Big data
How we’re using the vast amount of information available for research

Calculating the risk of cancer with whole genome sequencing

A year of advances in pancreatic cancer
I hope that you and your loved ones have had a wonderful festive season. Each new year at Garvan is filled with excitement and the possibility of life-changing research. This research is realised through the dedication of our scientists and the support of our donors.

Not everyone can say that they are part of the future of global healthcare, but as a supporter of Garvan and a reader of *breakthrough*, you are right alongside our researchers as they make exciting discoveries across all of our six divisions – Bone Biology, Cancer, Diabetes & Metabolism, Genomics & Epigenetics, Immunology, and Neuroscience.

Through the generosity of supporters, our researchers have had the funding to stay at the forefront of medical research. As they continue to produce outcomes that help to save lives, the reputation of the Institute has grown, which in turn attracts new researchers bringing fresh ideas and valuable insights from all over the world. Without the philanthropy of community members, none of this would be possible.

In one of our feature stories, ‘Big Data’ (page 6), we go into detail about how researchers are able to harness all of this information in the scientific world to start to improve health outcomes. Big data is changing the landscape of medical research and Garvan is passionate about staying at the forefront by utilising this information and the newest technologies available from around the world.

I hope that you will continue to be a part of this research. I am pleased to let you know that this year we will offer six public seminars. The seminars are a wonderful opportunity for you to come to Garvan and hear about the research taking place from our skilled and diverse team. If you can’t visit us, you are also able to view them online, with more details on the back page of this edition.

Thank you for the important role you are playing as a supporter of Garvan. Our breakthroughs, which are leading to better treatments and cures, are a result of the many generous donors who understand the impact that giving now to medical research can have on all of our lives in the future.

**From the CEO**

Andrew Giles, Chief Executive Officer, Garvan Research Foundation

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**Thank you, Geoff**

We thank Mr Geoff Dixon for the profound effect he has had on the lives of Australians through his support of Garvan. During his tenure as Chairman of the Garvan Research Foundation, Geoff has helped to inspire the generosity of the community. This has been an absolute necessity in supporting the work of the researchers at the Institute.

“Garvan in its current form would simply not exist without the work of the Garvan Research Foundation, chaired by Mr Geoff Dixon,” says Dr John Schubert AO, Chairman of the Garvan Institute of Medical Research. “We’ve benefited from, and are exceedingly grateful for, Geoff’s expertise and leadership.”

Geoff says, “I’m a passionate supporter of medical research, which is why I first became involved nine years ago. The Foundation continues to be very fortunate in receiving the ongoing generosity of individuals, foundations and corporations from across Australia and around the world to maintain Garvan’s research. The impact of this support cannot be underestimated.”

Geoff, we sincerely thank you, your wife Dawn and all of your family for your valuable contribution to Garvan over the years.

**55 years of breakthroughs**

This February we celebrate 55 years of solving the mysteries of disease. From origins as a small diabetes research department of Sydney’s St Vincent’s Hospital, to having more than 500 researchers studying more than 50 diseases and the only clinically accredited whole genome sequencing facility outside North America, Garvan has been instrumental in numerous breakthroughs that have impacted human health. For example, in 1973 we developed an insulin infusion technique that has saved the lives of people in diabetic comas. In 1993 we discovered that proteins called cyclins are involved in breast cancer development. In 2012 we opened The Kinghorn Cancer Centre with St Vincent’s Hospital. Thanks to support from the community, we’ve gathered a team of researchers recognised as among the best in the world. As we celebrate this milestone, we recognise it is one of many, many more to come. Thank you for joining us on the journey.

Professor John Mattick and Associate Professor Antony Cooper with Mr Geoff Dixon at a World Parkinson’s Day event.

The first laboratory at Garvan, 1964.
This image shows oil droplets formed in a cellular genomics process called Drop-Seq. Drop-Seq gives scientists the capability to understand how each cell is interpreting its genetic code (DNA) by looking at its transcriptome (RNA). The transcriptome, essentially ‘DNA in action’, helps define the function of each cell but also informs about the development of disease. Each oil droplet is a fully enclosed and separated reaction chamber. Here we see one co-encapsulating two objects: the dark circle is a microbead covered with barcoded primers, and the smaller, lighter circle is a cell. Through this method we can analyse tens of thousands, even hundreds of thousands of cells in a day in volumes as small as one nanolitre – a millionth of a millilitre – making the process economically viable. Droplet-based microfluidics are a major component of cellular genomics technologies and are fundamentally changing our understanding of cancer, immune disorders, neurological disease and many other normal and disease processes.

Rob Salomon
Technical Director, Garvan-Weizmann Centre for Cellular Genomics

For more information and a video about cellular genomics, visit garvan.org.au/cellular-genomics
Corporate sponsor paves the way for patient transport

Garvan is honoured to be supported by Sydney car business Suttons. Two new loan vehicles represent the latest in a longstanding relationship with managing director Laurie Sutton, who is also a philanthropic visionary of The Kinghorn Cancer Centre (TKCC) and The Garvan-Weizmann Centre for Cellular Genomics. Corporate sponsorship is a way in which businesses work with Garvan to change the future of medical research. A challenge that patients face is driving themselves to multiple ongoing appointments. The disability-accessible Honda Odyssey provided by Suttons City Honda will help TKCC provide a transport service to make it easier, more comfortable and less stressful for patients to attend appointments.

Making news

A YEAR OF ADVANCES IN PANCREATIC CANCER

Long-term survivors have ‘built-in immunotherapy’

Pancreatic cancer has a dismal five-year survival of just 6.8 per cent – but a select few live on for years, even decades. In these long-term survivors, the immune system sees the tumour as a kind of infectious disease – prompting it to attack and destroy cancer cells.

Over 150 Australian patients contributed clinical and genomic information through the Australian Pancreatic Cancer Genome Initiative (APGI). Garvan’s Professor Anthony Gill, chair of the APGI, says, “We’re able to go back to the tumours of these patients and to their DNA sequences, years later, and to look in detail at exactly what is different about their tumours.” This resource will continue to help researchers understand pancreatic cancer, with an end goal to improve survival rates.

Breast cancer drug could work in pancreatic cancer

A new breast cancer drug could be effective against some forms of pancreatic cancer, including metastatic cancer, Garvan researchers have found. Among 550 tumour biopsies, a cellular pathway known as Cdk4/6 was switched on in two-thirds, driving tumour cells to grow and divide.

“We know that the drug palbociclib switches off the Cdk4/6 protein, so we reasoned it might halt the growth of the many pancreatic cancers where this pathway is ‘ON’,” says project leader, Dr Marina Pajic. In preclinical models, the effects were dramatic, and were seen at all stages of pancreatic cancer progression.

Biosensor lights up

Garvan researchers have developed a biosensor mouse that gives a real-time readout of the rapidly changing ‘skeleton’ within cells. The researchers have watched cells respond dynamically to their surrounding environment in living tissues, including within invasive breast and pancreatic cancer.

“With the biosensor, we can see what each cell's minuscule skeleton is doing in a living animal, and how it responds and becomes addicted to its environment as diseases such as pancreatic cancer progress,” says study co-leader Associate Professor Paul Timpson, from Garvan’s Cancer Division.

This is the latest in a growing canon of ground-breaking biosensor mice developed by the research team, including one that makes it possible to watch in real time as pancreatic cancer cells prepare to spread beyond the primary tumour.

Here’s what you CanDo

When Rachael Lonergan was diagnosed at age 39 with triple negative breast cancer, she had to undergo surgery, chemotherapy and radiotherapy. Her sister was diagnosed around the same time with a rare sarcoma and their family was stretched trying to care for both of them. “Friends and co-workers would ask me what they could do, but I found that awkward, and my pride held me back from accepting the help I really could have used,” Rachael says.

After this experience, Rachael came up with the idea for the CanDo app, connecting people going through treatment with those around them who are eager to help and make their treatment as easy as possible. The app helps to eliminate social isolation and allows patients to focus on their general wellbeing, reducing the stress and anxiety around getting the ‘little things’ done.

Rachael founded CanDo to make dealing with a serious illness easier. CanDo helps to map out action plans, provides updates to a wider network with privacy settings and ensures that everyone who wants to help can contribute in a meaningful way. Visit candoapp.com.au to find out more.
A joint quest to stop pancreatic cancer

Jane Hemstritch and Garvan’s Dr Marina Pajic have formed a powerful bond over their shared mission to defeat pancreatic cancer

After tragically losing her husband, Philip, to pancreatic cancer in 2010, Mrs Jane Hemstritch threw her energies into raising awareness and making a difference in the deadly disease’s outcomes. Jane rallied the troops and formed ‘Team Phil’ in the Melbourne Marathon from 2011 to 2013. The team raised $328,000 for Garvan’s pancreatic cancer research.

Recognising the importance of ongoing funding security in medical research, Jane developed the Philip Hemstritch Fellowship in Pancreatic Cancer in 2013.

That’s how Jane met Marina.

Dr Marina Pajic leads the Personalised Cancer Therapeutics group at Garvan. Her research focuses on developing new and different personalised therapies for pancreatic cancer.

In 2017 alone, Dr Pajic was named the Outstanding Cancer Research Fellow from Cancer Institute NSW and led or co-led two significant research studies that explore innovative approaches to treating pancreatic cancer using existing drugs. In the first she showed (with Associate Professor Paul Timpson) how softening tissue around a pancreatic tumour can make chemotherapy more effective. In the second she demonstrated the potential of a breakthrough breast cancer drug to treat a common subtype of pancreatic cancer (see opposite). With a five-year survival rate of less than seven per cent, pancreatic cancer desperately requires further research and Jane has unquestionably made an incredible impact.

“I’m proud to be honouring my husband’s legacy as well as supporting an incredibly talented young researcher,” says Jane.

Dr Pajic says, “The Fellowship has been crucial for me to develop my ideas into sound projects with solid data, and recruit the best individuals to my team. There are significant funding challenges in medical research; we rely on philanthropy, sometimes exclusively, to get innovative ideas off the ground.

“But for me it goes far beyond the financial support. I’ve been incredibly lucky that Jane has taken the time to work closely with me as a mentor. She’s an eminent businesswoman and her knowledge and insights have supported my growth as a researcher,” says Dr Pajic.

For Jane too, there is an immense sense of satisfaction in supporting Dr Pajic’s work. “I can’t think of a better way to support medical research than to help a brilliant young mind make her ideas a reality, and make a difference in this dreadful disease.”
As we gather vast numbers of genome sequences, new approaches are transforming information into insight

In 2003, medical research changed forever. This was the year the human genome project concluded, which, after US$3 billion and 13 years, determined the first entire sequence of human DNA. From there, the sequencing cost per genome began a 15-year free fall to around US$1000 today. Meanwhile, the number of genomes sequenced has been equally dizzying in its ascent.

When Garvan acquired the Illumina HiSeq X Ten Sequencing System in 2014, it enabled us to move to the leading edge of medical science and the consequent transformation of healthcare. The clinical and research value of whole genome sequencing has since been amply demonstrated, such as in diagnosing rare genetic disease, guiding personalised treatment in cancer and enabling cohort studies to compare genomes en masse.

Indeed, personal genome sequences will soon become an intrinsic part of medical research and health management. While this has major implications for scientific discovery and the economy, it also poses an increasingly common challenge: as the sequences accumulate, so do the computing demands for analysing genomic data and its relationship to clinical information.

“Historically in medical research, generating the data has always been the bottleneck. But now science is changing,” says Dr Warren Kaplan, Chief of Informatics at Garvan’s Kinghorn Centre for Clinical Genomics. “We’re able to generate data much faster and progressively the work is happening on the analytical side of things rather than in the labs. If we’re going to continue to excel as a research institute we have to grow the pedigree of our data engineers and data.”

Big data describes the deluge of information that is a hallmark of our times. But how big is big? Let’s start with something small: one byte of storage. This is about the power needed for a single character. If we imagine a byte as a grain of sand, a kilobyte would be a pinch, a megabyte would make a small turret on a sandcastle, while a gigabyte would be a whole sandcastle. Increasingly, however, the data go-to unit is the petabyte, which satisfies the proverbial ‘long walk on the beach’.

The Garvan Institute’s genomics program has a presence of about three and a half petabytes at the National Computational Infrastructure facility in Canberra, which includes more than 15,000 whole genome sequences and grows by 50 genomes a day. Yet, this is only a fraction of what is becoming an unimaginably huge universe of all the data across the globe, which expands by 2.5 quintillion bytes every day. This is chiefly driven by the daily activities of casual data-makers the world over through search engines, social media, telecommunications, navigation and many other means. It is, in Dr Kaplan’s words, “an almost infinite scale of computation”.

The six billion bases in a genome sequence thus become six billion data points. Such detail allows for ongoing analysis as knowledge and needs evolve. “It’s almost exactly like the internet,” says Dr Kaplan. “No one wants to know everything about the whole internet at any given time. Today you might want to understand how Jamie Oliver makes a certain recipe, but by next Wednesday you want to know how a new model of car compares to the next one. Where all this sequencing data becomes really valuable is in enabling us to do Google-like queries across the genome, and we can start asking questions that are unprecedented.”

The ultimate potential of such information is only realised when it is integrated with parallel data sets, says Garvan’s Executive Director, Professor John Mattick: “The genetic data is not very useful without the clinical information.” Professor Mattick describes a multifaceted data landscape as an ‘ecology’ in which “we will have thousands, and soon
enough, millions of individual genome sequences with accompanying clinical records, supplemented by data from the Internet of Things [the huge range of connected electronic devices and sensors]. The more data we have, the more we can make correlations between our individual genetic idiosyncrasies and our health. That’s why we’re building capacity in analysis and artificial intelligence to prepare us for this new generation of medical research and medicine. Our job is to lead the way. Our job is to invent the future.”

The deep learning approach has much promise in interrogating the genome, which is, as Ersavas says, “an imperfect art at the moment”, with only about two per cent reasonably well understood. Dr Kaplan adds that there is greater potential still: “Where we think deep learning is going to become particularly valuable is that, in addition to the genome, there are many other ‘omes’ that could be computed, such as the epigenome, the transcriptome, the metabolome, the proteome, the exposome, and then on top of all of that are personal devices such as Fitbits, your social media presence and so on. How do we take all of this highly variable data and find meaning from it? This is where opportunities around artificial intelligence will become very interesting and useful.”

Currently Ersavas is working towards a process to interpret mitochondrial DNA as a pilot study for ultimate application to the whole human genome. This project is being undertaken in parallel with the Garvan-Deakin Program in Advanced Genomic Investigation, a collaboration with Deakin University’s Centre for Pattern Recognition and Data Analytics (PRaDA) established in March 2017. “This partnership aims to harness two great strengths: Garvan’s excellence in genomics and human biology, and PRaDA’s ability to search for patterns in complex data sets, with the mutual ambition of better understanding human biology, diversity and disease,” says Professor Mattick.

With the ongoing development of analytical methods, bigger data means better data, and Garvan’s thousands-strong library of genomes is recast as relatively modest in scale. As the volume grows, however, so do the avenues for transforming information into insight, and healthcare in turn.
Calculating cancer risk

It is always tragic when cancer is diagnosed in a young person. Now, thanks to whole genome sequencing, medical research is better able to predict an individual’s level of risk and offer management strategies.

At 28 years old, Hugh* was like any other person his age and living life to the full, with a young family, burgeoning career and enjoying living abroad in Australia. He had seen the GP about a lump on the side of his tongue, who prescribed antibiotics and then antivirals to no effect. Then came the evening of Monday 10 March, 2014. With wife Alice away sitting an exam, Hugh was eating dinner with his 18-month-old son when a crust of bread caught on the lump. “It was so painful I fell to my knees,” he recalls. “I called my neighbours and asked them to care for Henry while I took myself to hospital.” By Wednesday, Hugh was undergoing a biopsy. “The doctor said, ‘Don’t worry, it’s not going to be cancer.’ ‘I thought, pfft – cancer? Are you serious?’ But then on the Monday the doctor called and said, ‘You need to come in tomorrow.’ My heart stopped. I started thinking, I’m going to die, and about Alice and Henry and I just went into tears on the phone.” Within a fortnight, Hugh was undergoing surgery to remove the tumour and nearby lymph nodes, followed by six weeks of daily radiotherapy.

Though Hugh is unfortunately not alone in his experience of cancer, his relative youth places him in a minority, with the average age of cancer diagnoses in the Australian population at around 65 years. This raises the question, how and why does cancer arise in young people?

“We tend to think of cancer as a disease of ageing, where genetic changes accumulate over time,” says Dr Mandy Ballinger, Group Leader – Genetic Cancer Risk in Garvan’s Cancer Division. “If you are diagnosed with cancer at a younger age, it means you have a head start, which is generally a genetic predisposition. A number of papers have shown that the younger a person is diagnosed with cancer, the greater the likelihood that there are heritable factors at play.”

Certain genes are now well established in increasing cancer risk, such as variants in BRCA1 and BRCA2 and their relationship to breast and ovarian cancer, or in TP53 as the basis of Li-Fraumeni syndrome, which is associated with early-onset cancer in a number of organs. Increasingly, however, research is recognising the combined impact of multiple genetic factors as being significant in contributing to cancer risk, as enabled by whole genome sequencing.

“We’re approaching the point where we can look across all the genes and start to get a picture of the contribution of more than one variant to somebody’s risk – not only rare variants, but also the more common ones – and add all of those risks together and come up with a single risk score,” says Ballinger. Thus, the greater the number of harmful gene variants a person has, the greater their risk of developing cancer at a young age. The challenge for researchers is to pin down exactly what they are, how they work together and, ultimately, what we can do about it.

“This is the objective of the Genetic Cancer Risk in the Young study (RiSC), which Ballinger leads, and in which Hugh is a member of the cohort. The project sequences the whole genomes of people who have been diagnosed with cancer between the ages of 16 and 40, and analyses them for known cancer-causing features. “Ultimately, we want to identify the entire set of genetic variants that affect the risk of developing cancer,” says Ballinger. Over time, the unfolding medical stories of the cohort bring a wealth of clinical information to complement the genomic, with the potential to both validate and expand our understanding of genetic cancer risk. This paves the way for better monitoring, earlier detection and, as a result, improved outcomes for patients.

Among risk-management strategies, whole-body magnetic resonance imaging (MRI) is emerging as one of the most promising. In a major finding last year, Ballinger and colleagues validated this approach in Li-Fraumeni syndrome.

*The names of Hugh and his family have been changed.
I’m grateful to all the people who were in labs 50, 100 years ago, working to get to where we are today. If it was not for advances in medicine, I would not be here. Whole-body MRI detected early and symptom-free primary cancers in 10 per cent of participants, who were then offered lifesaving treatment. The significance of this is underscored when compared with the detection rate of the widely recommended breast MRI for carriers of harmful BRCA1 or BRCA2 gene variants, which finds cancer in approximately one in 50 people screened. Now, Ballinger and colleagues aim to assess the effectiveness of whole-body MRI in people with multigene cancer risk for whom, as with Li-Fraumeni syndrome, there is no way to predict where in the body cancer might arise.

Such insights would have enormous clinical potential, with whole genome sequencing centre stage in identifying people at high risk of cancer and providing scope to catch tumours at an early, treatable stage. “I think that within the next 10 years anybody diagnosed with cancer under the age of 40 will have the option of whole genome sequencing,” says Ballinger. “It’s becoming more and more obvious that this group is genetically predisposed, so being able to further identify those that are at the highest risk, interrogate them genomically and then offer them surveillance really is a no-brainer.” Would Ballinger extend this to assessing risk in people who have never had cancer? “In the distant future, it’s theoretically possible. But there would have to be lots more research to verify that the genetic profiles were validated.”

For Hugh, hopeful that his experience of cancer is behind him, RiSC represents a chance to help others. “Hopefully my contribution will be one tiny step in the bigger story of medical research,” he says. “I’m grateful to all the people who were in labs 50, 100 years ago, working to get to where we are today. If it was not for advances in medicine, I would not be here, Alice would be a widow and Henry would never know his father. This is my way of giving back.”

As one of the current generation of researchers contributing to this ongoing quest, Ballinger echoes Hugh’s sense of greater purpose: “At a young age, you often think you’re invincible, and a cancer diagnosis can be quite world-shattering. Having said that, many young people are resilient and find a strength that they haven’t had before. There are lots of things that can be done, as well. That’s the other side to think about. If we can understand cancer better, we can start to make steps to improve things.”

Leading the way in clinical genomics and cancer

The Genetic Cancer Risk in the Young (RiSC) study is part of Garvan’s Genomic Cancer Medicine Program, an innovative suite of research studies and clinical trials that match therapies, prevention and screening methods with individuals on the basis of their genetic information. In addition to providing enhanced understanding of cancer risk, such information makes it possible to select treatment approaches based on the genetics of the cancer, rather than where it is growing in the body, through a personalised treatment approach.

Find out more about the Genomic Cancer Medicine program at garvan.org.au/genomic-cancer-medicine-program
How did you come to be interested in polycystic kidney disease (PKD) and the potential for genomics to help?

I find PKD really interesting because it’s a relatively common condition and so it’s one we see in the clinic. In this disease, cysts progressively expand and damage the kidneys, eventually causing renal failure. As it’s an inherited disorder I often witness the powerful impact this chronic, lifelong disease has on multiple generations of the same family.

Why is it so important to find a better way of diagnosing this condition?

Previous methods to make a genetic diagnosis of PKD have been expensive and difficult for laboratories to perform due to six non-functional ‘pseudogenes’ near the specific disease-causing gene. As such, this testing has not been readily available, and patients are usually diagnosed only after they have developed a significant degree of disease. Our lab has worked to develop a more accurate and less expensive test using whole genome sequencing, and we hope it will help more patients. Earlier diagnosis allows patients to better understand the risks of passing on the disease before they start their families and allows better access to any potential treatments.

You’re also a clinical genetics fellow at Sydney’s Liverpool Hospital – how do your clinical practice and research intersect?

I enjoy working in both clinical medicine and research. Meeting patients in the clinic and seeing how much advances in science and technology have improved clinical care allows me to see the very real benefit of scientific research. Along with this, seeing the areas that we could improve in healthcare motivates me to keep pursuing research, so that we can provide patients with even better care.

What are your interests outside of work?

I love spending time with family and friends, cooking for them and eating too much food! Now that the warmer weather has arrived, I also love checking out Sydney’s amazing beaches.
A brother’s tribute

Peter Gallagher lost his life to prostate cancer on 19 September 2014, his father’s birthday. In Peter’s memory, his brother Terry seeks to make a difference to the disease that claimed him

Born 18 months apart, Peter and Terry Gallagher were close from the beginning and grew ever closer with the passing of time. Now, more than three years following Peter’s death from prostate cancer, Terry’s admiration for his older brother is undiminished.

“I’d like to honour him by telling his story and helping the Garvan Institute, through a gift in my Will, to find better treatments for this insidious disease prostate cancer.

“Peter was one of those people who couldn’t do enough, never took offence at anything,” says Terry. “He was an inspirational secondary school teacher, in English, history and economics. When we would walk down the street in Dee Why (Sydney), young people would stop him and say, ‘You’re the best teacher we ever had.’ He was a top-flight sportsman, played professional first-grade rugby league for Eastern Suburbs, 90-something games against the best of the era. Whatever he did he excelled in. Just an all-round good guy.”

Close to 100,000 Australians are living with prostate cancer, making it the most commonly diagnosed cancer among Australian men, and the third most common in the population after breast and colorectal cancers. It is also the country’s third most common cause of cancer death.

Prostate cancer affected both Gallagher brothers. Terry, with a low-risk, early stage of disease, was eligible for radioactive brachytherapy seed implants, which have successfully kept his cancer in check. By contrast, Peter’s cancer was already advanced when it was detected, and surgery could only remove part of the tumour. Doctors then found secondary cancers throughout his body and prescribed a chemotherapy regiment. Despite this, sadly Peter passed away within a year of diagnosis.

Peter’s experience left Terry seeking better solutions for prostate cancer. “I went on a tour of Garvan and learned about the research into personalised medicine, getting chemotherapy right so it doesn’t knock the hell out of people. I thought this was very worthwhile,” he says. The encounter motivated Terry to help progress medical research through becoming a Partner for the Future and nominating Garvan as a beneficiary of his Will.

“It all goes back to my brother,” says Terry. “I don’t want anyone else to lose someone close to them in the same way. My support of Garvan is a tribute to him.”

Thank you Terry for your future bequest and for granting Garvan the honour of Peter’s legacy.

If you would like information about leaving a gift to Garvan in your Will and becoming a Partner for the Future, please contact our Bequest Manager, Carol O’Carroll, on (02) 9295 8117.
Ask Garvan

Sandy from West Leederville in Perth, writes:

“My nephew has Prader-Willi syndrome. What’s happening in research to help this terrible condition?”

This issue’s answer comes from Associate Professor Alexander Viardot, Group Leader – Prader-Willi Syndrome and Genetic Forms of Diabetes, Diabetes and Metabolism Division

A: Thank you, Sandy, for your question about Prader-Willi syndrome (PWS). Garvan is one of the few places in the world actively studying this condition. While PWS is relatively rare it is also the most common genetic cause of obesity and has significant developmental impacts. It is characterised by intellectual disability, insatiable hunger and other physical disorders, making it a very challenging condition to live with for both the person and the family.

We have several new projects underway in PWS research, working towards understanding how critical muscle mass is for bone strength as well as the link between eating and inflammation in people with PWS. One of our past studies showed that the response of the autonomic nervous system to food intake is abnormal in PWS.

As insatiable hunger and obesity have the biggest impact on quality of life for people with PWS, our current research focuses on how appetite-regulating hormones and the speed of gastric emptying affect appetite sensation. We are also testing a new drug, already used for diabetes, which has potential to be the first treatment in PWS to curb hunger and improve eating behaviour. Such a discovery would immediately benefit people with PWS.

This research is a great example of the importance of studying rare diseases, as aside from helping people with these conditions, they also offer us unique insights into how the body works, which has the potential to improve everyone’s health.

Do you have a science question? Just ask!

Garvan’s experts are standing by to answer a selected medical research question in the next issue of breakthrough:

Email your question to breakthrough@garvan.org.au or post to Garvan Research Foundation

Reply Paid 68593, Darlinghurst NSW 2010

We cannot review personal health information or provide medical advice. If you are concerned about your health, please see your doctor.
In memoriam

Between 1 October and 31 December 2017, donations were made in memory of:

Bill & Barbara
Joanna
Mary
Les & Paula
Ron and Ted
Michelle
Mum and cousin
Natalie
Patty Adair
Trevor William
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Norman John
& Betty
Maureen Matthews
Brigid Jane
Maxwell
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McCreddie
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Patricia McIntosh
William McLachlan
Betty Constance
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Sidney Patrick
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Helen Richards’
grandmother
Mr Sydney Roberts
Martin Samociuk
Jim Sarikas
Ambrose Seagrave
Tan Chye Seng
Barry Sheedy
Robert Andrew
Smith
Lorna Star
John Steele
Edward Godfrey
Stephens

For further information, please contact:
Professor Jerry Greenfield and Dr Dorit Samocha-Bonet.

Clinical studies

Appetite study

Gastric emptying time is the time taken for food to leave the stomach and may affect your appetite and satiety. We are looking for healthy normal or overweight male volunteers who are between the age of 20 and 52 to participate in our trial investigating appetite regulation and gastric emptying in a genetic form of obesity called Prader-Willi syndrome.

For further information please contact:
Alex Viardot (02) 8382 2622 or email pws@garvan.org.au
(St Vincent’s HREC Ref 15/SVH/437)

Prediabetes study

Volunteers needed for a study testing blood sugar control in response to diabetes medications or weight loss. We are looking for men and women aged 20-70 years who DO NOT have diabetes, but have an increased risk of developing diabetes (e.g. have had an occasional increased blood sugar, or have had gestational diabetes, or a family history of type 2 diabetes). Study visits are conducted at the Garvan Institute of Medical Research and St Vincent’s Hospital, Darlinghurst, Sydney. Principal Investigators: Professor Jerry Greenfield and Dr Dorit Samocha-Bonet.

For further information, please contact:
(02) 9295 8215 or predict@garvan.org.au
(St Vincent’s HREC Ref SVH 17/080)