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Rare Disease Registries: A Call to Action

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ABSTRACT

When registries collect accurate clinical data over extended periods of time, they can act as fundamental support structures for patients and their families, and powerful cost-effective instruments to support clinical trials and translational research that improves quality of care, quality of life and survival. Registries are critical for Rare Diseases (RDs) with low prevalence and propensity for variation in treatment and outcomes. Rare Voices Australia (RVA) is leading a call for action to the research and clinical community to prioritise RD data collection and develop an integrated RD Registry strategy for Australia. Financial, operational and governance challenges exist for establishing and maintaining RD registries. As a multidisciplinary team whose interests converge on rare diseases, we highlight the need for the establishment of an Australian RD Registry Alliance. This “umbrella” organisation will: i) bring together existing RD registries across Australia; ii) establish National RD Registry Standards to support inter-operability and cohesion across registries; iii) develop strategies to attract sustainable funding from government and other sources to maximise the utility of existing RD registries and support the development of new RD registries. The most important role for the Alliance would be to use the RD registries for translational research to address current knowledge gaps about RD and to improve the care up to 1.5 million Australians estimated to live with RDs.

Rare Disease Registries: A Call to Action

The burden of rare diseases is unacceptably high for patients, families and the community in Australia and needs to be recognised as a national priority, with rare disease collectively affecting over 1.4 million Australians [1-3]. Instead the impact of rare disease remains largely hidden due to inadequate information systems of a healthcare system that is designed to respond to individual diseases with much larger patient numbers.

At present, even the most basic information about many RDs, including incidence and prevalence, are merely estimates given the absence of systematic patient data collection (i.e. registries) for most RDs in Australia. The need for a National RD Registry Strategy has been highlighted previously [1,2,4,5] and more recently echoed by Rare Voices Australia (RVA), the national alliance of people living with RD. Out of its Rare Disease Summit, RVA released a public Communique in 2014 highlighting the need for a national coordinated and collaborative approach, better data collection and information use for RDs, with coordinated care and equitable access to services, diagnostics, treatment and research for RDs. The Communique has now been endorsed by over 100 patient organisations [6] and used to inform the recent “Fair for Rare” campaign [7].

For progress to be made towards achieving a National RD Registry Strategy in Australia we need better coordination and alliances among existing RD registries and stakeholders. Stable, long-term government funding also is needed to ensure sustainability and to facilitate the significant networking and coordination effort required. This coordinated approach to managing multiple RD registries under the same umbrella is likely to provide significant cost savings while maximising impact.

Data collection is a critical aspect for RDs because the inherent features and lack of patient numbers result in a lack of data, evidence and knowledge. This has negative impacts including delayed diagnosis, suboptimal patient care, and limited access to treatments, either because they have not been developed, or where available due to high costs and/or lack of Government reimbursement. Registries are systems of uniform data collection to evaluate specified outcomes for a population defined by a disease, condition, or exposure, typically spanning different clinical or academic partnerships over an extended time period [8,9].

Registries are particularly important for rare or poorly understood diseases with small patient numbers, complex delayed diagnoses, a propensity for variable standards of care, and limited treatment options [1,2]. Registries act as a central networking point for all stakeholders around a particular RD, including patient advocacy groups, researchers, clinicians, industry and Government. Registries, if populated with accurate and high-quality clinical data over extended periods of time, enable health service planning, epidemiological research, clinical trial recruitment and post-marketing drug surveillance.

RD researchers and registry custodians from across Australia recently agreed to develop a National RD Registry Strategy, and given the limited resources and funding identified the following priorities:

- a) Create an alliance of existing RD registries in Australia through the RVA-based network to begin sharing best practices, establishing standards, cohesion and higher inter-operability;
- b) Conduct an audit of appropriate existing RD registries and databases (this is underway with examples of Australian RD registries and databases provided in Supplementary Table 1);
- c) Define how best to establish a single National RD database to act as a central repository for RDs, and define the consent, data capture and governance models, following FAIR principles [10];
- d) Develop strategies to minimize barriers for establishing new and more comprehensive registries for individual RDs or related disease groups;
- e) Consolidate a coordinated effort to attract sustainable government funding for RD registries.

Epidemiology & Impact of Rare Diseases

Rare diseases (RDs) can be defined as disorders with a prevalence of $<1/2,000$ [11]. Some 6,000-8,000 known rare diseases, approximately 80% of which are genetic in origin, exist across 27 different disease categories, many with childhood onset, and the remaining 20% comprise varying different aetiologies [1,3,11]. Although each individual RD is by definition infrequent, the combined health burden of all RDs in Australia is significant, with an estimated $\geq 6\%$ of the total population (or 1/17 people) affected, equating to at least 1.4 million Australians, i.e. similar to the number of those in Australia with diabetes [1-3] and matching the prevalence of RDs in England [11]. However, these estimates are limited by a scarcity of robust representative data, due to the lack of registries and data collection systems.

Some RDs such as cystic fibrosis, have benefited from long-standing Australian registries, however, there are no standardised data collections or registries for most disorders. Therefore there is uncertainty about basic information including; the number and prevalence of RDs in the population; how many patients with a RD are receiving treatment; variation in the quality of care and health outcomes across the health systems; and the overall burden of RDs nationally. Without registries or better patient data collection systems, it will continue to be difficult or impossible to ascertain even this basic, yet crucial information for RDs in Australia.

Given the existence of so many different RDs, it is impractical, cost-prohibitive and unrealistic to establish stand-alone high quality clinical registries for each individual RD. Modular approaches and shared platforms are required to collect data on multiple RDs or the grouping of related RDs. A central database or repository for many RDs in Australia would provide an important step towards better measurement of the burden of RDs on the health system, and would provide basic information like incidence and prevalence for RDs. A National RD database would not act as a clinical quality registry measuring outcomes and clinical variation for each RD, but instead would be a starting point to help collate basic data and then identify areas in need of further development and focus.

A National RD Registry Strategy should be consistent with the 10 key principles for RD Registries jointly declared by the European Organisation for Rare Diseases (EURORDIS), the National Organization for Rare Disorders (NORD) and the Canadian Organization for Rare Disorders (CORD) [9] (Table 1).

Table 1. EURORDIS-NORD-CORD principles for RD Registries

Registries should:

- Be recognised as a priority in the field of rare diseases;
- Encompass the widest possible geographic scope;
- Be centred on a disease or group of diseases rather than therapeutic intervention;
- Have interoperability and harmonisation between registries;
- Consistently use a minimum set of common data elements;
- Have a nationally recognised, integrated process for ethics approval and consent
- Where possible be linked with biobank data and biospecimen access
- Include the possibility of data directly reported by patients as well as healthcare professionals;
- Encourage public-private partnerships to ensure sustainability;
- Involve patients equally with other stakeholders in governance;
- Serve as key instruments for building and empowering patient communities.

The Case for Registries: Benefits to Patients & Treating Doctors

Registries have the potential to provide widespread benefit to researchers, clinicians, governments, industry, patients, patient organisations, and ultimately the community [12]. For registries to be effective, data input must be complete and comprehensive over long periods of time. Only then can registries enable monitoring of disease prevalence, incidence, natural history, interventions and their safety, and functional outcomes effectively. Registries can generate sufficient data on a given RD to make it appropriately visible to researchers, clinicians, governments and industry. Registries can facilitate planning and recruitment for clinical trials, enable post-market drug surveillance and allow the assessment of new interventions once established.

The evidence base for RDs is limited because of their low individual prevalence, and consequent propensity for variations in treatment and outcomes. Registry data can be used to reduce variation in practice, and strengthen the evidence base by generating hypotheses for new interventions and facilitating clinical consensus and practice guidelines. For any given RD, without basic data available through a registry, it is often impossible to take the first critical steps towards improved management or new research.

For RD patients, diagnosis is often difficult or delayed, with misdiagnosis or inappropriate and ineffective treatment commonplace [13]. Even with a correct diagnosis many RDs have no effective treatment, and when treatment is available insufficient data may be cited by funding bodies as a reason not to approve reimbursement. This can make treatment inaccessible to RD patients. Registries enable the accumulation of relevant evidence required to break this cycle.

There are several additional value propositions for establishing an enhanced clinical RD registry model across Australia. Registry data, once collected, could be used for benchmarking in research and clinical practice, for coordination of research groups, to facilitate phenotype/genotype correlations across small patient groups, and to connect researchers and clinicians with international registry initiatives. Registries can also provide transparency for patient advocacy groups to raise the profile and understanding of RDs in the community.

A national RD Registry strategy or alliance could improve consistency across RD registries, standardise governance and consent, and provide efficiencies by consolidating information technology platforms, and linking existing registries thereby improving inter-operability, sustainability and reducing costs. It could support the harmonization of data according to standardized definitions and procedures, enabling multiple clinics to transmit standardized data to a common national registry. Registries could effectively quantify and monitor the burden of RDs in Australia, and identify areas for improvement once accurate data had been collected.

For the approximately 80% of RDs that have a genetic aetiology, genomic analysis can have a major impact on diagnosis and research [14]. Registries can assist genomic research by facilitating the collection of biospecimens and genomic test results. Genomics and bioinformatics have also helped to facilitate international initiatives, such as RD-Connect [15], the Human Variome Project [16], the Global Alliance for Genomic Health [17,18] and Matchmaker Exchange [19], thus encouraging research groups to share vital phenotypic information and build international RD registry communities [15]. To significantly contribute to international efforts, linkages of RD registries with biobanks should be encouraged. International data sharing efforts are proving to be instrumental for advancing our understanding of causative genetic variation in RD.

The Logistics & Challenges of Establishing RD Registries

Registries face common challenges including difficulties in reliable and complete data capture, multi-site research ethics and governance approvals, data harmonization, data custodianship, access to reliable IT platforms, and financial sustainability. Inconsistent approaches in state-based health systems, disparate hospital data collection methods, and variable interpretation of the rules around data sharing are also problematic. These challenges are amplified for RD registries because there are fewer cases of individual RDs, limiting information about RD subtypes, the natural history of each disorder, or the safety of interventions. This leads to inadequacy of available information to effectively support RD advocacy.

Governance and funding difficulties arise for RD registries because RDs are collectively numerous, and even single RDs span multiple clinical specialties, making it difficult to achieve united advocacy and stable funding. Making the case for the sustainable funding of RD registries is challenging given the low frequency of specific RDs and limited treatment options, potentially limiting interest from

government and industry. RVA has an opportunity to unify many disparate disease groups to advocate for action to establish a cohesive national RD registry strategy and alliance.

Despite advances in genomics and bioinformatics and the associated hope these bring, many challenges still remain, including the accurate clinical interpretation of new sequence variants for RDs [20], the variable penetrance of known RD mutations [21] and the fact that many RDs remain undiagnosed even after whole exome or genome analysis. A push for increased international collaboration and consistent data-sharing is a crucial aspect of continued progress, particularly for Australia, given its relatively small population and limited RD case numbers [10]. The scale of ethnic diversity in Australia is also a challenge for genetic research, given ~3% of the Australian population is Indigenous, and 28% are first generation immigrants from over 200 different countries [22].

There is an opportunity to build on existing database frameworks and structures to support RD registries. [23] Systems that simultaneously collect data on many different rare diseases in children such as paediatric surveillance units are currently active in Australia, and have provided incidence estimates and clinical data on over 60 different rare diseases [1,2]. There is potential to expand these systems to support the enrolment of patients in RD registries to collect accurate data across more disease types. There is also an opportunity to collect more patient reported outcome measures (PROMs), especially around quality of life, for RDs where the collection of information from clinical centres may be more challenging. The Australian government has also developed a consumer-managed electronic health record system MyHealthRecord [24] which may also provide future options for data collection or linkage. Implementing a national RD data system requires long-term planning and consensus among many stakeholders with regards to data collection, storage and sharing. International guidelines provide the foundation for this work [15].

RD registries must also strive to capture data that are meaningful to patients and health professionals e.g. quality of life. In some cases, a parochial sense of ownership and a reticence to share for some Australian RD registries, has further restricted progress. To be sustainable, after data collection national RD registries must also be designed to deliver genuine and practical benefit to all stakeholders, including clinicians, researchers, government, health service providers, pharmaceutical companies, patient advocacy groups, and most importantly to RD patients themselves.

Finally, a National RD framework will probably never be achievable without significant government funding to help network and link existing registries, and provide the long-term sustainability required to make registries effective. There has been long-held enthusiasm for a National RD Plan to help address the growing needs of this fragmented and under-served patient community [1,2,4,5]. The new-found enthusiasm for RD registries is an important first step towards a national approach for rare diseases in Australia. The RD research and registry community has aligned itself with RVA to create better national cohesion and a scalable framework to ensure that government funding provided for RD strategies is spent most effectively.

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Supplementary Table 1. Existing Rare Disease Registries hosted and/or operated in Australia. NB This is not an exhaustive list.

<u>Name of registry</u>	<u>URL</u>	<u>State</u>	<u>Hosting Institution</u>
The Australian Cystic Fibrosis Data Registry	www.cysticfibrosis.org.au/data-registry	National	Cystic Fibrosis Australia/The Thoracic Society of Australia and New Zealand
The Australian and New Zealand Fontan Registry	www.fontanregistry.com	VIC	Murdoch Children's Research Institute
Victorian Cerebral Palsy Register	https://www.mcri.edu.au/research/projects/victorian-cerebral-palsy-register	VIC	Murdoch Children's Research Institute
Global Angelman Syndrome Registry	https://angelmanregistry.info/	QLD/WA	Mater Research, Royal Children's Hospital, Murdoch University
Australian Motor Neurone Disease Registry	www.mndaust.asn.au/Discover-our-research/Latest-research/AMNDR.aspx	NSW	MND Research Institute of Australia
The Australian Rett Syndrome study/database	www.interrett.org.au/about-us/australian-rett-syndrome-study/	WA/NSW	Telethon Kids Institute/ Children's Hospital at Westmead, Sydney
Perth Lupus Registry	www.medpharm.uwa.edu.au/Sites/QEIIIMC_Home/Research/Groups/Rheumatology	WA	University of Western Australia
Thrombotic Thrombocytopenic Purpura (TTP)	www.med.monash.edu.au/sphpm/depts-centres-units/registries/ttp.html	VIC	Monash University
Aplastic Anaemia (AA) Registry	www.torc.org.au/aar	VIC	Monash University
Haemoglobinopathy Registry	www.torc.org.au/hbr	VIC	Monash University
Global Myotubular and Centronuclear Myopathy Patient Registry	mtmcmregistry.org/	WA	TREAT-NMD at Newcastle University/ Murdoch University

Australian National Spinal Muscular Atrophy Registry	nmdregistry.com.au/sma/	WA	Department of Health WA (Office of Population Health Genomics) /Murdoch University
Australian National Duchenne Muscular Dystrophy Registry	nmdregistry.com.au/dmd/	WA	Department of Health WA (Office of Population Health Genomics)/ Murdoch University
HHT Alliance (Hereditary Hemorrhagic Telangiectasia)	www.hht.org.au/	VIC	Melbourne Health
Australian Registry: FKRП (Dystroglycanopathies) mutation	www.ann.org.au/dystroglycanopathies-fkrp-mutation/	National	Australian Neuromuscular Network. Registry linked to TREAT-DMD international registry
The Charcot-Marie-Tooth Registry	www.nmd-journal.com/article/S0960-8966(06)00626-2/abstract	NSW/VIC	University of Sydney/Children's Hospital at Westmead/Royal Children's Hospital, Melbourne
Chromosome 18 Registry and Research Society Australia	www.geneticandrarediseasenetwork.org.au/support-groups/view/16/	National	Appears to be linked to an American Registry
Australian Creutzfeldt-Jakob Syndrome Registry	ancjdr.path.unimelb.edu.au/	VIC	The University of Melbourne
The Australasia Epidermolysis Bullosa Registry	https://www.ncbi.nlm.nih.gov/pubmed/20566927	NSW	University of NSW
The Australian Birth Defects Registry (Cleft Palate/lip)	www.cleftpalswa.org.au/userfiles/Birth%20Defects%20report.pdf	WA	The Australian Birth Defects Registry
Registry of Kidney Diseases (ROKD)	www.med.monash.edu.au/sphpm/depts-centres-units/registries/rokd.html	VIC	Monash University
Neonatal Alloimmune Thrombocytopenia (NAIT) Registry	www.torc.org.au/nait	VIC	Monash University
Osteogenesis Imperfecta (OI) Registry	www.oif.org/site/PageServer?pagename=RS_Registry	Int'l/NSW	The University of Sydney
Australian Idiopathic Pulmonary Fibrosis Registry	lungfoundation.com.au/health-professionals/idiopathic-pulmonary-fibrosis-registry/	NSW	Lung Foundation Australia

Australian Bronchiectasis Registry	lungfoundation.com.au/health-professionals/bronchiectasis-registry/	NSW	Lung Foundation Australia
Lymphangioliomyomatosis (LAM) Registry	lamaustralia.org.au/join-us/	NSW	Various

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References

- 1 **Jaffe A, Zurynski Y, Beville L, Elliott E.** 2010. Call for a national plan for rare diseases. *Journal of paediatrics and child health* **46**: 2-4.
- 2 **Zurynski Y, Frith K, Leonard H, Elliott E.** 2008. Rare childhood diseases: how should we respond? *Archives of disease in childhood* **93**: 1071-4.
- 3 2016. Orphanet Report Series: Prevalence and incidence of rare diseases
http://www.orpha.net/orphacom/cahiers/docs/GB/Prevalence_of_rare_diseases_by_alphabetical_list.pdf.
- 4 **Molster C, Youngs L, Hammond E, Dawkins H, et al.** 2012. Key outcomes from stakeholder workshops at a symposium to inform the development of an Australian national plan for rare diseases. *Orphanet journal of rare diseases* **7**: 50.
- 5 **Kirby T.** 2012. Australia makes up for lost time on rare diseases. *Lancet* **379**: 1689-90.
- 6 Rare Voices Australia (RVA) - RARE DISEASE SUMMIT COMMUNIQUE, 2014
<https://www.rarevoices.org.au/page/57/communique>.
- 7 FAIR FOR RARE: A Fair Go for Australians Living with Rare Disease
<http://www.fairforrare.com.au/>.
- 8 **Nony P, Kurbatova P, Bajard A, Malik S, et al.** 2014. A methodological framework for drug development in rare diseases. *Orphanet journal of rare diseases* **9**: 164.
- 9 2012. EURORDIS-NORD-CORD Joint Declaration of 10 Key Principles for Rare Disease Patient Registries
http://download.eurordis.org/documents/pdf/EURORDIS_NORD_CORD_JointDec_Registries_FINAL.pdf.
- 10 **Wilkinson MD, Dumontier M, Aalbersberg IJ, Appleton G, et al.** 2016. The FAIR Guiding Principles for scientific data management and stewardship. *Scientific data* **3**: 160018.
- 11 2014. European Commission, Rare Diseases
http://ec.europa.eu/health/rare_diseases/docs/2014_rare_diseases_informationsheet_en.pdf.
- 12 2011. Rare Disease Task Force (RDTF) Report on Patient Registries in the Field of Rare Diseases (June 2011) - http://www.eucerd.eu/?post_type=document&p=1218.
- 13 **Zurynski Y, Deverell M, Dalkeith T, Johnson S, et al.** 2017. Australian children living with rare diseases: experiences of diagnosis and perceived consequences of diagnostic delays. *Orphanet journal of rare diseases* **12**: 68.
- 14 **Chong JX, Buckingham KJ, Jhangiani SN, Boehm C, et al.** 2015. The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities. *American journal of human genetics* **97**: 199-215.
- 15 **Thompson R, Johnston L, Taruscio D, Monaco L, et al.** 2014. RD-Connect: an integrated platform connecting databases, registries, biobanks and clinical bioinformatics for rare disease research. *Journal of general internal medicine* **29 Suppl 3**: S780-7.
- 16 **Ring HZ, Kwok PY, Cotton RG.** 2006. Human Variome Project: an international collaboration to catalogue human genetic variation. *Pharmacogenomics* **7**: 969-72.
- 17 **Rahimzadeh V, Dyke SO, Knoppers BM.** 2016. An International Framework for Data Sharing: Moving Forward with the Global Alliance for Genomics and Health. *Biopreservation and biobanking* **14**: 256-9.
- 18 **Knoppers BM.** 2014. Framework for responsible sharing of genomic and health-related data. *The HUGO journal* **8**: 3.

- 19 **Philippakis AA, Azzariti DR, Beltran S, Brookes AJ, et al.** 2015. The Matchmaker Exchange: a platform for rare disease gene discovery. *Human mutation* **36**: 915-21.
- 20 **Salgado D, Bellgard MI, Desvignes JP, Beroud C.** 2016. How to Identify Pathogenic Mutations among All Those Variations: Variant Annotation and Filtration in the Genome Sequencing Era. *Human mutation*.
- 21 **Song W, Gardner SA, Hovhannisyan H, Natalizio A, et al.** 2016. Exploring the landscape of pathogenic genetic variation in the ExAC population database: insights of relevance to variant classification. *Genetics in medicine : official journal of the American College of Medical Genetics* **18**: 850-4.
- 22 Australian Bureau of Statistics: 3238.0.55.001 - Estimates of Aboriginal and Torres Strait Islander Australians, June 2011.
<http://www.abs.gov.au/ausstats/abs@.nsf/mf/3238.0.55.001>, 3412.0 - Migration, Australia, 2014-15.
<http://www.abs.gov.au/AUSSTATS/abs@.nsf/Latestproducts/3412.0Main%20Features32014-15>.
- 23 **Bellgard MI, Render L, Radochonski M, Hunter A.** 2014. Second generation registry framework. *Source code for biology and medicine* **9**: 14.
- 24 MyHealthRecord:
<https://myhealthrecord.gov.au/internet/mhr/publishing.nsf/Content/home>.

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(vii) Abstract:

When registries collect accurate clinical data over extended periods of time, they can act as fundamental support structures for patients and their families, and powerful cost-effective instruments to support clinical trials and translational research that improves quality of care, quality of life and survival. Registries are critical for Rare Diseases (RDs) with low prevalence and propensity for variation in treatment and outcomes. Rare Voices Australia (RVA) is leading a call for action to the research and clinical community to prioritise RD data collection and develop an integrated RD Registry strategy for Australia. Financial, operational and governance challenges exist for establishing and maintaining RD registries. As a multidisciplinary team whose interests converge on rare diseases, we highlight the need for the establishment of an Australian RD Registry Alliance. This “umbrella” organisation will: i) bring together existing RD registries across Australia; ii) establish National RD Registry Standards to support inter-operability and cohesion across registries; iii) develop strategies to attract sustainable funding from government and other sources to maximise the utility of existing RD registries and support the development of new RD registries. The most important role for the Alliance would be to use the RD registries for translational research to address current knowledge gaps about RD and to improve the care up to 1.5 million Australians estimated to live with RDs.

Key words:

Rare disease, registries, clinical registries, genetics