Summary of the 2022 National Rare Disease Summit

Rare Voices Australia's (RVA) Chair, Joanna Betteridge, delivered a summary of the 2022 National Rare Disease Summit (the Summit) to conclude the two-day event on 11 and 12 November. We have transcribed Joanna's summary below. You can view the Summit Agenda here and speaker biographies here.

Summit Summary

There has been such an extraordinary buzz being back in person over the last two days and I hope those who have logged in virtually have picked up on this positive wave. In person the event has been extraordinary.

The theme of this Summit is implementing the National Strategic Action Plan for Rare Diseases (the Action Plan) in relation to each of its three pillars:

1. Awareness and Education
2. Care and Support
3. Research and Data

How well did we set out what we hoped to achieve, and what did we learn along the way? We asked ourselves these questions:

- How have we progressed so far from launch [of the Action Plan] in February 2020, and of course with the interruption of COVID-19? Has this stalled the implementation?
- Nicole has called on us to consider what is missing. What else do we need to do?
- How do we see [the Action Plan] progressing in the next 10 years in relation to each of the three pillars? How do we embed person-centred care into the implementation?

Opening Session and Panel Discussion: A Person-Centred Approach to Implementing the National Strategic Action Plan for Rare Diseases

Nicole observed that RVA had reached its 10-year anniversary, a milestone, but [there's] so much more to do.

We've been resilient as a sector, made solutions when there weren't any available, and those living with a rare disease are both the learner and the expert. And all parts of the sector have different strengths and expertise and we reminded ourselves of the importance of listening, asking questions, teaching each other and collaborating.

I want to spend a few minutes thinking about what we learnt from the commencement of the Summit.

We started by grounding ourselves in the overarching theme of our conference, person-centred care. We heard from people who experience living with a rare disease day by day. We had inspirational talks from some of our RVA Ambassadors, Beck Webber, Katie Alexander, Lachy Beckett and Tammie Rees.

As Katie reminded us, they are experts by experience. We were given insights into the challenges faced, and how much larger the issue is than just obtaining medical care for rare disease.

From Beck we learnt that there are additional challenges when others with your rare disease are family members. What are the implications of that in relation to putting together a care support team when often, the family is the care support team? What happens if the family is also affected? In addition to the challenges there are also the challenges of living in a rural environment. A theme that's come across from quite a few people.

The glue holding you together in the darkest times is often the family and friends said Beck, but the supporters also need to be supported. It's about the mind plus the body. Your support team needs to include a vast array of different professionals, as well as people who will support your goals and quality of life. Psychologists, occupational therapists, physiotherapists, speech pathologists, general practitioners (GPs), neurologists, the list goes on and on.

She talked about walking into her GP of 35 years and being told, "I don't do rare disease". Where do you go with that when you live in a place where there aren't specialists that you can go to readily? The education of GPs was a big issue with Beck.

Katie told us about some of the challenges she'd faced, both personally and medically. Again, people in rural Australia experience worse outcomes when living with a rare disease. There's a long-term failure that she's experienced in diagnosing her rare disease. Her nearest public hospital doesn't have a department willing to take on her rare disease. She spends, I think she said, $3000 annually on private health insurance. To reach her infusions in the city was $400 for a taxi fare. This all has a marked effect on her quality of life. There was some relief from the National Disability Insurance Scheme (NDIS), but still, how do you afford some of these things on a disability support pension?

The gut-wrenching reality of living with a rare disease, including having a child with a disability. Your vulnerability to family violence, having others dismiss your symptoms as not valid and all the while living with severe pain and disability, unemployment and at times homelessness. It's a very bleak picture. Doctors failed to believe and act on her complaints for over four decades, and she noted that invisible illnesses are extremely stigmatising. The importance of people listening, believing, and acting, and the fact that a failure to diagnose can lead to significant premature deaths. We have world-first acute care, but she said it's dehumanising and hard to bear over time. She has been carved into tiny little fragments, all the different medical specialities; they exist in silos but not collectively. Disadvantage and

distress go hand in hand. Allied health workers in her case have allowed her to be an advocate for her peers and have helped her live her best life.

Lachy's experience started at three years of age. He remembers being unable to walk between the ages of three and six and being in so much pain. He emphasised the importance of more education around rare disease. He was made to feel at times grateful for the help he received, but he can't help that he needs that help. How appalling that someone is made to feel they have to be grateful for something that should be automatically provided to everyone.

Society needs changing. The person-centred approach is so important because rare diseases affect everyone differently. Everybody has their own unique set of challenges.

Tammie gave us a window into the challenges faced by parents of children diagnosed with a rare disease. Her daughter was diagnosed with Maple Syrup Urine Disease in the first two weeks of her life. Thank goodness for the newborn bloodspot screening test that was done at birth, or shortly after. The exhaustion, the learning curve that you have to be on. The three-hour feeds. Three feeds an hour, measuring formula. The problems with the lack of expertise in pathology, and again in a rural setting. Having to drive 30 minutes to a regional hospital. Traumatic admissions to hospital. People in rural hospitals not knowing how to do things and having to do them yourself. Making up the solution for the nasogastric tube etc. Ignoring directions from parents who are the experts as to what to do. The discovery via a social worker's recommendation of the real source of personal support, the metabolic support group and the patient support group. The discovery of this. What a tremendous difference to have other families to talk to. To have other people who understand what you are going through. Even though the children may have different symptoms, it fills in the gap left by the health system.

It's exhausting being the patient, the educator, the mentor and so on. You are carrying the load, and you're educating the doctors in a great many cases. The GPs in particular. She suggested a great way forward. Get the Minister for Health and Aged Care and the Minister for the NDIS in a room with Nicole. Yep, at the same time.

Panel Discussion: Celebrating RVA's 10 Years of Rare Disease Advocacy and Shaping the Next Decade

Dr Mike Freelander MP then talked to us about his life experiences as a medical professional and his political role, and how much the landscape has changed for rare disease. It's so important to hear the stories from the political front. The *New Frontier Report - Delivering better health for all Australians*, how to advocate for people in relation to evolving treatments. A centre for precision medicine and rare diseases within the health department, he would prefer that to be an independent body. But, he noted, we are now seeing some curative treatments, and there are treatments for things that were not able to be given treatments for earlier. So, he felt that we were on the cusp of major change, and we were changing the world for future generations.

We heard from the perspective of one of RVA's Partner organisations and Sean Murray, how very complex rare diseases can be and the issues that arise when there's a genetic cause. The family connection. Being able to trace things that have happened throughout your family that were never diagnosed before. He talked about the power of uninformed optimism, to have a condition where there are no treatments or cures apart from symptomatic treatments, but now in other parts of the world, a body of research providing hope [but]that is not legal in Australia yet. So, what do you do on your journey of advocating for legislative change? The importance of the Action Plan in that. The legitimacy given to rare disease groups now seen and heard through the Action Plan and RVA's work. The care and support specific references to Mitochondrial donation as an example in the Action Plan. So, when they are advocating for this, they can point to the Action Plan, and say, "Look, here's an example of our situation." What does the next decade look like? He talked about RVA continuing to be the unified voice, bringing together and seeing the common ground, supporting and learning.

From the medical perspective, Dr Kaustuv Bhattacharaya noted that the Action Plan encompasses awareness. All health workers need to be made aware. We keep discovering new conditions, we have to find what people need and put together a package around it. The Action Plan he talked about is a framework or a scaffolding on which you can hang whatever you need. It's your tool to use so that needs are met and become a reality for patients.

From the medical research perspective, Dr Carol Wicking talked about the difference that the Action Plan has made to research. It has raised awareness in the community and among the government and funding bodies. It's made raising funds easier. The message is now there are a lot of people living with a rare disease in Australia. The perception in the past that research would be based on how many people suffer from a rare condition, well it's no longer just a handful of people living with an individual rare disease: it's two million Australians. The importance of gauging early in a research program and helping to set the agenda with clinicians and researchers being patient-centred and collaborative. She said funding bodies are now expecting to see those partnerships, and the Action Plan has helped with that. The importance of data, all types of research, consistent collections, safe storage shared within a good ethical framework. Registries are integrated and interoperable.

Dr Kathryn Evans talked from an industry perspective about how many people are touched by rare diseases, and there are many others that have connections with those people. Her company also works with patient groups but working at the national level with RVA enables them to bring stakeholders together around a common purpose with common ground, looking for the best outcomes for those living with a rare disease. There has to be persistence, and there has to be pushing of boundaries, but it's a privilege to be in the room with those working together to develop the Action Plan. She talked about being involved in Parliamentary Events and policy consultation with all stakeholders in the room and RVA bringing people together. The Action Plan, she said, has helped inform her company's approach to rare diseases and more widely helps them to anchor what they do and what

they say. She talked about having hope for the future, and we all work for that hope and for equity and the same opportunities for everyone to have the best lives possible. She said together, we can transform lives.

Panel Discussion: National Strategic Action Plan for Rare Diseases – Awareness and Education

Megan Donnell spoke to us about a foundation that communicates to the community and all its aspects in terms they can relate to and understand in childhood dementia. Raising awareness is key to achieving outcomes. A suite of information for health professionals. Lived experience. She talked about 50 families being involved in a range of activities. Publishing what families tell them are the issues. Outlining calls to action. Finding that hook for media to move that awareness forward. Campaigns like 'Face It', painting faces and memorable awareness raising campaigns. She said every activity has to be informed by lived experience.

Dr Amanda Choo bought us up to date with what's happening with the Rare Awareness Rare Education (RARE) Portal. The purpose, the key points of difference, its customisation for the Australian context, serving as a hub for people to access information.

Dr (Elizabeth) Emma Palmer gave us an update in relation to the Rare Disease Awareness, Education, Support and Training (RArEST) Project. A fairly unique partnership, she called it, with RVA and a group of clinicians, specialists and educationalists across Australia, embedding research into clinical practice. Principles of person and family-centred collaboration and capacity building. She talked about that very memorable comment about hearing hoofbeats. Not just looking for horses but looking for zebras as well. Signposting great resources, it can be overwhelming, but together we can make such a difference. Emma talked about the importance of the network approach, where you can't emphasise one speciality. It has to be a multi-disciplinary clinic, all the different specialists there in the one place. A model of care that families want. She talked about an ultra-rare clinic. Coming in on one day, all having a role to play. Allied health. Metrics that matter. Assessing the things that are meaningful, and what matters to people. Power in the use of terms that resonate with people. Collection of data and engaging with people who have rich natural history data.

Dr Falak Helwani bought us up to date about a sector-wide progress scan, monitoring and evaluating progress is the responsibility of all of us. We are just at the start. The RARE Portal is in the Action Plan and will cater for different stakeholders, communities and clinicians and work with all of the patient groups for the sector, by the sector. Bringing all the research together, and that will be launched next year.

National Strategic Action Plan for Rare Diseases: Care and Support

Lisa Schofield PSM spoke to us about the need for care and support to be more integrated. Education, employment, and every facet of life are affected, including mental and physical health. Enabling patient and support networks and some of the enablers we are now seeing, such as the RArEST Project and the RARE Portal, childhood dementia, newborn bloodspot screening, and national policy frameworks, the government is committed to improving these things. State and territory operated programs. Working from evidence, lived experience and ethical frameworks.

Dr Falak Helwani talked about the *Rare Metabolic Disease Workforce White Paper: Towards a Strengthened Rare Disease Workforce for Australia*, sustainable systems, again part of the Action Plan. Its key findings include poor recognition of the metabolic speciality, and a lack of clear and formalised clinical care pathways. Variations in the composition of maturity of metabolic services and where you live being a factor. Potential devastating consequences of living in a particular place. Critical workforce shortages. Insufficient resources for clinical trials. Lost opportunities. RVA has put together a strategy to tackle these things, and that will be published in due course.

Jo Watson talked about questions of access beyond one area. What is sustainable? Equitably available to everyone and can be repeated going forward. She talked about the Symposium on health technology assessment and the ongoing dialogue involved.

Louise Healy talked about the importance of mental health support for people living with rare diseases. Unambiguous evidence is that we are at much greater risk of developing mental health issues than the general population. Being the expert, and having to educate doctors, being an additional burden. Only one choice of a clinician, nowhere else to go in many cases, particularly in the rural environment. Mainstream mental health organisations don't understand living with a rare disease. Time is a barrier. There is often no time to look after yourself. Mental health first aid training and the fact that family and carers also need care.

Fiona Lawton talked about RVA bringing together a range of partners to meet with the National Disability Insurance Agency (NDIA). We were able to advocate across the sector, the first of its kind where they heard directly from the patient. A robust discussion, eligibility, access and planning issues were discussed. In many cases, gaining access to the NDIS requires more understanding of what evidence is needed by the consumer to access NDIS services. Some patient groups have developed resources, and they are unique to ensure they include the matters that the NDIS need to see in order to make up their mind about access. So, there is work to be done there. The intersectionality with rare diseases and poverty and domestic violence and all these other issues. Systems geared around language. Knowing the right words to say. Coach the clinicians to use the right words to open the door to services that are needed. Concerns about the cycle of funding review, you could lose your funding at any time. And this level of uncertainty hanging over people. Data needs to be far more transparent and if the data is not captured, it can't inform policy. The

RVA initiative to have been at the table. To collate and consolidate core issues to share stories with the agency. And the hope that this means they can tackle and strengthen the NDIS.

National Strategic Action Plan for Rare Diseases: Research and Data

In the research and data stream, Prof Adam Jaffé talked about the Action Plan requirement, and why there is a need for research. Placing consumers at the centre of participatory research. Identifying the shared national priorities, not focusing on rare individual diseases. There are common challenges. He talked about himself as being disease agnostic, which is a nice term. What are the common needs? RVA's Scientific and Medical Advisory Committee (SMAC) and other experts are coming together and looking at scope, mapping it out against the Action Plan all across the rare disease sector. Developing a top 10 list for research priorities, looking at where this has been in other countries. For example, with asthma in the UK. Coming up with that example of where the families said, "We want to know what breathing has to do with asthma?" And the researchers are thinking that is not something we would have followed, but it was patient-led, and it resulted in an interesting paper and action in relation to talking about the role of breathing in controlling asthma. So, your set of priorities can also sway your funding and you can map granting applications against the Action Plan framework to add to the legitimacy of your funding applications.

Prof Susannah Ahern talked about a registry audit that monitors undertaking comprehensive and clear recommendations in the Action Plan and the fact that registries can help to address the collection of data, but you can do more than that. You can develop population level accurate data that can be used for a number of purposes. There are 76 very disparate registries of rare diseases in Australia and they are disparate in terms of what they collect and where their data comes from. Most have demographic diagnostics and treatment info, but there are some real barriers to developing registries. There is funding, and there is the burden of data collection. There is consent. There is ethics. There are a whole range of issues. And the problems with the rare disease registries are that they have grown organically and are difficult to fit into a model. The conclusion is that they are feasible and they are valuable. They need to be clinician-led and patient-centred. There needs to be low burden approvals, coded data that can be extracted, and multiple data uses with opt-out options. And while we worked on consolidation, she said, let's start capturing everything, from now on, going forward and particularly improving diagnostic coding.

Prof David Coman spoke about an example of a national research clinic for Ataxia Telangiectasia (AT) where collaboration and listening have a profound effect. AT is a particularly difficult rare disease. Every couple of months people with AT would come in, and you would see 40 people, and you'd learn a lot. It has driven research as well. The researchers have become intimately familiar with what's important to the families. They come to the clinic, they know the families. They've had information from an overseas conference that has led them to do trials on the repurposing of a drug. The research gave them enough information to apply for a funding grant. And this was something that was

patient-led. The patient said we want you to work on this. It needs a clinician to pull everyone together, so it is very much a collaborative space.

Panel Discussion: Flying the Flag for Rare Diseases

Clin/Prof Gareth Baynam updated us about Indigenous rare disease care, it needs to be a national approach. There is incredible work going on. There is an integrated, coordinated, interagency care program but inclusion doesn't equal equity and the big question is, how do you make care fair? The same dynamic with Indigenous health, thousands of years of knowledge and wisdom can and should be applied, but you've got what he called the triple whammy; remote, Indigenous and having a rare disease. You start where the burden is highest, and where the most equity can be achieved. You include capacity building in the health system, and Aboriginal health primary care. You increase partnerships with universities, institutes, centres and an evolving consortium of works. And a national Indigenous network stimulus to get this happening and involving the families. Having a voice within government, primary care capacity building, a centre for rare and undiagnosed diseases in Western Australia (WA), the Rare Care Centre. Improving the lives of those living with a rare disease. Integrated health care delivery, a model for other states and territories. It's not about research: it's about treatment and care. The Action Plan has been an incredible lever in relation to that, as has been the United Nations (UN) Resolution on Addressing the Challenges of Persons Living with a Rare Disease and their Families. Governance is co-designed through reference groups, youth reference group, advisory groups, and the model is the whole of the person and their life experience. There are internal referrals from hospitals and other sources and accredited training for GPs.

Prof John Christodoulou talked about the diagnostic odyssey and explained why it can take years to reach a diagnosis. How do you try and short-circuit some of those diagnostic barriers? Being able to remove clinical diagnostic data to be used in another area. Huge barriers to overcome a completely unsexy area, he said. Almost impossible to get funding, but they've been building resources at the local level and looking for new ways to achieve the end result. And he talked about the Undiagnosed Diseases Network (UDN) Australia, working with undiagnosed diseases. Looking to recruit up to 600 individuals apparently undiagnosed, upskilling clinicians and exploring different approaches. Developing streamlined processes for sharing data around the country. A very encouraging initiative. And they are hoping that they can identify the next modalities needed to reach a diagnosis. They can leverage funded projects and it will be collaborative and person-centred, translating research and innovation into clinical care.

A/Prof Jodie Ingles talked about their rare disease program bringing together individual groups within the Garvan Institute. Access to the right patients and asking the right questions. Patient-focused, inclusive, collaborative and diverse. A lot of people have no diagnosis. What is the underlying genetic cause? A lot of issues to consider. Make it affordable, cost-effective, and have an impact. Provide tools for support and be inclusive and diverse in its population.

Prof Adam Jaffé talked about the development of a large patient-centred network in Sydney. Increasingly, genomics has been integrated into specialist care, the therapeutic pipeline. He talked about find it, fix it, prevent it. He gave us an example of the power of early detection and treatment in relation to spinal muscular atrophy. One injection before three weeks of age can save a life. In relation to cystic fibrosis, early detection completely changed the outcome of children with cystic fibrosis who can live to old age taking a tablet a day. Embedding this approach into health care would completely change the model of rare disease care. You need to build on healthcare systems and strengthen them, and we need to think as a group about how we can do that and how centres of expertise can assist.

Prof John Rasko talked about the next phase to use genes as therapeutics and use them as drugs. Many diseases have genes as their root causes, and they are driving clinical trials to use genes as therapeutics. Many colleagues didn't think they would succeed, but they are now glimpsing the future of success and addressing the goals of the Action Plan and bringing together a group of busy clinicians. Put out a call, asking them to come in if you are looking after someone with a rare disease. None of them identified they were looking after rare disease. They just identified they were looking after their particular problem that arises in that rare disease. Cardiac problems or whatever it may be, once you pull them altogether and get them thinking in a different space, that opens up opportunities. And looking at people with rare diseases, the problems that occur when you transition out of childhood, when you've got a rare childhood disease and how assistance often dissipates at that point. So, creating a core group of clinical champions, gaining the trust of medical specialties translates expertise and knowledge across the whole. The Sydney Local Health District embraced all of the organisational approaches. They're the first ones doing this. Partnered with families and all of the health sector but also with industry. Enormous investment by industry. A vibrant culture of technologies. You need to invest now so that it doesn't end up offshore. And each person is treated as unique, not just rare.

Themes at the End of the Last Two Days

As Falak has said, we've seen amazing participation. RVA will take all of the feedback and engagement from the different voices and stakeholders to identify gaps and a willingness to problem-solve. We will take a closer look, and we will report back to the sector.

So, what are the themes at the end of the last two days?

The critical importance of all aspects of care is holistic and person-centred and the power of strategic collaboration.

The Action Plan implementation has been an opportunity we've all taken up and used to great effect, and when you bear in mind that it's only been active for less than three years, that is three years during a global pandemic. There is so much positive movement forward that's been achieved in that short and difficult period of time. Imagine what we will achieve

in the next three years, as hopefully, we move out of COVID-19. So, the hope for the future is really an amazing reality.

I was chatting to Andrew over lunch, and I asked him to summarise what he thinks the major issues and themes of the day were, and he came up with two which I thought were terrific. The first was, we are doing this all together. We are in it together. The second was that these two days have been powerful. So, I thought that they would be a great way to finish the 2022 Summit. Thank you all for your extraordinary engagement.