FIGHT MND.

Researcher.
PROF NAOMI WRAY

Research Initiative.
SPORADIC ALS AUSTRALIAN SYSTEMS GENOMICS CONSORTIUM (SALSA-SGC)

Where do you work?

The University of Queensland.

Can you give us a summary of your research background and experience?

The key questions driving my research are: Why do some people get disease and others do not? Why do some people progress in their disease faster than others? Why do some people respond to drug treatment and others do not? These are difficult questions, for which data collected on people - clinical, lifestyle and biological - are the key to generating evidenced-based answers.

My research group works on many common diseases, not just ALS, and we use statistical methods and genetic and genomic data. Working across diseases helps to bring different perspectives and ideas into our ALS research. We use "big data" approaches because the human genome is complex. There are about 3 billion letters (called base-pairs) in our DNA code, most of which are the same between all people, but about 10 million vary between people. My research focuses on these 10 million DNA letters and also on environmental factors that interact with the DNA.

What led you to pursue research into ALS/MND?

I was recruited to the University of Queensland in 2011 by Professor Perry Bartlett and he asked me to expand my research portfolio to include ALS. The previous five years had seen important technology-driven discoveries in other common diseases and he

was keen to see the approaches used in my research applied to ALS. The award of grant funding generated by the 2014 Ice Bucket Challenge cemented my commitment to ALS research.

How did the Sporadic ALS Australian Systems Genomics Consortium (SALSA-SGC) begin?

After starting to work on ALS, I realised that in Australia DNA was collected from less than 50% of those with ALS. The 2014 Ice Bucket Challenge, which raised $1.1M for research, provided an opportunity to change that. The Motor Neuron Disease Research Institute of Australia awarded these funds to the SALSA-SGC project, an Australia-wide collaborative proposal led by Ian Blair at Macquarie University and myself.

What excites you most about the big-data research resources SALSA-SGC is providing?

We have generated a resource that researchers from across Australia will be able to access for many years to come. The biological samples are stored for use in research today but are also ready for the technologies that have not yet been developed and the research hypotheses that have not yet been asked.

Data accumulated through SALSA-SGC will help my team, and others, investigate many hypotheses about causes and consequences of ALS that can be detected in biological samples. Since we are generating multiple layers of clinical and genomic data, it is indeed a big-data resource that will grow richer over the years.

People with ALS are very enthusiastic about research. The SALSA-SGC provides an opportunity for people with ALS to make a lasting gift for research that will help those that come after them.

What difference will this funding make to SALSA-SGC and ALS/MND?

When the Ice Bucket Challenge funding finished in May 2019, we began winding back our efforts because the funding available to us was not sufficient to keep SALSA-SGC going. This FightMND Award means that we can now continue and expand the SALSA-SGC project. We aim to include participants living regionally, in addition to those attending the major clinics. We plan to expand our data collection to include a self-report questionnaire for environmental risk factors. SALSA-SGC will dovetail with FightMND-funded clinical trials and will be well-placed to help quicker identification of those meeting specific inclusion criteria, which in future may include specific genetic markers. I am proud of what SALSA-SGC has achieved so far, which reflects the dedication of many people. We will build on these strong foundations in the next 3 years, making new research discoveries that will ultimately contribute to cures for, or better still, prevent, this most horrible disease.

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In 2015, Prof Naomi Wray was awarded the Ice Bucket Challenge Grant from MND Australia to establish and support the Sporadic ALS Australia - Systems Genomics Consortium (SALSA-SGC) for 3 years. SALSA-SGC brought together seven major MND clinics across Australia with the goal of establishing systematic and uniform collection of clinical data (symptoms, rate of disease progression), self-report data and biological samples from MND patients, creating a research bank that integrates clinical, lifestyle and biological information. The long-term goal was/is the generation of "layers" genomics data (DNA, epigenetics, gene expression across multiple tissues) to build a more complete picture of the complex genomic causes and consequences of MND/ALS. The by product is an important data resource to support many research projects.

SALSA-SGC has enabled the development of an online data collection platform where we work with clinicians and provide them with clinic reports. We have developed a portal which the clinical sites use to manage their patient cohort. Necessarily, it took time to establish the portal and to roll-out the system to clinics. But by the end of 3 years we had collected clinical data and DNA on 598 people with ALS and 223 controls.

This funding from FightMND provides continued support of the SALSA-SGC Program for the next 3 years. By investing in SALSA-SGC, FightMND will support researchers in the program and contribute to support for research nurses at 8 MND clinics across Australia, giving MND patients at each of these sites the opportunity to be involved in important research that advances the development of effective treatments for MND.

SALSA-SGC participant visit the research lab to see what happens to their DNA, all explained to them by SALSA-SGC project manager Anjali Henders.