abstracts were published

## EXECUTIVE SUMMARY

### Welcome to the 2013 Annual Report of the Australian Twin Registry (ATR)

This report summarizes the major achievements, activities and research developments of the Registry in the past year.

The 2013 reporting period coincides with the fourth year of the renewed Enabling Grant (2010 – 2014) from the National Health and Medical Research Council (NHMRC) that funds the ATR.

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 2013 the ATR made significant progress towards achieving this vision. Following below is a summary of the 2013 highlights and achievements:

#### Highlights and Achievements for 2013

- The ATR announced an international Patron, HRH Crown Princess Mary in January 2013;
- ATR Membership numbers grew, exceeding the goal set in the Enabling Grant, with 2,262 new sets of twins and triplets registered in 2013.
- The ATR recruited for 11 studies in 2013, 3 of which were completely new, looking at health issues from growing pains, bone health and mental wellbeing. This resulted in 10,775 approaches to ATR members to participate in research.
- The research benefits enabled by the ATR were significant, with 60 peer-reviewed articles based on ATR-related studies published, as well as 4 book chapters and 25 abstracts and posters.
- Throughout the year the ATR has had enormous success in reaching out to the wider community with research messages leveraging off our high profile patron, Princess Mary, and providing unique media stories involving twin research. In 2013 the ATR gained significant media exposure with 134 mentions in the media.
- In 2013, 28,377 twin and other contacts (i.e. parent or secondary carer) records were updated. The ATR maintained accurate information on 92.3% of the members.
- The ATR sponsored and participated in the 2013 21<sup>st</sup> Convention of AMBA, which was held in Canberra on the 25<sup>th</sup> - 26<sup>th</sup> October 2013. The ATR Club Initiative Support Grant continued to support AMBA clubs around the country.
- The Travel Grant Scheme supported a total of 17 early-career to attend relevant national and international research workshops and conferences relating to twin research.
- Establishing the “Friends of the ATR”, a group of members that wish to participate more actively in the ATR with assistance in study feedback, volunteering in twin activities’ such as festivals, media events and fund raising opportunities;
- Distributing the 2013 Newsletter to 53,300 members by post and 5,113 members by email, whilst running an i-Pad competition to encourage members to update their address details;
- Providing an information session on Twin Research by Tim Spector, a leading international twin researcher and epigeneticist.
- The ATR reviewed and enhanced a range and quality of services that it currently provides to the research and twin community, optimising the ATR's ability to



support research that has the potential to contribute to the health and wellbeing of Australians. This was achieved by:

- Monitoring our services to current researchers with a Quality Assurance Survey collecting feedback over 3 stages of the life span of the study;
- 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;
- Developing a Health and Lifestyle Questionnaire for our members to complete so as to better target twins suitable for the researcher's areas of interest (to be completed in 2013);
- Providing a comprehensive "Working with the ATR" Guidelines and Data Transfer Agreement. This will request the return of all study data for the purpose of enabling future ethically approved studies; and
- Continuing to lead the initiative to be part of an International Network of Twin Registries.



ATR International Patron Launch: Director of the ATR, Professor John Hopper presenting twin platypuses to the ATR International Patron, HRH Crown Princess Mary of Denmark in Odense, Denmark, January 2013.

Overall, 2013 has been a year of great success for the ATR in research, funding and Registry growth. The outlook for 2014 promises further progress in all these areas.

## Outlook for 2014

The ATR will continue to focus on strengthening its relationships with both the research and twin communities and by doing so assist with the translation of research knowledge to society.

This will be achieved in the research community by:

- Implementing the Health and Lifestyle Questionnaire for all ATR members and making the data available for future studies;
- Implementing the collection and archiving of research data into the Ark;
- Supporting the establishment of a Brazilian Twin Registry; and
- Responding as needed to feedback from Quality Assurance Surveys.

This will be achieved in the twin community by:

- Raising the profile of the ATR through the International Patron, HRH Crown Princess Mary;
- Establishing the “Friends of the ATR”, a group of members that wish to participate more actively in the ATR with assistance in study feedback, volunteering in twin activities’ such as festivals, media events and fund raising opportunities; and
- Providing an information workshop on Twin Research by leading international twin researchers in December 2014.

A key strategic priority for the ATR will be to develop a funding strategy that seeks to secure funding streams to sustain the ATR and its operations beyond the term of the existing funding grant (2014). This initiative is of utmost importance to ensure that the Australian research community has efficient and reliable access to the benefits available from the utilisation of twins in research, as enabled by the ATR.



## ATR OVERVIEW

### *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 2013, the ATR updated information on 28,377 sets of twins and triplets, and supported 80 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](#)). ATR's significant national and international media coverage helped raise the profile of twin research in the wider community.

### *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 the many health and medical issues that affect Australians. Twin studies have started to play a vital role in the emerging search for epigenetic effects. Such epigenetic effects have been linked to 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 their shared environments and genetics.



ATR Researcher Professor Sam Berkovic with ATR members Amanda and Nicole Campbell undertaking tests for Epilepsy Research Study

## ***Funding***

The ATR is supported by an Australian NHMRC Enabling Grant. The grant covers a 5-year period, *1 January 2010 - 31 December 2014*.

This enables the ATR to be a national resource to all researchers within Australia and worldwide.



Carla and Mia Lubbock at the ATR International Patron Launch in Melbourne, January 2013



## 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*

### Database

The ATR maintains an up-to-date register of twins 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 meaningful 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.

As at 31 December 2013, the database held data on 76,136 individuals representing 37,669 twin pairs and 399 HOMs (total number of 38,608 sets).

In 2013, 28,377 twin and other contacts (i.e. parent or secondary carer) records were updated. There were 10,775 study approaches to ATR members to participate in research. The ATR maintained accurate information on 92.3% of the members.

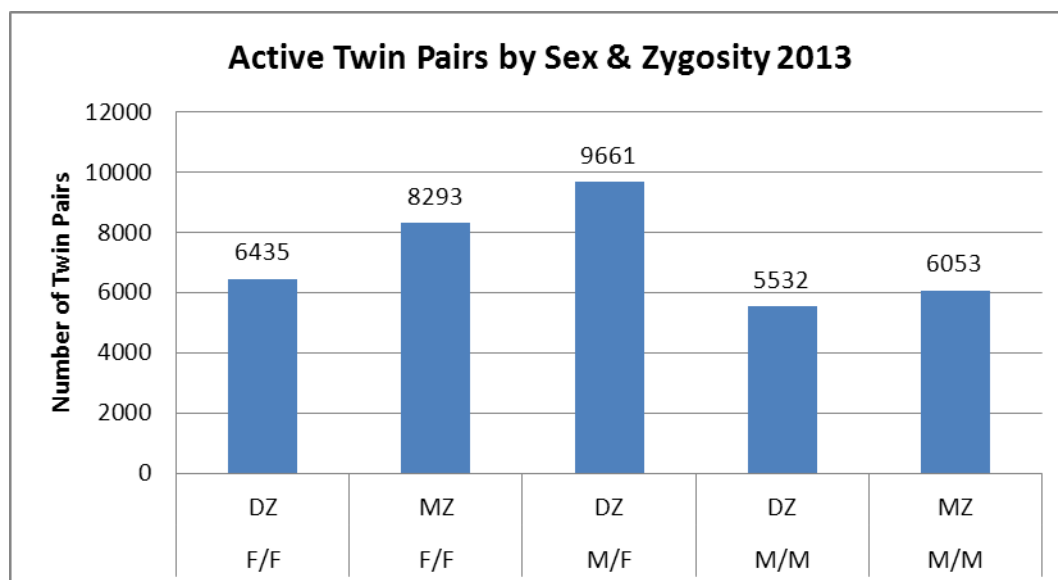
Members of the ATR are recorded under a specific status, depending on the currency of their contact details and individual preference for involvement in research activities. The majority of members enrolled in the period covered by this report (99%) are categorised as Active/Active, Active/Questionnaire and Questionnaire/Questionnaire pairs, indicating that they are willing to consider participating in research. The current status of members of the ATR is summarized in **Table 1**. A total of 70.4% twin pairs have both members active, and an additional 5.35% of members' contact details require updating (recorded as Pending). Junior members represent approximately (28%) of the entire registry, the remaining (72%) being senior members.

**Table 1:** Twin Pair Status Combination as of 31 December 2013. *Top row details T1 (twin one) status, and left most column details T2 (twin two) status. OS identifies twin members who have moved overseas, but are still available for electronic surveys.*

T1/T2 Status	Active	Deceased	Lost	Questionnaire	Newsletter	Inactive	OS	Pending	Total
Active	28075								28075
Deceased	767	585							1352
Lost (O/S)	1	0	750						751
Questionnaire	656	11	0	138					805
Newsletter	181	51	0	3	104				339
Lost	167	16	0	1	6	29			219
Inactive	881	468	0	4	35	1560			2948
OS	180	2	0	2	2	6	61		253
Pending	2564	116	54	52	33	164	21	2132	5136
	33472	1249	804	200	180	1759	82	2132	39878

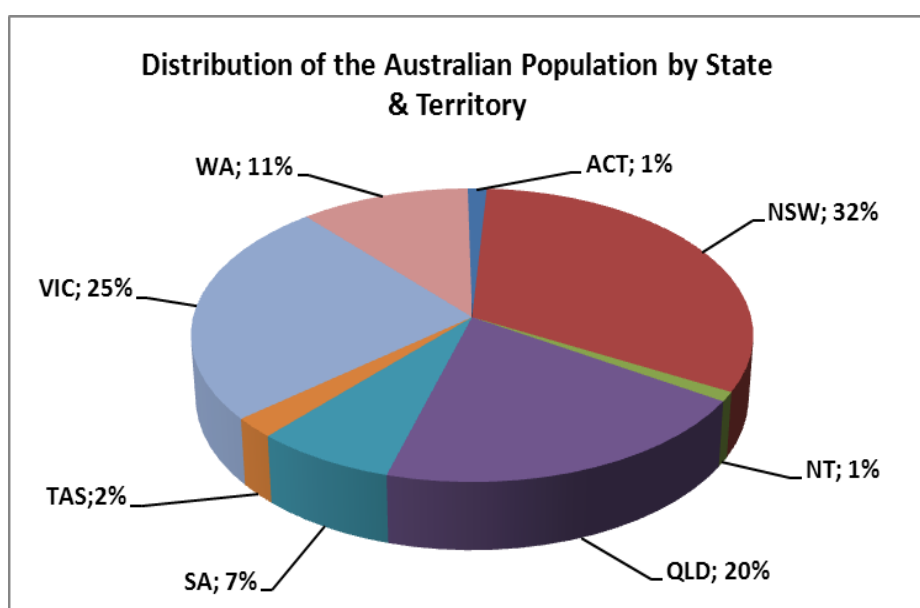
Currently, one third of the active ATR members are junior (0-19 years) and the rest of two thirds are adult. The current numbers of active twin pairs by sex and zygosity are shown in **Figure 1**. It does not show the 'lost' of 'unknown' twin pairs of which there are 1,819 pairs.

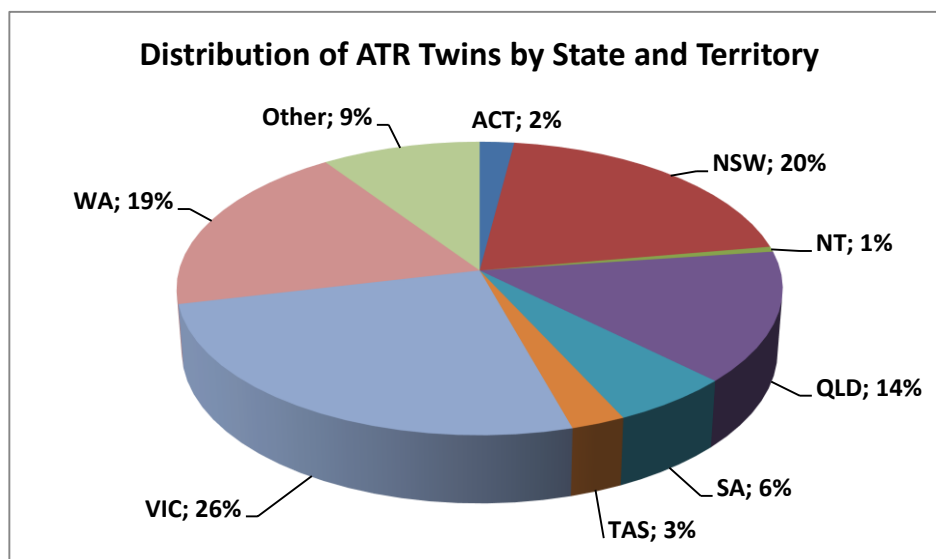
**Figure 1.** Active Twin Pairs by Sex and Zygosity as at 31 December 2013.



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 2012). Comparison of the two graphs shows that most populated states, New South Wales, Victoria and Queensland, are the same ones where most active ATR members reside.

**Figure 2.** Active Twin Pairs Combination by Location as at 31 December 2013 (**Top**) and overall distribution of Australians by State and Territory (**Bottom**). *Note: because some twins live in separate states or one twin member in a pair lives overseas, this figure captures only 26,852 of the 28,075 shown as Active/Active in **Table 1**.*

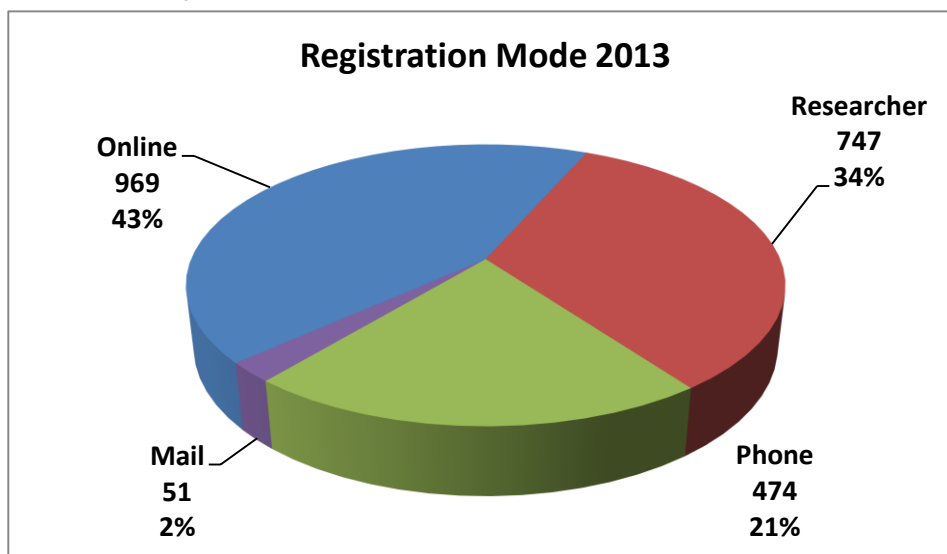




## Recruitment

Continuous recruitment of new twin members is vital to ensure the future viability of the ATR. The ATR's goal is to increase membership by more than 5,000 over 5 years. In the reporting period, 1 January to 31 December 2013, the ATR added membership details on 2,209 sets of twins and 53 sets of higher order multiples (homs)<sup>1</sup>, for a total of 2,262 sets, which exceeded the ATR's goal of recruiting 1000 members annually. The majority of new members (969 of the total 2,262 sets; 43%) were completed online (**Figure 3**), 34% by researcher involvement, 21% by phone and 2% via mail.

**Figure 3.** Mode of new registrations (twin and triplet sets) in 2013.



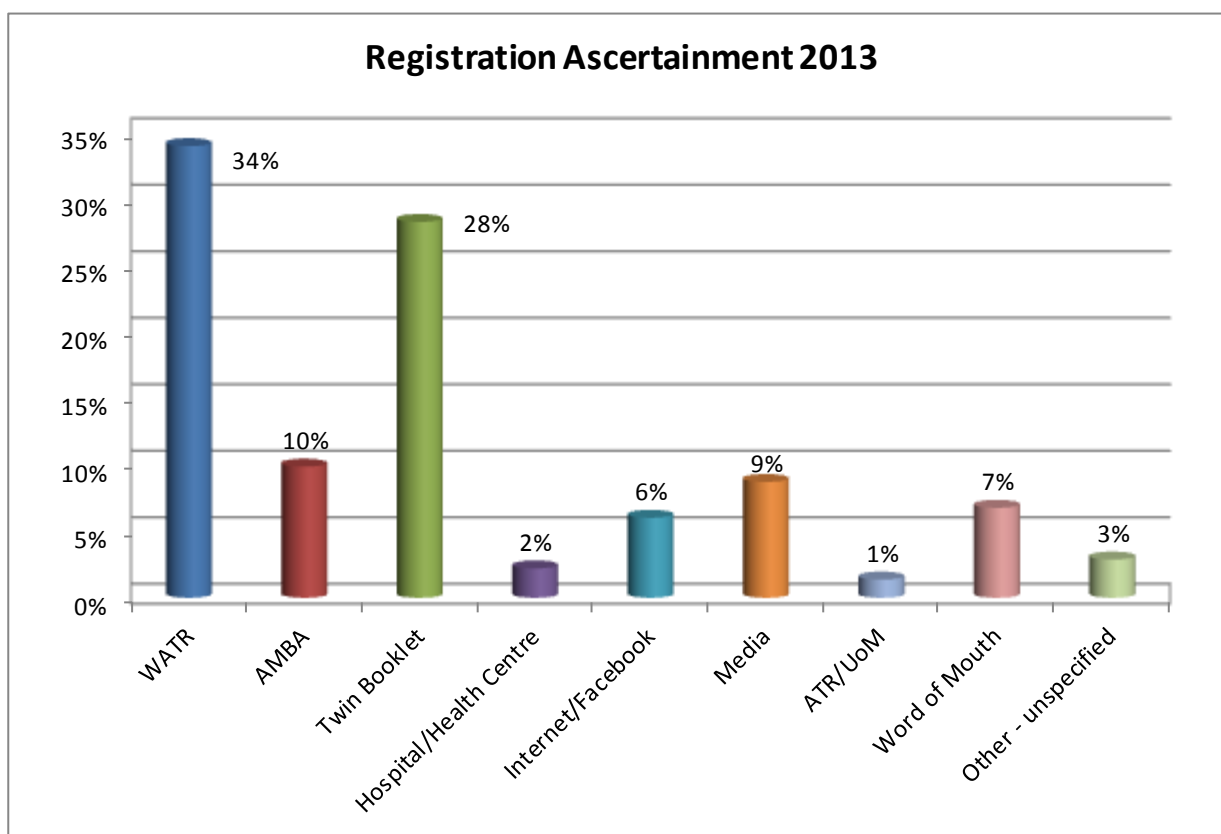
While online registrations were more than double our phone registrations; 969 vs 474, the booklet conversions would most likely account for a fair proportion of the phone registrations. Many booklet requests are online – but the follow up and eventual conversion is often via phone.

New registrations to the ATR are asked 'Where have you heard about the ATR?' From this we can ascertain that 34% of the 2,262 new registrations were from the Western Australia Twin Registry, while Other-Unspecified is 3% (**Figure 4**).

<sup>1</sup> Homs – higher order multiples refers to triplets, quadruplets and quintuplets.

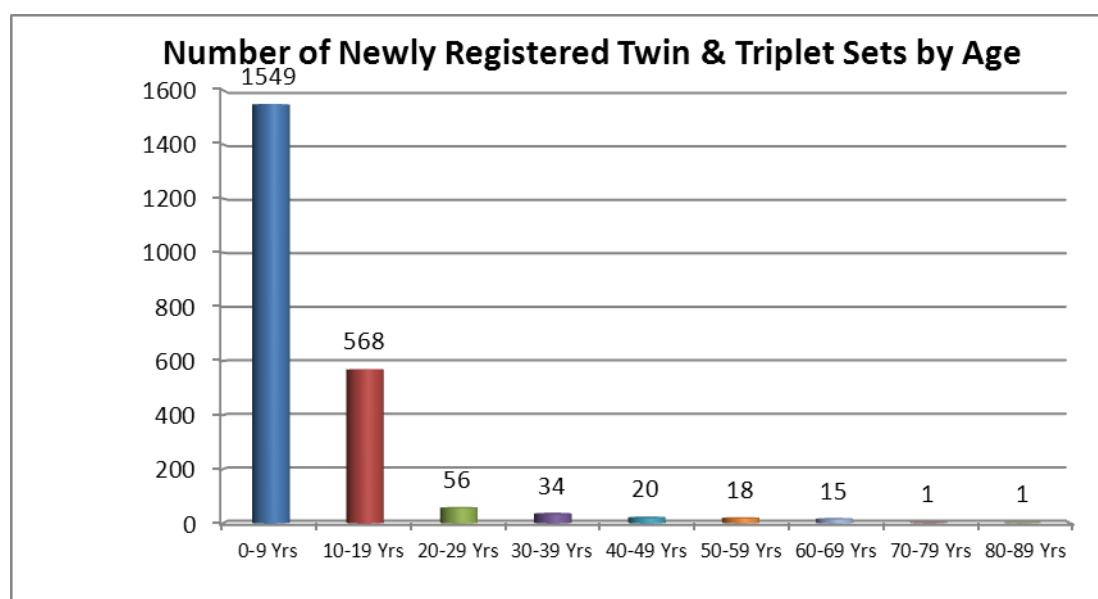


**Figure 4.** New registrations by Ascertainment (twin and triplet sets) in 2013.



Every effort is made to ascertain where new registrations to the ATR are from, as this informs where the ATR should focus future promotion. The remaining proportion is due, in part, to the focus on promotion of the ATR through the twin pregnancy booklet provided to parents in hospital (28%), AMBA (10%), media (9%), word of mouth (7%), internet/ Facebook (6%), hospital and maternal health centres (2%) and ATR/University of Melbourne (1%).

**Figure 5.** Number of new active twin and triplet sets registered in 2013 shown by age range.

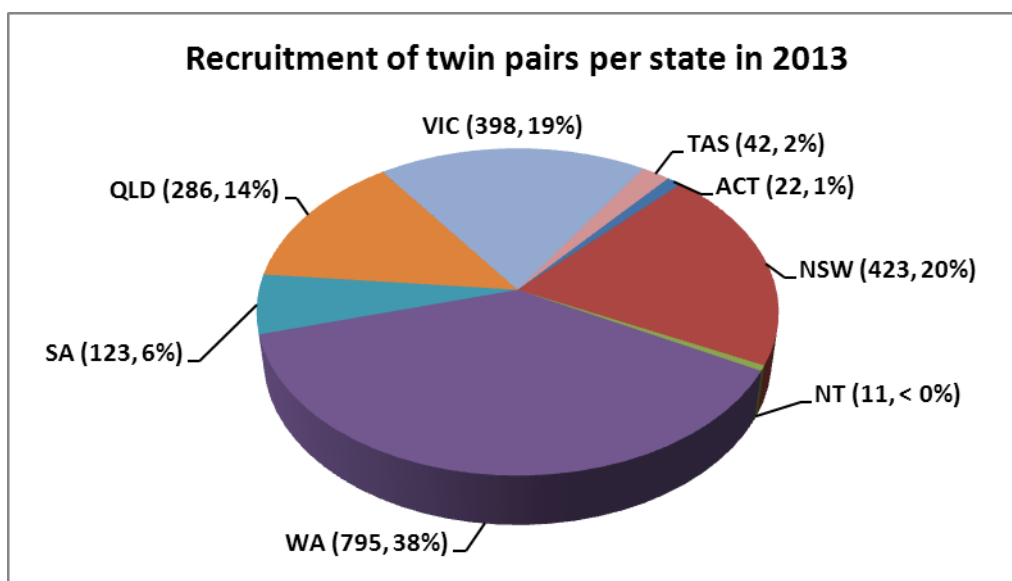


The majority of new members (68%) enrolling with the ATR during the reporting period were aged 0-9 years (**Figure 5**). This is consistent with the previous 20 years, where the 0-9 years have been the relatively consistent majority of new members. The 10-19 year group follows with

25%. The final transfer of twins and triplets from the Western Australian Twin Registry occurred in 2013. An increased focus on the promotion of the ATR could also have played a factor in the increase of this age bracket. The 2013 figures see the combined senior group, or 20-99 year age, represent the combined figure of 145 new registrations.

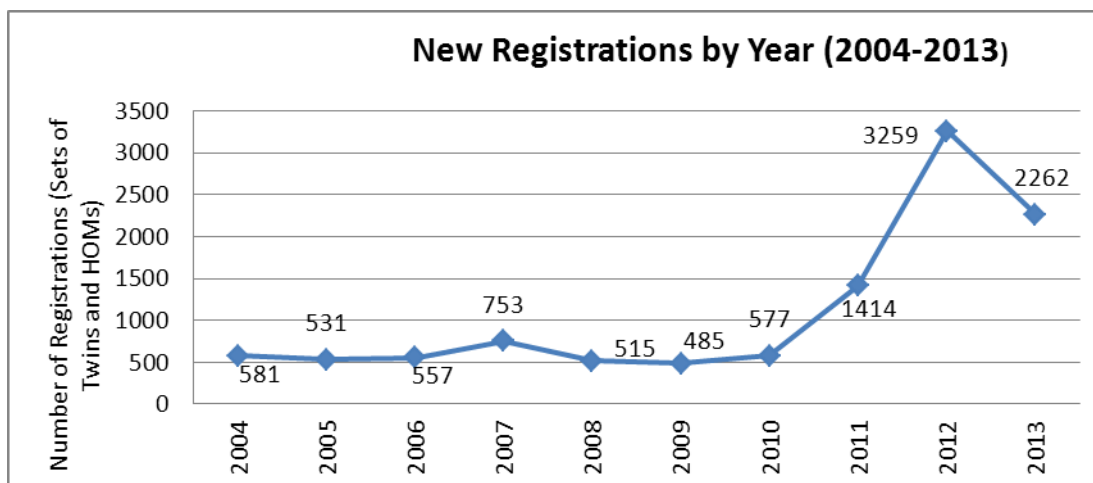
Compared with previous years, when most new registrants came from Victoria and New South Wales, in agreement with the overall population distribution in Australia, the collaboration between ATR and the Western Australian Twin Registry resulted in an increased number of new members from this state in 2013. Thus, most new members came from Western Australia (38%), followed by New South Wales (20%), Victoria (19%) and Queensland (14%) (**Figure 6**). Registrations from the remaining states were all under 10% with South Australia (6%), followed by Tasmania (2%), the ACT (1%) and the smallest proportion of registrations coming from the Northern Territory (<0%).

**Figure 6.** Distribution of active twin pairs, by State or Territory, recruited during the reporting period in 2013. *Note: numbers of triplet sets are not included in the graph.*



The numbers of new registrations by year since 2004 are represented in **Figure 7**. The drop in registrations in 2013 was due to a larger proportion of new registrations that came from the Western Australian Twin Registry in 2012. The greater number of new registrations came from Victoria or New South Wales as in the past.

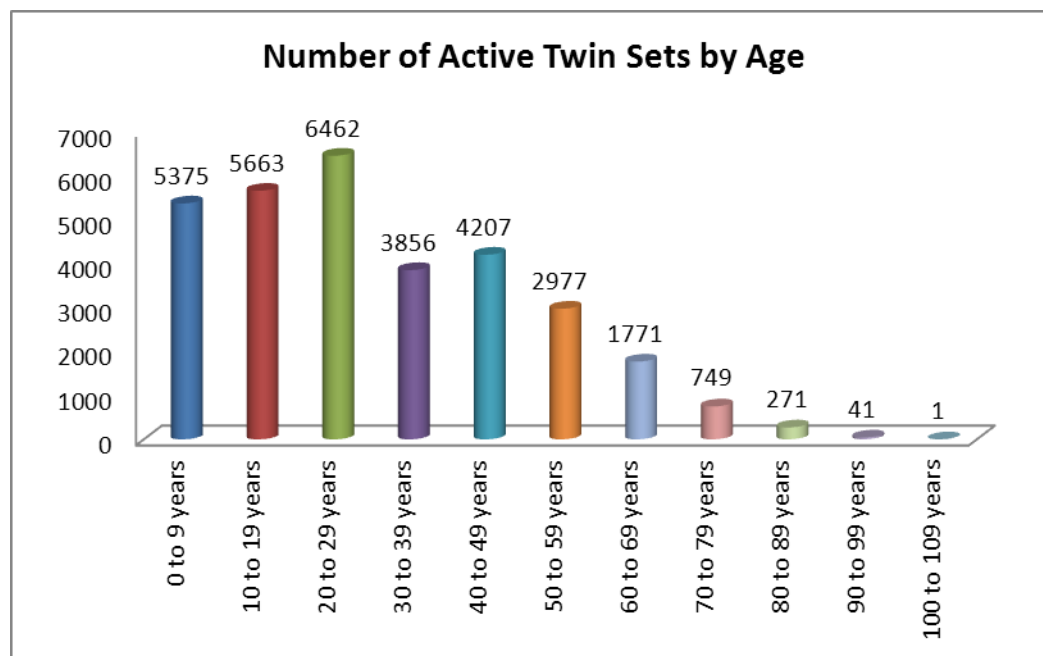
**Figure 7.** Number of new twin and HOM sets registered with ATR by year since 2004.



The distribution of active and lost pairs of twins, across the Registry, is shown relative to the age groups of members in **Figure 8**. As stated above, the anomaly with the 20-29 year group is the WATR members. 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 8.** Active twin pairs shown by age group, as at 31 December 2013.

*Note: data on triplets are not included in the graph.*



### ***Currency and Accuracy of Membership Data***

The goal of the ATR, as outlined in the NHMRC Enabling Grant, is to maintain current information for at least 85% of the membership. This acknowledges that a proportion of twin members who have moved require tracing to update contact details.

Although the *Twins* Newsletter is first and foremost a tool for communication with members, it also presents an opportunity to assess the accuracy of member address data. Using the Newsletter and other communication methods, during 2013, the Registry received 3,048 Return to Sender (RTS) envelopes and 4,182 Replied Paid Envelopes (RPE), of which 2,707 and 1,253 were as a result of the sending out the printed ATR Newsletter, distributed to over 53,000 members. Mailed study approaches also provide an opportunity to verify members address details. This response is vitally important to the ATR, alerting us to members who have moved. Upon receiving the RTS, the ATR initiates the tracing of relocated members through a range of tools, such as the White Pages, as well as following up on the second or third contacts provided by the ATR members. At the end of 2013, the Registry maintained current information on 92.3% of the members (only 5.35% of members' contact details required updating and are listed as "pending" in **Table 1**).

## **ATR Website**

The ATR continues to improve the face of the ATR through the website, which provides up-to-date and current material for major stakeholders, the twin and research community. The most popular pages are 'facts and figures', followed by 'frequently asked questions' on the resource page, twin services and update your contact details.



ATR members sent in photos for the iPad Competition 2013. From top left: Alita, Loki and Michael; Chloe and Nina; Hayley, Joanne, Kelsey and Lyndon. From bottom left: Mia, Lily and Chloe; Pat and Will; Yakira and Elena.

The ATR continues to be active in social media with 280 'likes' a month on Facebook as a means of engaging members on a more informal and current platform, with a total of 4751 as of the end of 2013. In late April 2012 the ATR joined Twitter and had 225 followers at the end of the year. By the end of 2013 the ATR has over 413 Twitter followers and 5 new 'likes' a week, which normally increases after the e-News is published online. The Twitter is linked to the Facebook page, so all Facebook posts are shared to Twitter. Facebook and Twitter keeps members up-to-date with the latest on twin health and research, and enable twins to connect with each other. This allows the ATR a more immediate means of disseminating and sharing information about the multiple community and calls for participants in research studies.

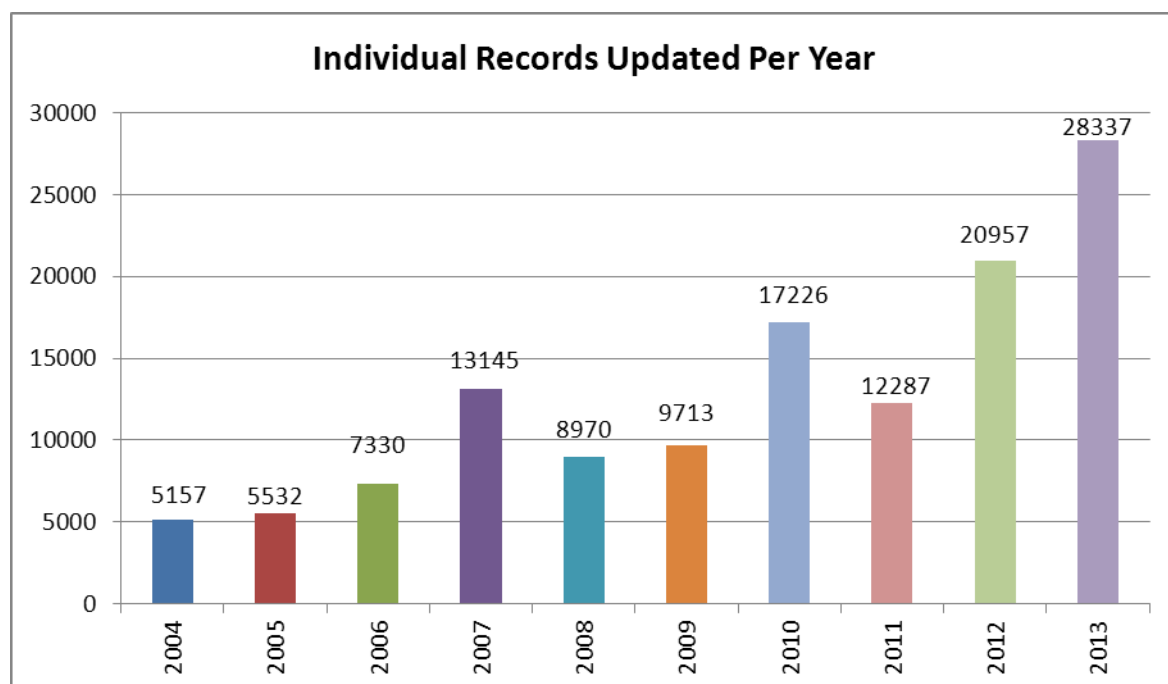
## **Record Updates**

The ATR is aware that not all misdirected mail is Returned to Sender, and as such, 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 2013, a total of 28,377 individual twin and other contacts member records were updated in the ATR database. A count of all individual records updated yearly since 2004 is shown in **Figure 9**. The 2013 spike was due to an iPad competition in the Newsletter, eligible to members who updated their address details.

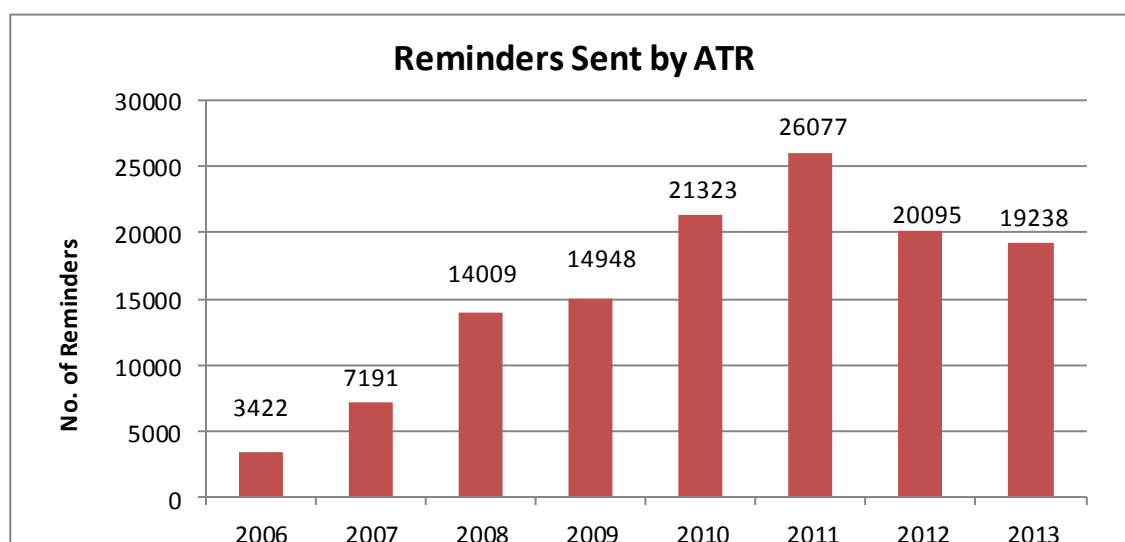


**Figure 9.** Numbers of individual records updated per year (2004 - 2013).



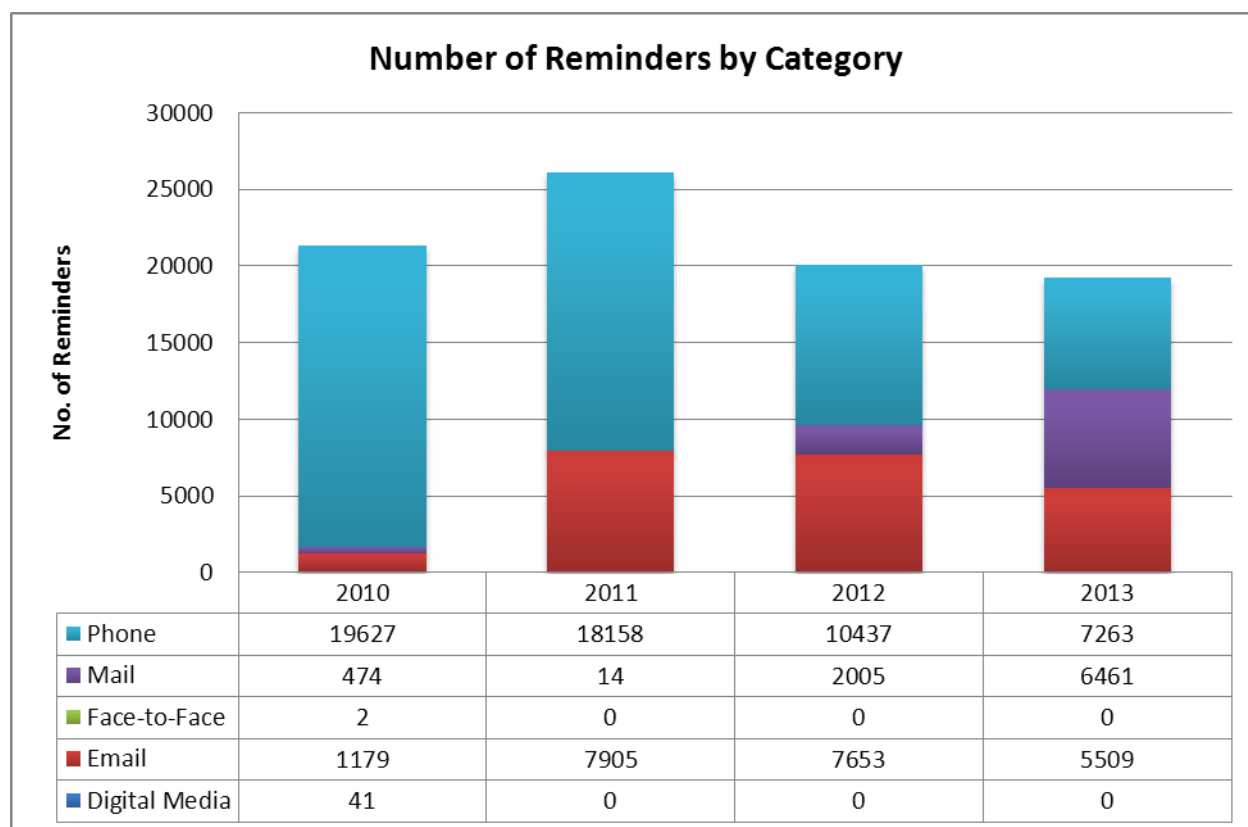
Telephone follow-up for studies remains a major tool for reaching high numbers of updated records. **Figure 10** illustrates the number of reminders and follow-ups being generated by ATR staff in the past seven years. There has been a decline in phone reminders in 2013 partly due to less study invitations being sent as in previous years and rising costs associated with study engagement.

**Figure 10.** Numbers of reminders sent by the ATR staff (2006 – 2013).



Of the 19,239 reminders sent by the ATR, less than half (38%, 7263) of all reminders are undertaken by telephone (**Figure 11**), which although is a labour-intensive and costly exercise, is of great benefit as it provides a personal touch with the ATR members and also increases response to studies facilitated by the ATR. The remaining reminders are through mail (34%, 6461) and e-mail (28%, 5509).

**Figure 11.** Number of reminders by category (phone, mail and email) 2010-2013.



**Figure 11** highlights the steady increase in email use as a tool to send reminders to ATR members, a declining number of phone reminders and an increase in the mail reminders. 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. Thus, ATR is working to use both communication methods and to increase recorded email addresses, so that researchers and the ATR have this option of both communication tools.

## **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 E-news to researchers. In 2013 the ATR aimed to raise awareness and increase support in the general community for the role twins play in research.

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

In 2013 the ATR focused on increasing exposure in the media through the promotion of particular research projects. During the 12 months there were 135 in press articles in newspapers, magazines, TV, radio and other media including online. The majority of the media coverage relates to the ATR International Patron launch, the Singing Ability study, the ATR Research event held in Sydney with the ATR Patron and the Tim Spector forum (see below). Refer to [Appendix 2](#) – In Press Articles for all of the ATR media coverage during 2013.

### **ATR International Patron Launch**

Her Royal Highness Crown Princess Mary of Denmark agreed to become the international patron of the Australian Twin Registry as well as the Danish Twin Registry. The ATR celebrated the announcement on both sides of the world – in Australia and Denmark. The ATR was successful in securing a high profile international patron, HRH Princess Mary of Denmark, in January 2013. As a mother of twins – HRH Princess Josephine and HRH Prince Vincent - and with her connections to both countries, the Crown Princess is the ideal patron of the two (twin) registries.



ATR International Patron Launch January 2013. Left: ATR International Patron, HRH Crown Princess Mary of Denmark, Danish Twin Registry Director, Professor Kaare Christensen and ATR Director, Professor John Hopper

The ATR Director, Professor Hopper, met with the Crown Princess on 15 January at a twin research symposium in Odense, Denmark where she undertook her first official engagement in her new role. Professor Hopper presented the Patron with 'Aussie' gifts for the Princess's twins which had special significance – Healesville Sanctuary near Melbourne is undertaking a special twin study of platypuses as part of its breeding program (photo in **Executive Summary**). The sanctuary has found that twin platypuses, also called a puggle, are common in the wild but uncommon in places of captivity such as zoos - and it would like to understand why.



The ATR International Patron launch in Odense Denmark and Melbourne Australia, January 2013 with the ATR International Patron and twin boys from Denmark and Luke and Rohan Whitby from Australia.

It seems we can learn much from twins in the animal as well as the human world. These twin platypuses have a special relationship with the ATR – hence the choice of gifts for the princess's twins. The Crown Princess is excited to be supporting twin research and was amazed by the extent of twin research happening around the world and is looking forward to supporting our work.

Meanwhile in Melbourne, the ATR's head office highlighted twin research and twins at a special media launch on 16 January, whilst the Melbourne event was attended by over 50-60 twins and associates, mixed cohorts of adults and children.



The ATR International Patron launch in Melbourne with ATR members, January 2013

Professor John Hopper said the Australian Twin Registry was “incredibly excited” to have the support of the Crown Princess. “We are very grateful for the Crown Princess’s patronage as we hope it will raise awareness of the importance of twin research and of the wonderful contribution of twins and researchers whose efforts go largely unheralded. Through her patronage, the Crown Princess very much recognizes the great contribution of our nearly 80,000 volunteer twin members - of all ages, identical and non-identical - across Australia who have registered their interest to participate in twin studies. Our vital work could not continue without their support,” Professor Hopper said.

This announcement generated much media interest with coverage in Denmark and other places around the world, and on all [TV channels](#) and print media in Australia. This resulted in 183 new registrations over the two weeks after the patron launch - the ATR normally receives 20 new registrations per week.



## Research and the Media

### ATR Singing Study Launch

An innovative world-first research study, launched in Melbourne on May 9<sup>th</sup> 2013, will investigate if singing ability is inherited in twins. Led by the University of Melbourne with the Melbourne Conservatorium of Music and the Australian Twin Registry, the study will explore if it is nature or nurture that most influences whether we can sing a pitch-perfect tune.

While much research has investigated the influence of environmental factors such as exposure to music on singing ability, this study aims to investigate the genetic factors at play focusing on twins. The findings from the study may help us better understand how singing is best taught.

Lead researcher Associate Professor Sarah Wilson from the University of Melbourne's School of Psychological Sciences said singing has been a part of human existence throughout history and yet the abilities of individuals vary widely.

"Today singing, and musical ability and appreciation are an integral part of education. We hope this study will contribute to the development of more sophisticated classroom singing programs that take into account differences between individuals and provide the best possible experience," she said.



The singing ability in twins study launch in May 2013. Top Left: A/Prof Sarah Wilson, ATR twin siblings Daniel and Matthew Thomson and Yasmin and Yolanda Absolom, with Yin Ting Tan.

The launch of the singing ability study received the 2<sup>nd</sup> highest hit in TV, radio, print and online (436 out of 3842 hits in 2013) for coverage of the singing ability research within the Faculty of Medicine, Dentistry and Health Sciences at the University of Melbourne, and was the busiest month in 2013 for media coverage with 2,059 hits (radio, TV and print).

This story was covered by many in-print modes around Australia on May 2013. Please follow the links to read more about the [Absolom Twins](#) to view the [ATR media release](#).

## ATR Research day with Patron

On October 26<sup>th</sup> the ATR hosted the Patron, her Royal Highness Crown Princess Mary of Denmark, who toured twin studies in Sydney, marking her first official engagement in Australia as the [ATR's International Patron](#). The Crown Princess and HRH Crown Prince Frederik visited ATR-supported twin studies at the Sydney Children's Hospital.



ATR members Jared and Noah with ATR Patron, HRH Princess Mary at ATR Researcher event, Sydney Children's Hospital, October 2013.

According to ATR Director, Professor John Hopper, it was an historic day for the ATR, recognizing the tireless efforts of twin researchers and ATR twin members around Australia. "The royal couple enjoyed meeting researchers, and many ATR twin members and their families," he said. "It was great to see how interested they were in everything, being proud parents of twins. They were telling me stories about their twins and asking lots of questions."



ATR and AMBA twins meet HRH Crown Prince Frederik and HRH Crown Princess Mary at the Sydney Children's Hospital, October 2013.

Professor Hopper said the royal couple found the studies into childhood conditions of particular interest. Twin researchers spoke about their studies into childhood pain disorders, children's



literacy and numeracy skills, and emotional resilience. It was a memorable day for the many ATR twins and their families who met the royal couple.

“They were both so down to earth,” said mother of twins, Janine Geerin. “The princess asked my 15-year-old twin boys about their experiences at school – whether they are in separate classes, have the same friends, and how close they are. The prince spoke to them about sport and music.” Another mother of twins, Tracy Triggell, said “it was lovely how they took the time to chat to everyone about their experiences as twin parents and the many unique situations that arise”. “They also spent time with all of the children and this was a fantastic thrill for my six-year-old twin boys. It was truly a day we will never forget,” Tracy said.



Kai and Siobhan Pereira and Lillian and Charlotte Harding presenting ATR International Patron HRH Princes Mary and HRH Crown Prince Frederik with flowers at the ATR Researcher visit to the Sydney Children's Hospital, October 2013.

The royal couple's visit helped spread awareness globally about the importance of twin research with a huge media contingent from Australia, Denmark and around the world following the tour. “We very much appreciate the prince and princess taking time out of their busy schedules to make this visit possible,” Professor Hopper said. “Not only did the royal couple raise awareness about twin research, but they made time for each and every one of our members, providing lifelong memories to all.”

We thank our event sponsor and ATR members, The Ritchie Family, and our event partner, the Sydney Children's Hospital. Our members can share in the highlights of this special day with photos and video at our [website](#), [Facebook](#) and [Twitter](#).

## Tim Spector Forum

On 28<sup>th</sup> November 2013 the ATR hosted a forum, *Twins: changing the future of genetics* with world renowned twin researcher and epigeneticist, Professor Spector, Director of the UK Twin Registry and Professor at King's College London, raising awareness of the vital role of wins in research. He asked questions on how genes shape our personal characteristics, health and identity. Professor Spector says even genetically identical twins can be very different, and we can learn much about diseases and our own health by understanding similarities and differences between twins.



Panel members A/Prof Jeff Craig, Prof Tim Spector and Prof Nick Martin at the Twins: Changing the future of genetics hosted by the Australian Twin Registry, Thursday 28<sup>th</sup> November 2013.



Prof Tim Spector with ATR twin members attending the Twins: changing the future of genetics forum in November 2013.

The forum was attended by over 100 twin members and researchers and was widely reported in the media through print, radio, TV and online and broadcast up to 9 million people.



## SCIENTIFIC MERIT

*Collaboration 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*

### Current Research Studies

Building on a previous average (in 1999-2003) of 10 - 15 studies per year, the ATR aims to increase the number of studies supported per year to 15 - 20. This includes studies that:

- are in the initial stages of planning and development;
- are involved in active recruitment;
- have completed or paused recruitment, but may require additional support for follow-up and clarification with members.

The total number of active and ongoing studies utilising ATR services and/or involving ATR members increased to 83 studies in 2013. This includes the active processing of 6 Expressions of Interests (EOIs) for new research, 2 new research applications (all as a result of approved EOIs) were processed throughout the year. In total, there were 13 active recruiting studies with participants in 2013, 3 of which were completely new, and the provision of ad hoc support to a further 69 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 2013, is shown in **Table 2** and a count of active recruiting studies in the past seven years is shown in **Figure 12**.

**Table 2:** Studies by Status 2013.

Study Status	Number
Application (EOIs, Full Application, Protocol Change)	14
Recruiting	13
Data Collection	5
Data Analysis	34
Writing Up/Publishing	17
<b>TOTAL in 2013</b>	<b>83</b>

**Figure 12.** Number of active recruiting studies between 2006-2013.



## 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 2013), major achievements for this period, and future plans.

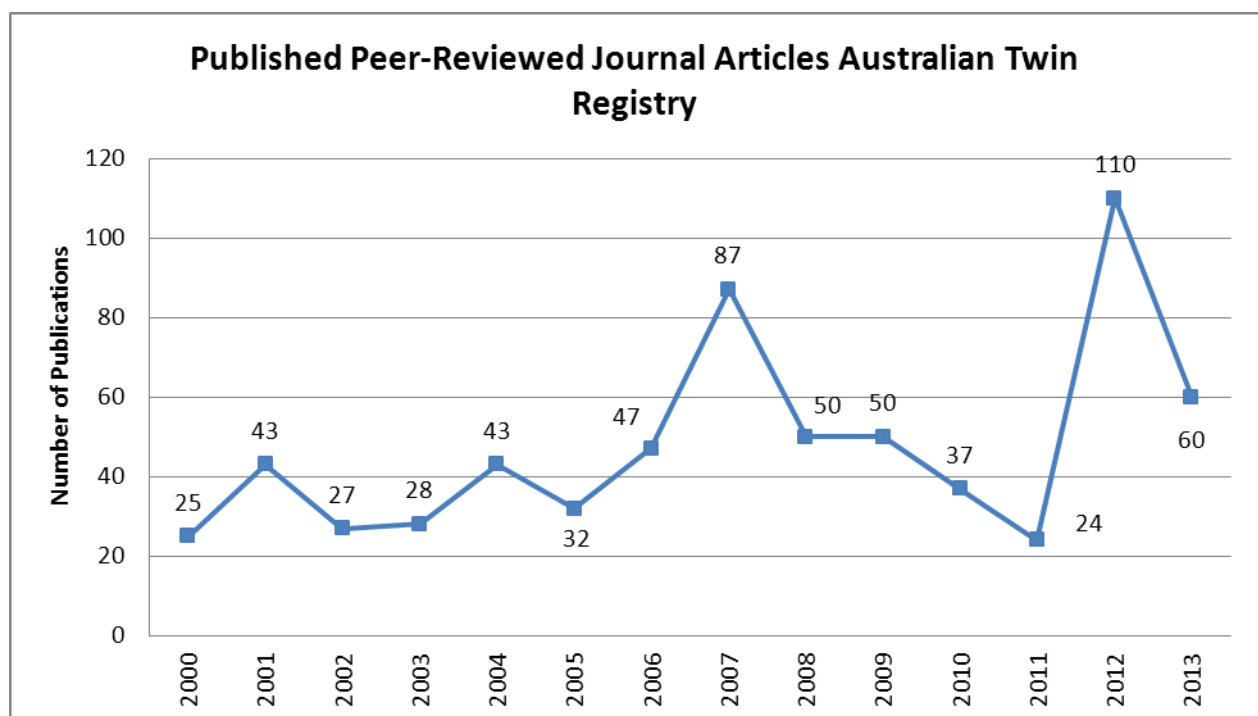
The Annual Researcher Satisfaction Survey has been fully integrated with the ATR's Annual Progress Report. Refer to Value Add section.

## 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.

In 2013, the ATR has recorded 60 peer-reviewed journal articles (**Figure 13**), 4 book chapters, and 25 conference proceedings, for a total of 89 publications. The goal of having 50 peer-reviewed articles yearly was reached, although several additional publications were reported to be in press or in preparation from 2012, were published and recorded in 2013. The list of all 2013 publications can be found in [Appendix 3](#).

**Figure 13.** Number of articles arising from ATR studies that were published in peer-reviewed journals every year since 2000.



## **Meetings and Conferences**

- *ATR International Patron Launch with HRH Princess Mary of Denmark, in Denmark and Melbourne, January 2013 (refer **Media Activities**)*
- *Lecture presented by ATR Director to 250 university students, 18<sup>th</sup> March 2013.*

ATR Director presented a lecture to 250 University of Melbourne science students on Genetics, Health and Society. This could provide a basis for lectures in other Institutes and Universities.

- *The 2<sup>nd</sup> International Network of Twin Registries (INTR) Seoul, South Korea, 31<sup>st</sup> May – 1<sup>st</sup> June 2013.*

The ATR Director and Deputy Director, A/Prof Jeff Craig, attended the 2<sup>nd</sup> International Network of Twin Registries (INTR) held in Seoul, South Korea on 31<sup>st</sup> May – 1<sup>st</sup> June 2013. This meeting involved 37 people from 15-20 registries from Europe, China, Japan and other Asian countries and the USA.

- *ATR Research event with ATR Patron HRH Princess Mary of Denmark, researchers and twin members at the Sydney's Children Hospital, 26<sup>th</sup> October 2013 (refer **Media Activities**)*

The ATR held a special event with ATR International Patron, HRH Princess Mary, at the Sydney Children's Hospital on 26<sup>th</sup> October 2013, showcasing twin research with ATR researchers and twin members from the ATR and AMBA, with coverage to over 9 million people on all prime TV.

- *The ATR hosted a public forum "Twins: Changing the future of genetics" with Prof Tim Spector at the University of Melbourne on Thursday 28<sup>th</sup> November, 2013 (refer **Media Activities**).*

The ATR Director invited Prof Tim Spector, a world renowned epigeneticist and twin researcher, to Melbourne to present a public forum of researchers and twin members. This generated coverage on TV news broadcasts, radio newspapers and online media around the country.

## **Research Travel Grant Scheme**

The Travel Grant Scheme supported a total of 17 early-career researchers in Round 11 to attend relevant national and international research workshops and conferences relating to twin research.

Round 11 of the ATR Research Travel Grant Scheme was awarded in April 2013. Congratulations to the following successful recipients:

- Amir Batouli - Brain and Ageing Research Program, School of Psychiatry, Faculty of Medicine, University of New South Wales
- Jeffrey Craig – Early Life Epigenetics, Murdoch Children's Research Institute, The Royal Children's Hospital
- Teresa Lee – Older Australian Twin Study, Brain and Ageing Research Program, University of New South Wales
- Jane Ebejer - Genetic Epidemiology, Queensland Institute of Medical Research
- Paulo Ferreira - Discipline of Physiotherapy, Faculty of Health Sciences, University of Sydney
- Negar Shahmoradi – Department of Endocrinology, Heidelberg Repatriation Hospital
- Xiaofang Wang – Department of Endocrinology and Medicine, Austin Health, the University of Melbourne

Grants provided opportunities for new researchers to attend diverse meetings and conferences. To date \$34,200 has been allocated to 61 young researchers since the beginning of this Enabling Grant.

### ***Australian Multiple Birth Association (AMBA)***

The ATR and AMBA continued to collaborate together in a range of different ways including speaking at national and state AMBA meetings and supporting the AMBA Club Initiative.

- *North-West Multiple Birth Association on 22<sup>nd</sup> May, 2013.*

The ATR Manager spoke at the North-West Multiple Birth Association AMBA club in North Melbourne on 22<sup>nd</sup> May, 2013 about the ATR and how twin research benefits the wider community.

- *The 21<sup>st</sup> Convention of the Australian Multiple Birth Association (AMBA), Canberra, Australian National Capital, 25-26<sup>th</sup> October 2013.*

The ATR presented a brief update on ATR and AMBA developments, including the launch of the AMBA Club Initiative. Participation of ATR in the AMBA National Convention in 2013 strengthened the close relationship between the Registry and AMBA and also increased awareness of the ATR brand amongst the twin community.

In 2013 the ATR continued the ATR Club Support Initiative in financial support. The grants go towards supporting administration costs for promoting the ATR at club level, resulting in the transfer of contact details of parent with twins. In 2013 a total of 35 AMBA clubs around Australia applied for the grants. All clubs are asked to include a section in their membership forms which permits AMBA to pass on their contact details to receive more information about the ATR. In 2013 the ATR received 435 new registrations from this initiative.

## PARTICIPATION

*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*

### Study Approaches

Mailouts to prospective participants for individual studies are a core component of the Registry's daily operations. Scheduling of mailouts and the total number of approaches sent is dependent on the requirements of the researcher. During 2013 a total of 10,773 letters or emails were sent (**Table 3**).

**Table 3:** Total number of mailouts and total letters by study in 2013.

Study ID	Study Title	Num Mailouts	Total Letters	RTS Rec'd	RP Mail Rec'd
2013-001	Why do we get addicted? A population based twin study of the relationships between impulsive and compulsive behaviours	3	906	0	0
2012-001	Bone health in children and young people with epilepsy treated with anti epileptic drugs (AEDs)	1	17	0	0
2012-012	Why do some people over consume fatty foods?	5	2004	67	161
2012-010	The genetics of singing ability	6	1439	0	0
2012-005	How does host cell influences HIV diversification	4	400	20	93
2012-003	A Twin Study of the NAPLAN	9	4109	152	2176
2008-004-1	The Emotional Well-being project phase 2	6	152	0	1
2008-002-3	Type 2 Diabetes and brain function – a co-twin study of brain metabolism	1	26	0	12
2007-005-6	An investigation into the nature of Growing Pains in Australia: Abdominal Pain Ext	1	18	0	57
2007-005-4	Back Pain and Other Regional Pain Disorders in Australian Twin Families	6	1702	19	193
<b>TOTAL</b>		<b>42</b>	<b>10773</b>	<b>258</b>	<b>2693</b>

**Table 3** shows these numbers by study and summarizes 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. There were 10 studies that had <5 RTS and Reply Paid mail received returns that were not included in the report as they relate to responses from recruited members mailed out in 2012.

### 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 most of the ATR actively recruiting studies during the reported period.

Telephone follow-up for studies is a significant component of the day-to-day work of Registry staff. 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 study calls and hours spend on the calls by the ATR staff in 2013.

Study ID	Study Name	Num Ph Calls	Total Hrs
2010-006	Beauty and the Eye of the Beholder	1	
2012-001	Bone health in children and young people with epilepsy treated with anti-epileptic drugs (AEDs)	27	1.75
2012-003	A Twin Study of the NAPLAN	5536	481.2
2012-005	How Does Host Cell Influences HIV Diversification	423	39.25
2012-012	Why do some people over consume fatty foods?	398	33
2005-003-2	OATS Extra Component	68	6.25
2007-004	Role of genetic and environmental factors in atrial fibrillation	5	1.75
2007-005-4	Back Pain and Other Regional Pain Disorders in Australian Twin Families	494	38
2007-005-6	An investigation into the nature of Growing Pains in Australia: Abdominal Pain Ext	27	2.25
2008-002-3	Type 2 Diabetes and brain function – a co-twin study of brain metabolism	24	5.75
2008-004-1	The Emotional Well-being project phase 2	260	26.75
<b>TOTAL</b>		<b>7263</b>	<b>635.95</b>

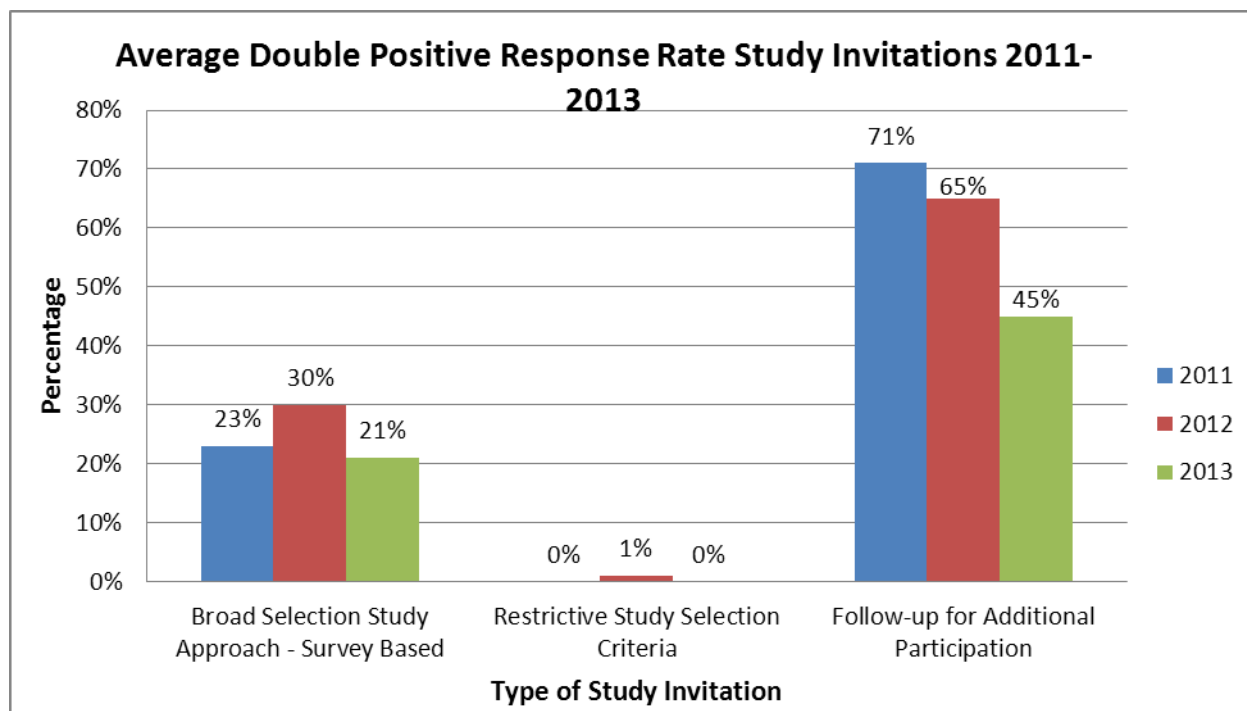
A slight decline in researchers requesting Telephone Follow Up and the Registry's adoption of a Verbal Response protocol, where a twin gives agreement over the telephone regarding their willingness to participate in a study, has reduced the number of follow up mail outs and approaches required. (Refer to **Figure 11** to see the changing response modes to studies). In 2013 there were less mail outs compared to the previous year, and as costs and overheads have risen, researchers have requested an increase in email follow up, resulting in a decrease in Telephone Follow Up, from 10,437 in 2012 to 7,263 in 2013.

## 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.

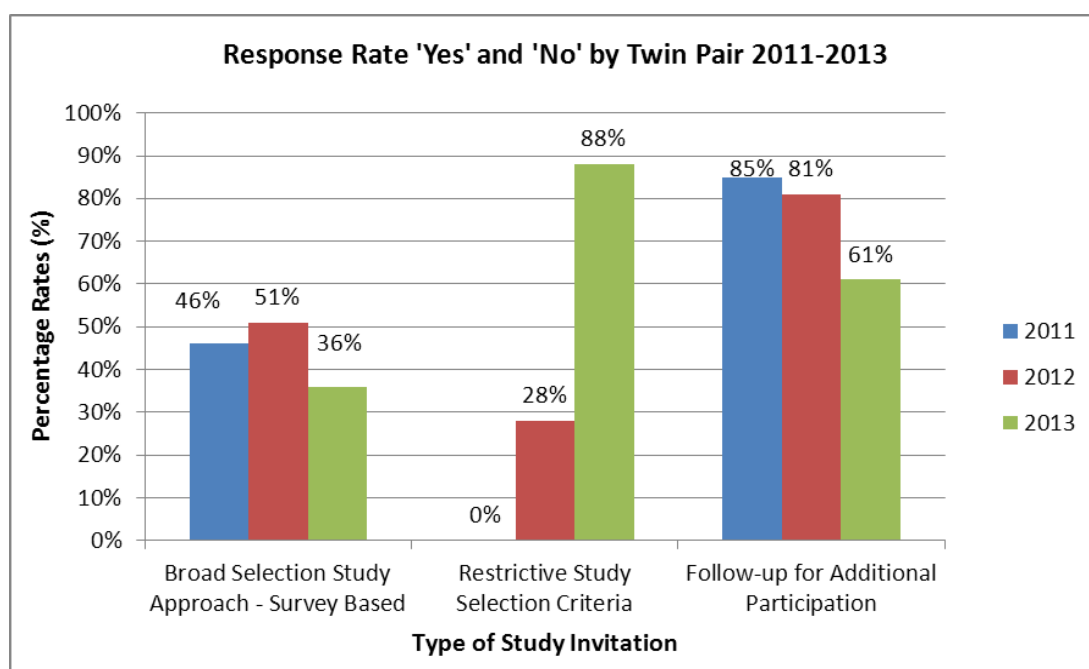
**Figure 14** shows double response rates, or double "yes", for most active studies in 2013, and for those studies that have been ongoing since 2011 in three categories – Broad Selection Study Approach – Survey Based, Restrictive Study Selection Criteria, and Follow-Up for Additional Participation.

**Figure 14:** Study response statistics to date for most active studies 2011-2013 (ongoing and recruiting)



**Figure 15** shows study response rates with a hard 'yes' and a hard 'no' for 2011-2013 years. The positive strong 'yes'/'no' response shows that the engagement of the ATR membership.

**Figure 15:** Study response statistics to date for most active studies 2011-2013 (ongoing and recruiting)



Response rates to Registry mail outs will depend on the study showing different study levels and responses. 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.

### ***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 ATR. ATR ensured the resolution of this complaint was addressed by the study researchers in a timely manner.

## VALUE-ADD

*Development of projects and programs to value-add to twin research in Australia*

### Quality Improvement Project

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 2013, the 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:

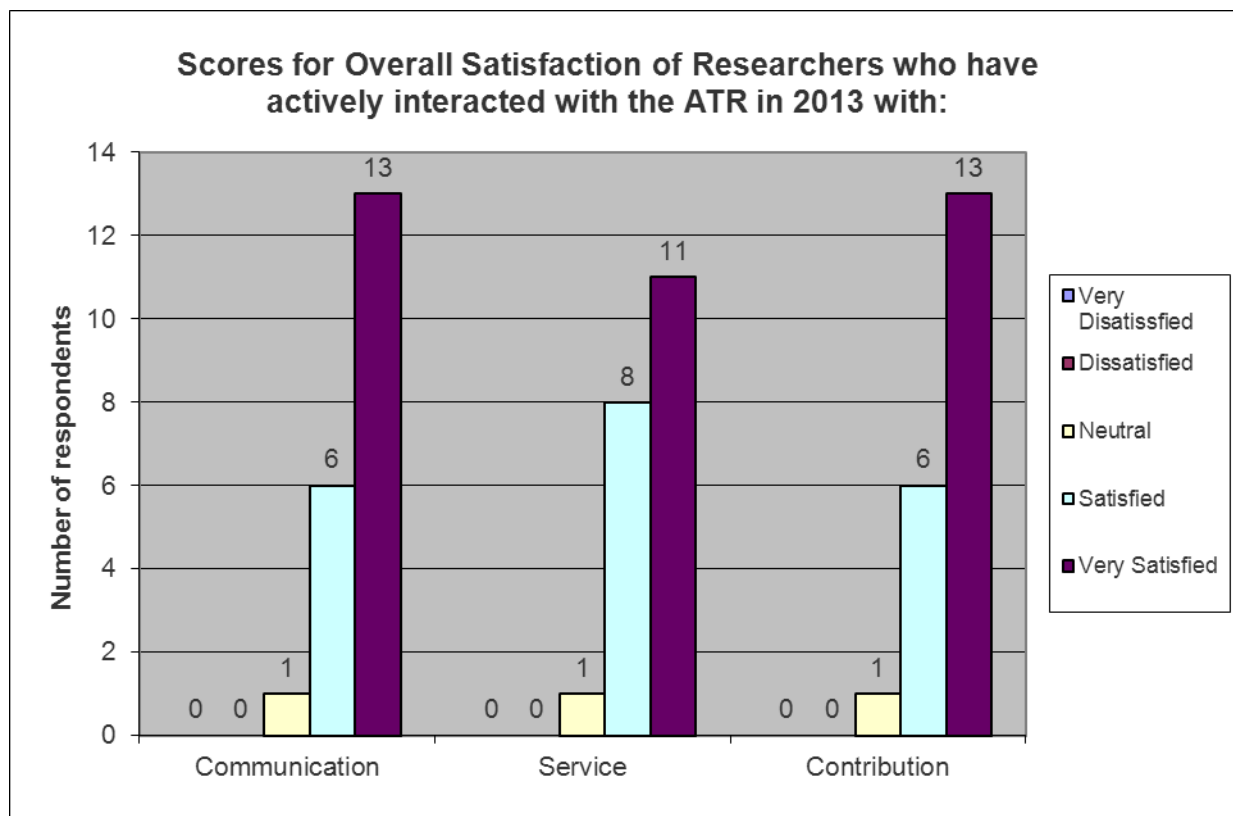
- (1) overall satisfaction with the researchers' communication with the ATR;
- (2) overall satisfaction with the services that the ATR provided; and
- (3) the value of the contribution that the ATR made to the overall research project

Responses were recorded as

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

We received feedback from 34 research groups. Overall, researchers were very satisfied with the communication and service provided by the ATR and the contribution the ATR has made to their project (**Figure 16**).

**Figure 16.** Overall satisfaction scores from researchers, evaluating communication with ATR, and service and contribution provided by the Registry.



Feedback from researchers accessing the ATR is gathered annually. 2013 feedback indicates that overall researchers are very satisfied with the communication and service provided by the ATR and the contribution the ATR has made to their project – refer **Figure 16**. Examples of comments provided by researchers include:

Communication: A number of respondents commented that ATR staff *'are always reliable and prompt in their response to requests'* and **'friendly, efficient and well informed'**, *'helpful with ideas around recruitment and issues on zygoty testing'* and *'are in contact as required and have been exceedingly helpful with regards to any issues that have arisen'*.

Service: *'Support from the staff at the ATR has been extremely fast and efficient and provided timely, accurate advice and have followed-up on any queries we have had.'* Whilst some studies had low rates of participation this did not impact on the **'excellent services provided professionally'** and *'Communication has been good and whilst we would have liked a higher rate of participation to date I do not believe this was due to ATR.'*

Contribution: A large number of researchers commented that **'the foundation and success of the projects could not have happened without the involvement of the ATR'** and *'Outstanding contribution, the project could not be implemented without the ATR'*. One researcher commented *'Working carefully to a grant budget we found it difficult that costs from ATR increased considerably during the process'*.

Other comments: **'The ATR has been tremendously helpful with this study and our engagements internationally'** and *'The ATR is invaluable to recruiting twins. Without their help and support, it would be very difficult for researchers to recruit twin participants, particularly the required sample size for various statistical methods.'*

### **ATR Data Index Project**

The ATR created a web-based, searchable index of questions and topics covered by all previous studies for researchers, launched in 2009. The index is updated each month and is accessible at

[http://www.twins.org.au/study\\_index/BasicSearch.php](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.

Researchers interested in utilising these existing data 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. The open source licensing of The Ark means that researchers are also free to install separate local instances to support single studies or whole institutions, depending on their needs. Ark modules to support recruitment, subject management, electronic questionnaires, phenotypic and biospecimen data, and model and visualise pedigree structures are currently in place.



The 2013-2014 period has seen a significant amount of Ark development work. Pedigree modelling functionality is complete, enabling the system to model complex family structures and twin relationships. Monozygotic and di-zygotic relationships can be captured in a flexible and user-friendly manner, and dynamic configurable pedigree diagrams can be created at the click of a button. The Ark's Laboratory Information Management System (LIMS) has been revamped to extend its capabilities and stability. The improved LIMS module will be a key asset in managing the data from the ATR's forthcoming initiative to collect biospecimens from members. In addition to these changes, over 100 miscellaneous improvements have been implemented, along with resolutions for small issues and bugs. The remainder of 2013 will be spent undertaking highly-focused programming on the Ark to improve the user-interface of the phenotype module, introduce new export formats (for analysis, e.g. Stata), add localisation (foreign language) support, and implement a new configurable module to generate study feasibility reports.

## GOVERNANCE

*Governance of the ATR in a fair, transparent and equitable manner*

### **ATR Management**

As of 31 December 2013, the ATR Management comprised:

- Prof John Hopper, AM, Director, Australian Twin Registry; NHMRC Senior Principal Research Fellow; Director (Research), Centre for MEGA Epidemiology, 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
- 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 2013, the Advisory Board members were:

- Mr Vincent Pollaers (Chair, Twin Representative, New South Wales)
- Mrs Ann Marie Harli (AMBA Representative, Victoria) stepped down in September 2013.
- Mrs Karen Willetts (AMBA Representative, NSW) was appointed in November 2013.
- Dr Keith Horsley (Australian Institute of Health and Welfare, Retired)
- Dr Paul Jelfs (Australian Bureau of Statistics, Australian Capital Territory)
- A/Professor Paul Lancaster (University of Sydney, Retired)
- Mr William Mackerras (Twin Representative, Australian Capital Territory) stepped down in December 2012.
- Ms Sue Carrick (Twin Representative, NSW) replaced Mr William Mackerras in January 2013.
- Prof Margaret Otlowski (University of Tasmania, Tasmania)
- A/Prof David Whiteman (Queensland Institute of Medical Research, Queensland)
- Prof David Ravine (Western Australian Institute of Medical Research, Western Australia) stepped down in December 2013.

Ex-officio:

- Prof John Hopper (Director, ATR, University of Melbourne)
- Dr Debra Foley (Deputy Director, ATR, ORYGEN Research Centre) and Mr Paul White, Deputy Director (Informatics), University of Western Australia stepped down as Deputy Directors in June 2013
- 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

### ***ATR Staff***

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 2013, the ATR continued to employ an ATR Manager, ATR Study Coordinator, ATR Assistant Manager, a Marketing Officer, Project Support Officer, Project Assistant and an Administration Assistant, five casual staff (equivalent of 2.0 EFT), and a part time Database Manager.

The ATR provided an honorarium to the Director and 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 2013.

### ***ATR Budget***

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 2013 towards a total of \$20,355.

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

## **Appendix 1: Researcher Reports 2013**

### **2005-003-2 THE TWIN STUDY OF BRAIN AGEING AND COGNITION – Extra Component**

#### **Principal Investigator:**

**Professor Perminder Sachdev**

**Neuropsychiatric Institute, Euroa Centre**

**Prince of Wales Hospital, NSW**

Although dementia is an age-related illness, not everyone who reaches old age falls victim to this disease. Researchers of the Older Australian Twins Study (OATS), the most comprehensive ageing study with elderly twins ever undertaken in Australia, aim to identify what factors help to prevent dementia, what environmental or genetic factors are responsible for dementia, and what genes are involved. OATS is a collaborative project between researchers of the University of New South Wales (UNSW), the Queensland Institute of Medical Research (QIMR), and the National Ageing Research Institute (NARI).

Study participants are 65 years and older identical and non-identical twin pairs, living across the Eastern seaboard. They undergo rigorous medical and cognitive function tests, give blood samples for research, and have an MRI scan of their brain. Information about environmental factors, such as lifetime physical and mental activity, physical and psychological trauma, and nutrition is also collected. Comprehensive cardiovascular tests, and in some instances, falls and balance checks are also provided. Participants receive written feedback in relation to the tests that they undergo. Two-year and four-year follow-up tests are carried out to measure change. Researchers hope the study could eventually become longitudinal, whereby the same group of participants is followed-up for many years. Ideally, the results may eventually be used to develop strategies for the prevention of dementia.

In 2013, researchers applied for more funding for this successful study, through the submission of an NHMRC Project Grant for Wave 3 (2013-2015), to continue Wave 2 activities, start the PiB-PET pilot study, and complete zygoty analysis. We have applied for funding for further scans to improve our ability to conduct sensitive statistical analyses.

The study collected pilot PiB-PET data from 60 twin participants (17 monozygotic [identical], 13 dizygotic pairs). All scans were performed at the Austin Hospital, Melbourne. Participants receive written feedback in relation to their cognitive strengths and weaknesses, brain imaging, cardiovascular and blood results shortly after their face-to-face assessment for the main OATS study. A 2013 Christmas newsletter was prepared for current OATS participants to give an update on the study.

Data analysis is complete and the study found a moderate correlation of amyloid deposition in monozygotic twins and a lack of correlation in dizygotic twins. Heritability estimates were moderate, suggesting significant environmental contributions to amyloid deposition.

The study aims to publish 10-15 papers and in 2014 will prepare manuscripts for publication that present the pilot data.

**Current status: ACTIVE – RECRUITING**

**2007-005-6 AN INVESTIGATION INTO THE NATURE OF GROWING PAINS IN AUSTRALIA:  
ABDOMINAL PAIN - EXTENTION**

**2007-005-5 COMMON PAIN DISORDERS OF CHILDHOOD**

**Principal Investigator:**

**A/Professor David Champion**

**Paediatric Pain Research**

**Sydney Children's Hospital, QLD**

The aim of this study is to investigate the heritability of growing pains, associations with restless legs syndrome, functional pain syndromes and psychological measures. Growing pains appears to be more genetically influenced than other common primary pain syndromes. The associations with restless legs syndrome, migraine and non-migraine headache, which these researchers have identified, have potential implications with respect to genes, nature of the disorder, and therapeutic applications.

Analyses in 2012-2013 have supported heritability of growing pains, migraine, recurrent abdominal pain, back pain, regional pains and restless legs syndrome, but not non-migraine headache. There are multiple associations in twin individuals between these conditions and between each pain disorder (except headache) and restless legs syndrome. This raises opportunities for further research directions based on pathogenesis, molecular genetics and pharmacotherapy of restless legs syndrome. Based on recent evidence in other contexts, it is increasingly likely that gene variants in common are conferring risk for multiple pain disorders of diverse category and anatomical site.

In 2013 the pain disorders are showing multiple associations with each other and with restless legs syndrome, anxious depression, iron deficiency and multiple sensory sensitivities, each of which is heritable. Akin to recent evidence regarding psychiatric disorders and selected autoimmune disorders, it is likely that a set of gene variants influences the risk of multiple pain disorders. The restless legs syndrome, anxious depression and iron deficiency associations suggest that the pain disorders share neurobiological mechanisms with that condition, and this gives promise about new genetic, pathophysiological mechanisms, and pharmacotherapeutic concepts.

A model constructed by Prof Carl von Baeyer and David Champion, published in the Journal of Pediatric Psychology, suggests the data results so far indicate probable genetic influence for growing pains, migraine, non-migraine headache, recurrent abdominal pain, back pain, and regional pain disorders, the heritability estimates ranging from 39% to 70%. These findings were presented to national and international conferences and will continue in 2014. The study will consider a supplementary component into dysmenorrhea in adolescent girls and is currently in protocol development.

A Bachelor of Science (Med) Honours project on the relationship between iron deficiency and the common pain disorders of childhood was instigated to assist with the analysis of data from the study. David Champion presented the study to ATR Patron HRH Princess Mary of Denmark in October 2013.

**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. These test results will be

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).

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.

Bone density results are sent pout to participants within 2-4 weeks after their visit. A letter is sent to further explain how to read the results. A number of papers were written and published of the findings, including the 2013 paper from M Bui, A. Bjornerem, A. Ghasem-Zadeh, G.S.Dite, J.L.Hopper and E.Seeman's titled 'Architecture of cortical bone determines in part its remodeling and structural decay' Bone 55 (2013) 353-358.

**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 plans to continue with recruitment and to explore other avenues such as international collaboration to optimise participant numbers.

**Current Status: ACTIVE – RECRUITING**

**2008-004-1**

**PATHWAYS TO AFFECTIVE DISORDERS: INTERACTIONS BETWEEN GENES,  
ENVIRONMENT AND BIOLOGICAL MECHANISMS or EMOTIONAL WELLBEING PROJECT  
PHASE 2**

**Principal Investigator  
A/Professor Leanne Williams  
Westmead Millennium Institute  
Westmead Hospital, NSW**

The TWIN-E Study (the Twin study of Wellbeing using Integrative Neuroscience of Emotion) is a large national Australian study focusing on identifying the predictors of emotional wellbeing in 1500 MZ and DZ twin pairs. This is a collaborative study between the University of Sydney, NeuRA, and Flinders University. By focusing on wellbeing instead of mental illness per se, researchers hope to understand for the first time the genetic and biological underpinnings of 'resilient' individuals; that is, what characterizes those people who are able to bounce back from adversity. Adopting a multidisciplinary approach, they are assessing healthy twins on a number of measures including genetics, cognitive function and personality assessed using web-based assessments, and brain function assessed using the EEG and MRI. By comparing MZ to DZ twins, these researchers will be able to assess the relative contribution of genes versus one's environment in resilience, providing a focus for preventative health regimens.

The new development of the TWIN-E project in 2010 involved a second phase (2008-004-1), whereby twins had the option of receiving personalized 'brain scores' and were then recommended certain types of brain training to optimize their cognitive function. The aim of this study is to identify the gene-environment predictors of response to cognitive brain training seeking to optimise cognitive and mental health. In addition to providing added confirmation of the utility of preventative health strategies in promoting emotional wellbeing, by comparing twin pairs, researchers will be able to identify whether there are certain genes or environmental life experiences that predispose one to favourably respond to such intervention techniques. The study provides a one-page report of average heritability of different cognitive factors to all participants once they complete the 12-month follow-up session. Participants also receive their personalized cognitive scores if they participate in Phase II (brain games phase). For the Phase 2, researchers completed the Phase II testing and data collection. They conducted heritability statistical analyses for the full sample in baseline measures.

In 2013 preliminary results were reported at national/international conferences and published in papers where possible. Study analyses has been summarized and published in the *Twin Research and Human Genetics* (Gatt et al., 2012). An additional four papers were published in 2013. Two examples of researchers attending conferences include Kylie Routledge's presentation 'Testing the impact of online brain training in promoting wellbeing and resilience' at University of NSW Brain Science Symposia in Sydney during October and Justine Gatt's presentation 'Testing the impact of online brain training in promoting wellbeing and resilience' at the University of Sydney e-health Showcase in Sydney in November 2013.

During 2014 the study will finalise the data analyses and share results for publication and presentation at conferences and workshops.

**Current Status: ACTIVE – RECRUITING**

**2008-006**

## **GENETICS OF SYNCOPE AND BREATH HOLDING**

### **Principal Investigator**

**Prof Sam Berkovic**

**Epilepsy Research Centre**

**Melbourne, VIC**

Syncope is a common problem with multiple causes, where clinical genetic evidence strongly suggests a major genetic contribution. The genes for syncope are unknown. In this study, researchers recruit twin pairs and families to determine the role of genetics in breath holding and syncope. Twins are particularly important to this study because, with the same or extremely similar genetic make-up and similar environmental exposure, subtle differences explaining the aetiology of diseases can become apparent.

Outcomes of this study will include a deeper understanding of how these conditions are inherited and the nature of the genes involved. This should lead to better diagnostic methods, and an improved knowledge base to counsel patients. Moreover, gene discovery will provide novel insights into the fundamental cause of these phenomena that could aid treatment in severe cases.

By studying twins and individuals with no family history of epilepsy, fainting or breath holding and also families where more than one person has experienced seizures, fainting or breath holding the researchers hope to gain a better understanding of the genetic relationship and improve understanding of the mechanisms that cause these events.

In December 2011, the study recruited and studied a further 28 twin pairs in which one or both twins were reported to be affected by breath holding or syncope. In 2012 the study continued recruiting more twin pairs and to carry out data collection and analysis.

During 2013 the study recruited and studied two twin pairs with epilepsy, two with syncope and four twin pairs with febrile convulsions. The Epilepsy Research Centre sends out annual newsletters to all study participants. The newsletter includes updates on the various research studies. The study published 3 peer-reviewed publications in *Epilepsy Research*, *American Journal of Neurology* and *Epilepsia*, and presented 2 posters at workshops in the USA and Australia.

The plans for 2014 are to recruit and study the remaining twin pairs who expressed interest in the study.

**Current Status: ACTIVE – RECRUITING**

**2010-003**

**OBSESSIVE-COMPULSIVE SPECTRUM DISORDERS: A POPULATION BASED TWIN STUDY OF MAJOR SYMPTOM DIMENSIONS**

**Principal Investigator**

**Dr Ben Harrison**

**The University of Melbourne  
Melbourne, VIC**

The study aims to investigate the prevalence and twin concordance of a wide range of anxiety related feelings and behaviors, in particular, focusing on putative obsessive compulsive spectrum disorders. This will be the first study to explore whether there is a common genetic influence across the proposed range of putative obsessive-compulsive disorder symptoms

List of formal scales used:

OCI-R (Obsessive Compulsive Inventory-Revised) HRS (Hoarding Rating Scale) DCQ (Dysmorphic Concerns Questionnaire) MGH-HPS (Massachusetts General Hospital- Hair Pulling Scale).

SPS (The Skin-Picking Scale)

WI (Whiteley Index, Hypochondriasis)

ASI-R (Anxiety Sensitivity Index)

SPIN (Social Phobia Inventory)

DASS-21 (Depression/Anxiety Symptom Scale) WHODAS 2.0 (Quality of Life).

In 2013 extensive data analysis was conducted and researchers attended multiple conference presentations. A manuscript was accepted in American Journal of Medical Genetics by Lopez-Sola C, Fontenelle LF, Alonso P, Cuadras D, Foley D, Pantelis C, Pujol J, Yucel M, Cardoner N, Soriano-Mas C, Menchon J, Harrison BJ. Prevalence and heritability of obsessive-compulsive spectrum and anxiety disorder symptoms: A survey of the Australian Twin Registry. American Journal of Medical Genetics: Neuropsychiatric Genetics (In Press).

During 2014 further data analysis, conference presentations & manuscript submissions will be undertaken. One manuscript is ready for submission and two additional manuscripts are underway.

The study will also include news of the recent publication and ongoing analyses in the ATR newsletter.

**Current Status: ACTIVE – RECRUITING**



**2012-001**

**BONE HEALTH IN CHILDREN AND YOUNG PEOPLE WITH EPILEPSY TREATED WITH ANTI-EPILEPTIC DRUGS (AEDs) (BONE HEALTH AND ANTI EPILEPTIC DRUGS IN CHILDREN AND YOUNG PEOPLE)**

**Principal Investigator**

**Dr Peter Simm**

**Royal Children's Hospital**

**Melbourne, VIC**

The use of anti epileptic drugs (AEDs) has been shown to be associated with lower bone mineral density (BMD) on dual-energy Xray absorptiometry (DXA) scanning, while fracture rates in patients with epilepsy are higher than the general population. The limited paediatric data shows similar findings to the adult reports, however fracture risk is not well defined in this age group. While there are multiple potential risk factors for poor bone health outcomes in patients with epilepsy, the low bone density seems to be independent of other risk factors such as immobility. There are also preliminary data suggesting adults with epilepsy treated with AEDs since childhood have lower bone mass accrual than those who commenced treatment as adults, pointing to a specific effect of AEDs on the developing skeleton.

To date, there are no data on pQCT findings in young people treated with AEDs and therefore their underlying skeletal geometry. Children and young people with epilepsy treated with anti-epileptic drugs (AEDs) have impaired skeletal development and bone mass accrual as compared with non AED treated controls. The study aims to 1) assess the impact of anti epileptic drugs (AEDs) on skeletal development, including volumetric bone density and skeletal geometry; 2) examine the effect of AEDs on other parameters involved in musculoskeletal development (such as muscle strength, Vitamin D levels, growth, soft tissue composition) and 3) assess the rate of fractures after the commencement of AEDs.

Twins participating in this study will have a bone health assessment, which will consist of a one-day visit (lasting 3 – 4 hours) for 2 different bone density scans, one Xray, one blood test, assessment of growth, measurement of grip strength, force plate testing of muscles (non-invasive) and completion of questionnaires.

During 2012 the study will be recruiting to undertake the assessments. We would hope to recruit 30 – 40 sibling/twin pairs over 12 – 18 months. In 2013 ongoing recruitment/study visits with twins recruited, with a further 11 pairs found and completing the study (14 in total now), almost all through the RCH epilepsy clinic. The main change proposed is to include first cousins (same gender, within two years of age) as possible controls. This will not affect the ATR twin recruitment.

In 2014 the study will focus on ongoing recruitment through ATR and RCH and preparation for an abstract entered for 2014 US Endo. A report will be prepared for all participants, as well as their individual bone density reports, at the completion of the study.

**Current Status: ACTIVE – RECRUITING**

**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 2200 families were recruited to the study. Questionnaire data concerning home demographics, parent attitudes and twin zygosity collected from all families; child-specific questionnaire data 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 was sent via email newsletter to all participants with an email address (approximately 2095 families out of 2200 recruited) in December 2013 and a copy of this newsletter was also provided to the ATR.

In 2014 the final collection of NAPLAN data from remaining states and territories will be conducted and the child-specific questionnaire sent to families of students sitting NAPLAN tests in 2014. Analyses of the data will commence and written in readiness for submission to publication.

**Current Status: ACTIVE – RECRUITING**

**2012-005**

## **HOW DOES HOST CELL INFLUENCES HIV DIVERSIFICATION**

**Principal Investigator**

**A/Prof Johnson Mak**

**Deakin University/Burnet Institute**

**Melbourne, VIC**

HIV continues to be a major cause of morbidity and mortality worldwide. Significant progress has been made in understanding the virology and immunology of infection, and a number of successful drug therapies have been developed to control the virus. Despite this, HIV infection still causes a large number of deaths in the developing world where access to drugs is limited. In the developed world, the requirement for lifelong therapy and the complications of this remain a significant source of morbidity. The most obvious solution to this problem is an effective vaccine, but progress on this has been slow and remains uncertain. In the absence of a successful prophylactic vaccine, it seems that the likely outcome of most interventions will be the chronic infection with successful control of the virus within the host, allowing the infected individual to reach a near-normal life expectancy. To achieve this, we need a good understanding of the host-pathogen interactions in chronic infection, and how these evolve over time. There is now a consensus that the immune response and the diversity of the HIV is likely to play a major role in driving HIV pathogenesis. Since HIV evades the immune system through rapid diversification of the viral population, (the viral diversity in a single patient is far greater than the annual global diversity of the influenza virus) any information which helps prevent viral escape and exhaustion of the immune system will be beneficial in combating AIDS progression.

The development of genetically diverse viral populations is one of the hallmarks of HIV infection and a key contributor to disease progression, distinct recombination and mutation rates amongst different donors may in part contribute to the varying rates of diseases progressions as seen in patients.

The overall goal is to define the mechanisms of HIV mutation and recombination and their role in AIDS pathogenesis. The aims of this study are to investigate the role of 3 key factors on the development of HIV diversity. Specifically, we will determine whether: (1) immune activation via vaccination; (2) genetic background (by obtaining primary cells from identical twins); or (3) expression of candidate host cell factors (via knockdown or deletion of nucleic acid editing/repairing enzyme) can influence the recombination and mutation rates of HIV infection.

During this period we have successfully advertised the study and recruited 20 participants. In 2013 the study continued the recruitment process for new twin sets and follow-up on those twins already recruited to the study. In 2014 the study will continue processing infection samples and create library for deep sequencing, analyse sequencing results and begin preparation of results for manuscripts.

**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). To date, 1,077 twin pairs have received email invitations from the ATR to participate in the online study. 106 twins have participated in the study, of which there are 34 complete twin pairs.

A paper has been prepared for publication by Y.T. Tan, G.E. McPherson, I. Peretz, S.F. Berkovic and S.J. Wilson, titled 'Investigating the genetic basis of music ability: A review in Frontiers in Psychology.

In 2014 the study will undertake another round of large-scale recruitment via email to 300 twin pairs has commencing in January 2014. As the uptake is still rather slow, in order to boost the participation rate, a lucky draw will be held for those twin pairs who both complete the online survey, where each member of a twin pair will stand to win an iPad mini. An application for an ethics amendment has been submitted and is currently awaiting approval before we can proceed with notifying twins who have been previously informed about the study about this new lucky draw. We aim to reach our recruitment target of 120 twin pairs and complete the data collection phase by June 2014 so that data analysis can commence in the latter half of 2014.

**Current Status: ACTIVE – RECRUITING**

**2012-012**

## **WHY DO SOME PEOPLE OVERCONSUME FATTY FOODS?**

**Principal Investigator**

**Prof Russell Keast**

**Deakin University**

**Burwood, VIC**

The study investigates if there is a genetic reason why some people consume more dietary fat than others and follows on from previous studies to broaden the understanding of fat taste sensitivity and fatty food preference. Oral fat sensitivity is strongly associated with overweight and obesity. Excess fat consumption is also associated with overweight and obesity, a condition linked to serious health consequences, such as cardiovascular disease and type-2 diabetes. However, the factors involved in individual variation in fat sensitivity remain unknown.

Differences in taste sensitivity influence food choices. It is likely that the genotype may explain some of the differences in food preferences among individuals. By analysing the within pair correlations of monozygotic twins, researchers want to investigate the heritability of fat sensitivity and its influence on fatty food preference, while minimizing environmental factors.

Feedback will be posted to all participants following completion of the study in the form of a report. The findings will be reported as group means. Participants will know how many twin pairs participated in the study, the mean age and BMI and we will also provide the concentration range at which people were sensitive to the taste of fat. Any association between fat taste sensitivity and genetic predisposition, diet and the liking of high or low fat foods will be noted. If a participant would like to know their individual results, for example their height or weight, or their liking for low or high fat foods, the details of the researchers will be provided and may be requested. Participants will also be able to find out the group results in the ATRs newsletter and on their website.

By December 2012, 20 twin pairs had completed the study. A number of pairs who initially expressed interest in the study were contacted and provided with additional information/questionnaires, however after receiving further details, wished to no longer take part. It was expected the remaining twin pairs will be contacted and data collection will be completed and preliminary results will be analysed and presented at the 10th Pangborn Sensory Science Symposium to be held in Rio de Janeiro, Brazil.

In 2013 the study successfully obtained ethics approvals and recruited 10 participants into the dietary intervention for an early 2014 start. Due to the low participant rate the study has applied to allow same sex siblings >5 years of age into the study.

In 2014 the study will begin the dietary intervention, data collection, further recruitment.

**Current Status: ACTIVE – RECRUITING**



**2013-001**

**WHY DO WE GET ADDICTED? A POPULATION BASED TWIN STUDY OF THE  
RELATIONSHIPS BETWEEN IMPULSIVE AND COMPULSIVE BEHAVIOURS**

**Principal Investigator  
Prof Murat Yucel  
Monash University  
Melbourne, VIC**

The words “impulsive” and “compulsive” are often used to describe how a person is acting. Although everyone experiences times in which they may be impulsive or compulsive, these styles of acting are found to occur more frequently in people who have some types of mental illness. There is also often a lot of overlap in the experience of impulsive and compulsive behaviours but we do not understand the extent to which these behaviours overlap. As such, this study will focus on understanding whether there is shared or unique heritability associated with these behaviours. Such knowledge will have direct implications for our understanding of disorders such as substance and gambling addiction, and obsessive compulsive disorder.

In 2013 the study obtained ethics and started the recruitment and data collection during this period. Almost 1000 potential participants were sent letters of invitation to be part of the study.

During 2014 the study will continue with data collection and recruitment and anticipate this will be completed by mid-year. Once this is done, we will begin data analysis and should have this completed by the end of the year. We anticipate publications to arise from this data in early 2015.

Upon completion, a summary of the study’s results will be provided to the ATR. These results will be available for inclusion in the ATR Newsletter and via the ATR website. Participants are also free to contact the researchers on the details provided in the plain language statement should they wish to receive other information about the group-level results of the study and any publications arising from the data.

**Current Status: ACTIVE – RECRUITING**

## **ATR 2013 IN PRESS – Articles in newspapers, magazines, online**

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- Georgakopoulos, C. (2013). Pereira twins meet Danish Royals. Blacktown Advocate. Blacktown.
- Gortan, A. R. (2013). Crown Princess Mary and Prince Frederik visit Sydney Children's Hospital. Daily Telegraph. Sydney.
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- (2013). Double Act. ABC News Perth. Perth, WA.
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- (2013). Mary in Sydney for Opera House birthday. National 9 News, Channel 9: 0:30.
- (2013). Mary to support twin registry. 7News.
- (2013). Mary to support twin registry. Channel 9 News Melbourne.

(2013). Princess Mary to support Australian and Danish twin registries. [Mix 101 FM](#).  
 (2013). Princess Mary to support Australian and Danish twin registries. [3AW Breakfast](#).  
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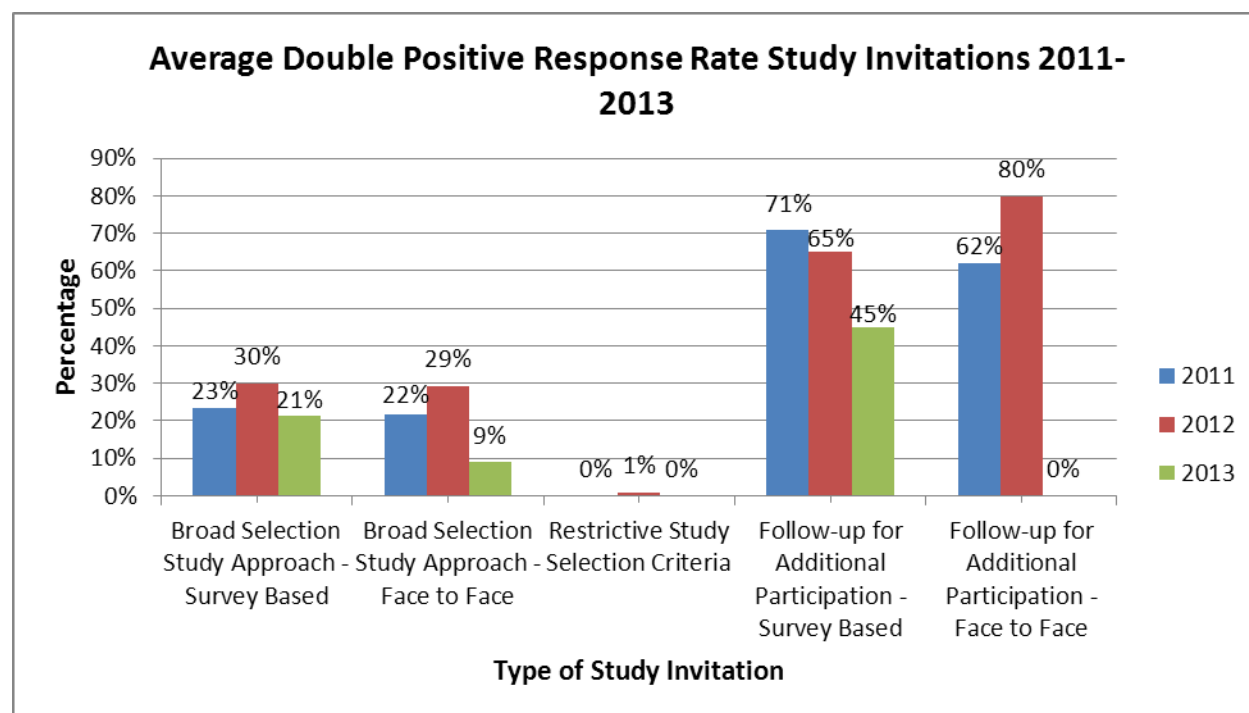
## ATR Response Rates in Active Studies 2011 - 2013

**Appendix 4** provides further breakdown the response rates for various modes of response, sex and age between 2011 and 2013. 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 of Twins who Responded Positively between 2011-2013

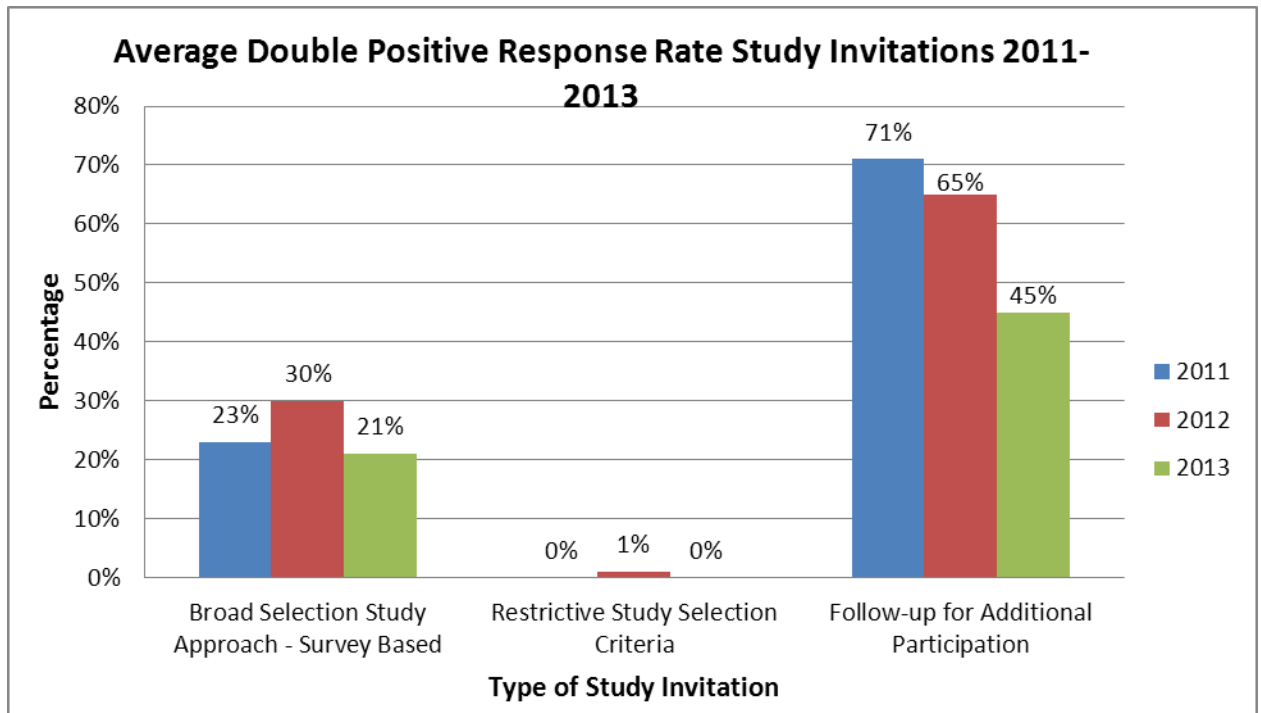
Average Double Response Rate for Study Invitations 2011-2013 include positive response from both individuals in a twin pair, or an individual response from a parent of a junior twin pair. This figure shows a positive pair response rate for each year for studies with mail outs from 2011 - 2013. Some study response rates will show responses for each year between 2011 – 2013, whilst other studies will only show for one or two years depending on the format of the study. For example, 1 study had a response rate for all three years, 10 studies have a response rate for 2 consecutive years and 22 have a response rate for only one year.

**Figure 1 (Figure 14):** Average Double Positive Response Rate for Study Invitations 2011-2013



Response rates to Registry mailouts will depend on the study showing different study levels and responses. 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.

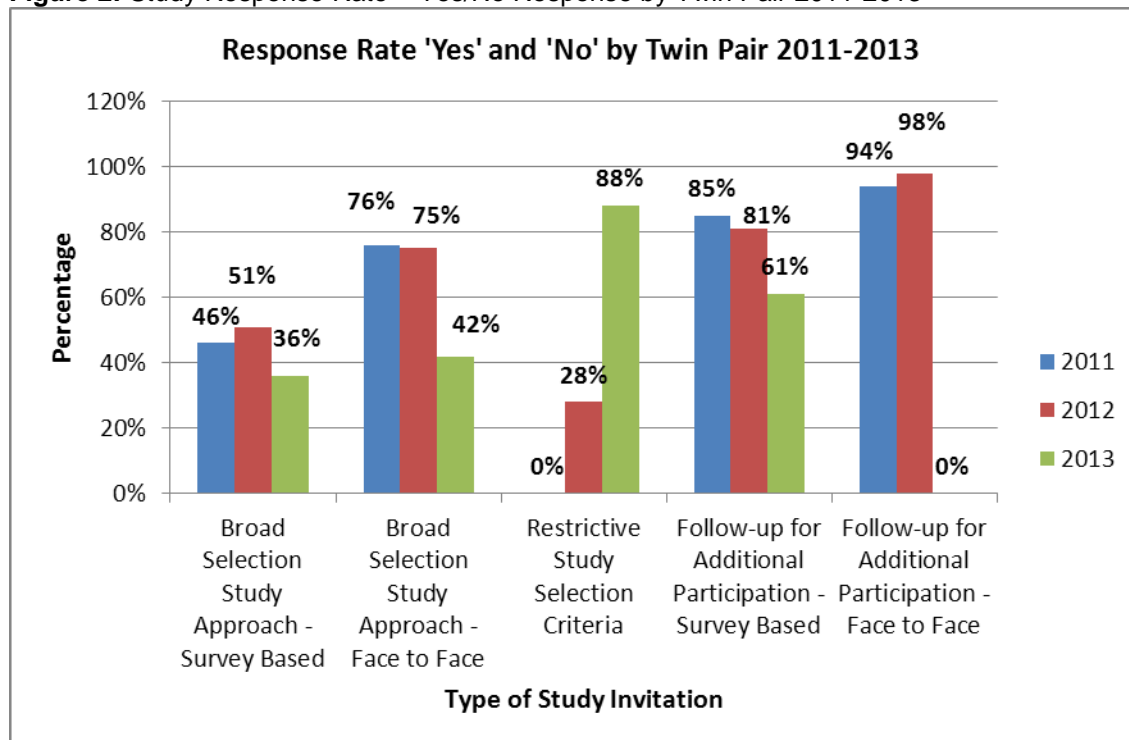


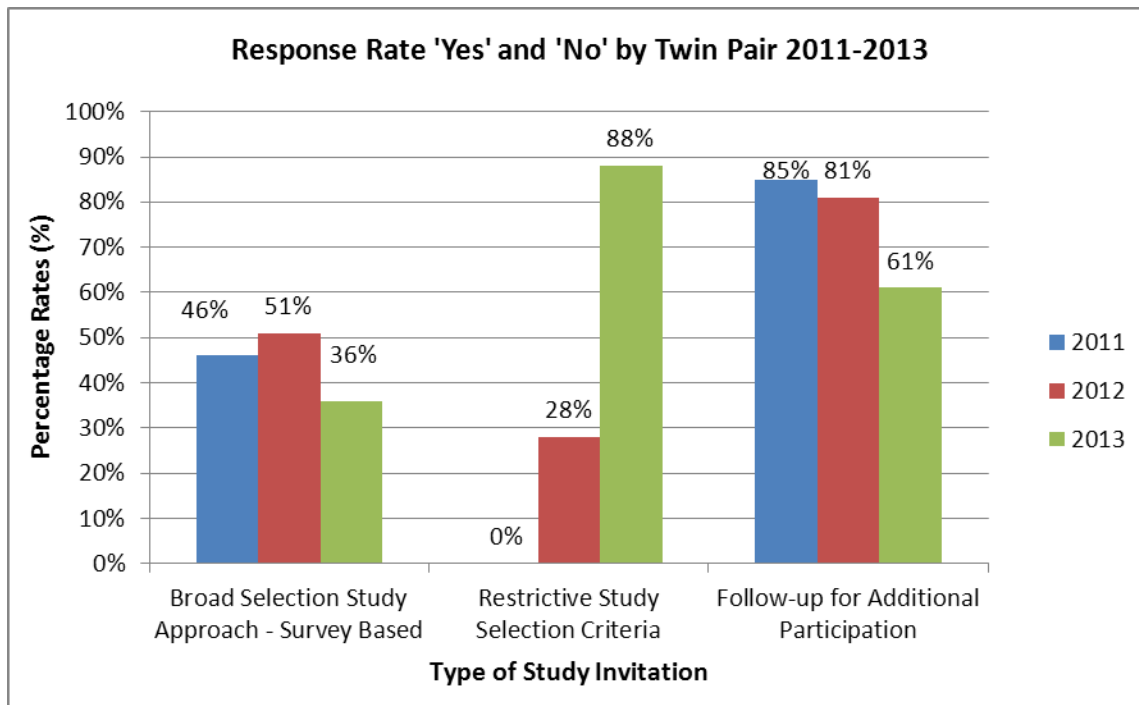


## 2. Study Response Rate - Yes/No Response Rate by Twin Pair

**Figure 2** shows a pair response rate for each year for studies with mailouts from 2011-2013. The twins must have responded Yes or a Hard No. The study response rates illustrate the response rates for each year, 2011 – 2013. The positive strong 'yes'/'no' response shows that the engagement of the ATR membership.

**Figure 2:** Study Response Rate – Yes/No Response by Twin Pair 2011-2013

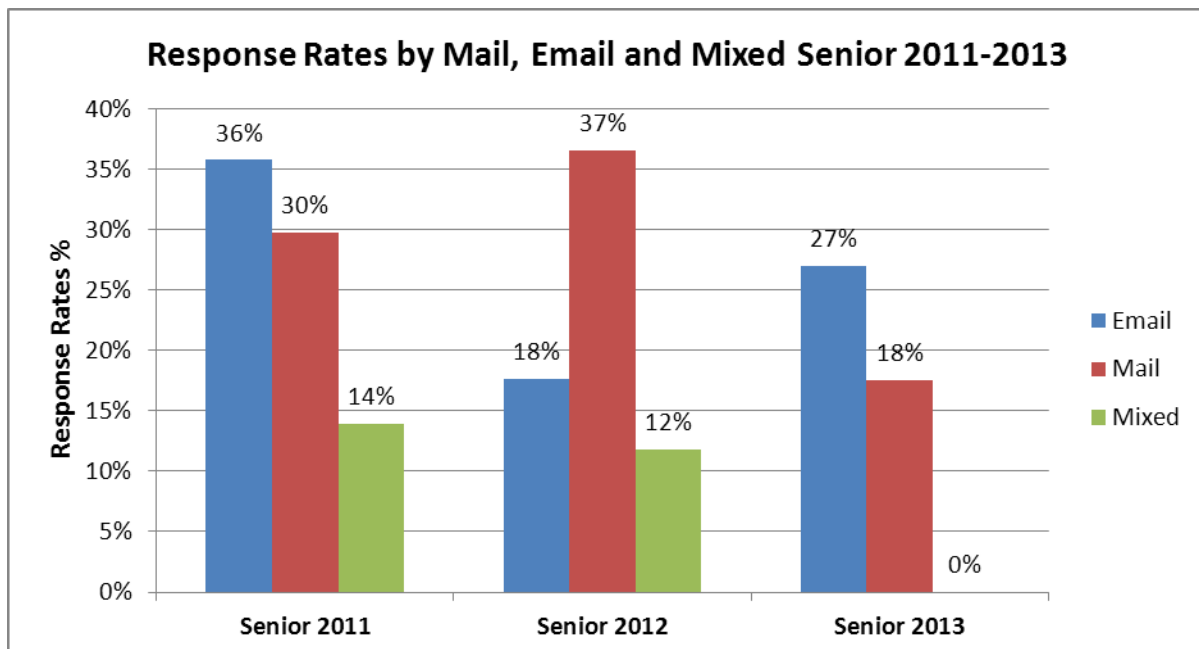




### 3. Study Response Rate – Twins who Responded Positively – Age Group, Mail and Email

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 3** below takes into account senior twin pairs who responded via mail or email during 2011-2013. Response rates for senior twin pairs appear to maintain an average 30-40% for email response rates which is encouraging for future study approaches.

**Figure 3:** Study Response Rate who Responded Positively by Age, Mail, Email and Mixed



#### 4. Study Response Rate – 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 2011-2013. 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, that is, excluded studies that re-approached already participating twins for further involvement. The response rates in the combined 70-99 age group are higher as the number approached and the positive response are equal to 100% are generally less than 20 twin pairs. The positive response rates for the younger groups are more responsive as a Yes or a Hard No. and the numbers are greater in number, thus the average 50% positive response rates in the 0-19 and 20-29 age years.

**Figure 4:** Study Response Rate – Age, Gender and Zygosity 2013

