Agilent Case Study

Australian Genomics Pioneers Clinical Data Sharing

If you work in the genomic medicine space you are probably familiar with the titanic challenges of the industry: data silos, lack of interoperability, and preserving patient privacy. Although large-scale data-sharing efforts hold enormous potential for driving the field forward, the infrastructure required to achieve that ambitious goal does not exist.

To address these challenges, two innovative medical scientists at Australian Genomics (a national network of hospital, university, research institutes), decided to start a variant curation and sharing project to address these challenges. So, in 2017, Emma Tudini, BSc (Hons) and Professor Amanda Spurdle both from the Queensland Institute of Medical Research (known as QIMR Berghofer) launched Shariant, a system for real-time national sharing of vital data. The Shariant project has an important mission: to help clinical genetic testing labs safely share structured insights on germline variants in disease genes, while identifying and resolving discordances in variant classification.

Establishing Shariant

A key, initial step was to ensure the buy-in of the laboratories that would be sharing this bioinformatic data: the clinical genetics labs of Australia.

"Our very first meeting we had with labs was an ad hoc meeting at the end of a conference. It was Sunday night, and the meeting ran from 6:00… it ended at half past 11:00 at night. People stayed on to talk about the topic, they were so engaged. That was the starting point for us. We knew that labs wanted to do something, they just needed to be brought together," stated Dr. Spurdle.

From that point, the Shariant team spent a significant amount of time figuring out how to automate the process of submitting structured data to the Shariant platform. They continuously modified the formats that can be exported out of Shariant to fit into a given lab's interpretation system. Since several labs use similar systems, the work required to get up and running at one lab often could be carried over to the next one. This helped Shariant generate momentum.

"Things are always slow when you start, but once we got [the labs] on board and showed that it could be automated, it really changed perspectives. It was nice to see the evolution of labs being excited about it – it just generates momentum," Dr. Tudini said.

Emma Tudini, BSc (Hons)

Amanda Spurdle, Ph.D.

Medical Scientists
Australian Genomics
Australia

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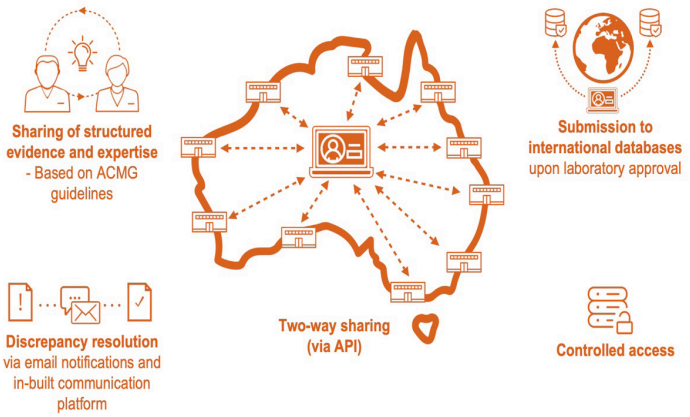

Figure 1. Illustration by Shariant from Australian Genomics, https://www.australiangenomics.org.au/tools-and-resources/shariant/

Because so many laboratories showed interest in the data sharing platform, Shariant continues to grow rapidly. They increased their network of clinical labs by 90% in the last year alone. A major focus as they scaleup is to improve automated systems for a more seamless user experience to transfer and upload data to the network. Dr. Tudini holds a regular user group meeting to gather feedback from the participating labs, who bring new, interesting questions every session.

Shariant Impacts Patient Care with Alissa Interpret

Dr. Spurdle recalled the moment Shariant's potential to impact patient care truly became apparent. A critically ill infant with an unknown condition had a sample sent to Victorian Clinical Genetics Service (VCGS), a Shariant member laboratory, for whole genome sequencing.

When VCGS consulted Shariant, they were able to see that a variant they had classified as a variant of unknown significance (VUS) had been tagged as pathogenic by another laboratory. Importantly, VCGS could review the evidence for the variant using the Agilent Alissa Interpret platform, the software that helps to power data exchange with the Shariant system, and were able to identify this variant as the recessive condition. In a matter of hours, they found a diagnosis for the patient and made a life-changing impact on the family.

What's next for Shariant?

Dr. Spurdle is excited by the impact that sharing clinical and variant information across the bioinformatic community, through Alissa Interpret, brings to diagnostic testing. As more laboratories join Shariant the sharing of variant information will enable all members to "…provide better and faster classification for their variants".

Today, the team is focused on germline applications. But coming on the heels of their success with genetic disorders, they have turned an eye toward cancer. Amanda cheekily shared with us, "If I can clone myself, we may be able to get that project going toward the end of the year."

What does the future hold for Shariant? If past performance is any indicator, we can think of a few things: improved quality of care for Australian patients, faster and better variant curation with every lab in the Shariant network, and a big step forward for Australian Genomics' mission to accelerate the real-world translation of genomics into the clinic.

"We believe that this is going to lead to more accurate and faster diagnoses."

– **Dr. Spurdle**

Learn more about the Shariant network at:
https://www.australiangenomics.org.au/tools-and-resources/shariant/
https://shariant.org.au/accounts/login/

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