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APARDO NEWSLETTER

iConnect

YOUR BI-ANNUAL RD COMMUNITY NEWS, INFORMATION AND EVENTS

NOVEMBER | ISSUE 01 | 2020

On the Board Welcome

Welcome to our inaugural APARDO newsletter and thank you to the amazing response and sharing of fantastic articles from many of our collective members. It is truly uplifting to have insight into the major initiatives, projects and conferences that have occurred across our region. Rare diseases across Asia Pacific region deserve a platform and voice with opportunities to connect for added strength, unity and cohesion and this is only possible with the effort and willingness of our members. One of the great challenges facing our rare communities is COVID-19 and the way that each country responds will

be vital to effective recovery with equity for those with the most vulnerabilities hopefully being at the core of efforts to rebuild and recover. As a vulnerable group, we hope that each Government will ensure that people living with rare diseases are included in planning for a more robust health system.

“Some people focus attention at the core; others are moved to address symptoms. We need both to make change possible.”

~Menachem Kniespeck



**Asia Pacific
Alliance of Rare
Disease Organisations**



Dr. Ritu Jain



Dr. Durhane Wong-Rieger



Dr. Ramaiah Muthyala



Ms. Lisa Foster



Dr. Shuan-Pei Lin



Mr. Tsang Kin Ping



Ms. Eileen Li

CALL FOR SUBMISSION OF ARTICLES

We want to share our stories, hopes, and successes across our region as together we are strong! APARDO is seeking your contributions to its bi-annual newsletter. Kindly submit your organization's planned activities so we may share your events on our website and social media platforms.



Minister of Health Chris Hipkins and myself

Raising Awareness About Rare Diseases

RARE DISEASES — IS SOMETHING SO COMMON REALLY SO RARE?

New Zealand Curekids Professor of Paediatric Genetics at the University of Otago (and Board member of RDNZ) Stephen Robertson has written an article for GP magazine. Circulated to 5,500 GPs, this article talks about the importance of support groups and patient organizations like RDNZ in helping patients to improve their health and wellbeing

https://www.rnzcgp.org.nz/GPPulse/Opinion/Opinion_Rare_diseases.aspx

https://www.rnzcgp.org.nz/GPPulse/Clinical_news/2020/Individually_rare_collectively_common.aspx



Rare Disorders NZ Campaign

By: Lisa Foster



The importance of health and wellbeing has never been so clear in the light of the COVID-19 pandemic. Along with the whole world New Zealand has been focused on protecting, minimising and managing the crisis while balancing health needs with economic impact. Our vision for a 'RESET' economy includes an intention to build back better within our recovery for our health and disability sector. The main question we have is *'Are people with rare diseases included in this rebuild or an afterthought?'*

Medicines New Zealand hold an annual parliamentary dinner to bring together industry partners, charities and politicians so transformative conversations can occur. The focus this year was on: **Medicines Inequity - the problem and potential solutions.**

I was able to meet with our

Minister of Health Chris Hipkins to request a meeting to talk about rare diseases and how we can help improve access to medicines along with other positive possibilities.

Sadly, New Zealand ranks last out of 20 OECD countries for market access to modern medicines and invest only 5% of their total health budget to publicly fund medicines as opposed to 15% for Australia and 12% for the UK.

NZ had only two modern medicines to treat rare diseases funded between 2011-2018 out of 36 launched in the OECD.

'We must not allow inequality to take hold in our recovery'
NZ Finance Minister

Taking Care of Ourselves: Getting Through Together

FOCUS ON WELL BEING

The importance of mental wellbeing during this time of constant uncertainty and change has been a key concern in New Zealand with some great resources and apps provided by our Mental Health Foundation.

One key factor was continuing to reach out to others, as this can really help in people feeling less alone and scared. The value of our collective community in New Zealand, within our regions and throughout the world holding strong together has never been so important.

WINNING WAYS TO WELLBEING



INTRODUCE THESE FIVE SIMPLE STRATEGIES INTO YOUR LIFE AND YOU WILL FEEL THE BENEFITS.

Mental Health Foundation of New Zealand
www.mentalhealth.org.nz



New Zealand Campaign and Petition

- Rare Disorders New Zealand are running **Fair for Rare NZ campaign** and parliamentary petition to gain acknowledgement and fairness for people living with rare diseases.
- This campaign is asking the Government for a National Framework and Action Plan for comprehensive patient-centred care across sectors with a focus on seven priority areas. These include access to early diagnosis, coordinated care pathways, access to disability services and social supports, access to medicines, research on rare diseases and a rare disease registry to capture data plus workforce development.
- RDNZ, alongside mum and advocate Sue Haldane, launched a parliamentary petition in Feb 2020 with a push for our 140+ NZ rare disease support groups to visit their local Members of Parliament (MP) to ensure awareness and inclusion in the wellbeing budget here. We are feeling encouraged with a new Labour Government elected and their mission to listen, act and lead the country to equity for all New Zealanders and to build back better.

It is vital that New Zealand Government include people with rare diseases and we have 4170 signatures so far and many more to show it matters so please sign and share with your networks (the petition can be signed by people living outside of New Zealand).

https://www.parliament.nz/en/pb/petitions/document/PET_94998/petition-of-sue-haldane-for-rare-disorders-nz-develop



10 Years in the Making: Australia's National Strategic Action Plan for Rare Disease

By: Nicole Millis



For over a decade, Australia's rare disease sector has advocated for a national policy framework to respond effectively to rare diseases. With bipartisan support, the National Strategic Action Plan for Rare Diseases (the Action Plan) was commissioned by the Australian Federal Government in November 2018. Its collaborative development was led by Rare Voices Australia (RVA), Australia's peak body for people living with a rare disease. Informed by extensive stakeholder consultation, the Action Plan addresses the countless commonalities among over 7,000 different rare diseases.

The Action Plan was launched in February 2020 by Australia's Minister for Health. The launch was accompanied by an initial funding announcement from the Australian Government of \$3.3 million for implementation activities. It is anticipated that the Action Plan will also guide future Government investment into rare diseases.

The Action Plan is built on three key principles: person-centred, equity of access and sustainable systems and workforce. It is comprised of three core Pillars, with each Pillar outlining priorities, actions and implementation recommendations. The Pillars are: Awareness and Education; Care and Support; and Research and Data.

Covering a wide scope, the Action Plan aligns with and expands on RVA's Call for a National Rare Disease Framework:



crucially, the Asia-Pacific Economic Cooperation (APEC) Action Plan on Rare Diseases.

Throughout 2020, the Action Plan has provided a sound case study as to how a comprehensive policy framework can reduce uncertainty through policy while remaining flexible enough to respond to changing policy contexts. For the estimated two million Australians living with a rare disease, triggers such as the current COVID-19 pandemic only exacerbate the high levels of uncertainty faced. As such, RVA has used the Action Plan to guide its advocacy around the pandemic.

Through national leadership and coordination, the Action Plan can drive and future-proof much needed reform.

6 Strategic Priorities, published by RVA in June 2017, and

When, 19 years ago, my son was diagnosed with a rare disease, I was left with many questions and few answers. Why did this happen? How do I fix it? What do I do? I know first-hand that effective policy can transform the lives of people living with a rare disease. The Action Plan is already providing opportunities for systemic change with the House of Representatives Standing Committee on Health, Aged Care and Sport announcing a Parliamentary Inquiry into approval processes for new drugs and novel medical technologies following a referral from the Minister for Health. The inquiry will include a particular focus on access to the treatment of rare diseases.

With effective policy, we can reduce the disempowering uncertainty of rare diseases.

New Hope for Those with Rare and Undiagnosed Diseases

By: May Ho

It is a common fact that rare diseases are tough to diagnose. A patient may have to undergo a years-long and tortuous journey with countless visits to different specialists, numerous tests and feelings of despair before getting the correct diagnosis and right treatment. Delays in diagnosis can lead to inappropriate management, or



which was initiated in 1990 and led by an international team of researchers, the human genome sequence was completed in 2003. Genome is the complete set of genetic material (i.e. DNA) passed down from parent to child. Information about the human genome sequence and its variants can be applied in order to develop highly effective diagnostic tools, to better understand the health needs of people based on their individual genetic make-ups, and to design new and highly effective treatments for diseases. As the majority of rare diseases are caused by changes in DNA that affect proper gene function, genetic testing can be an effective method to diagnose rare diseases.

Ironically, a fast-paced city like Hong Kong is lagging behind on the implementation of genomic medicine when compared with the US, Western Europe and some Asian countries like Japan, South Korea, Singapore, Thailand and Sri Lanka.

It was only after more than a decade's lobbying and advocacy that the Steering Committee on Genomic Medicine was set up to lead the study on strategies for developing genomic medicine in Hong Kong. About two and a half years later, in May 2020, the Government eventually announced the

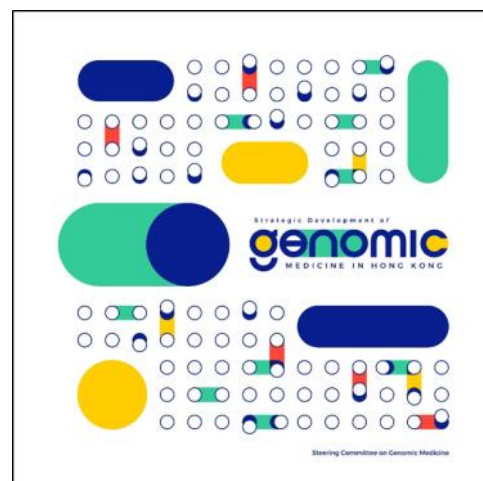
Strategic Development of Genome Medicine in Hong Kong, in which HK\$1.2 billion (around US\$154 million) has been set aside to execute the **Hong Kong Genome Project** (HKGP) to set up a genome sequencing database to help detect undiagnosed genetic disorders and cancers.

The HKGP aims to perform 20,000 cases (or 40,000 to 50,000 whole genome sequencing) in two phases for a period of six years. The pilot phase (2,000 cases or about 5,000 genomes) will cover patients with undiagnosed disorders, and cancers with clinical clues linked to possible hereditary or genetic components. The latest plan is to start recruiting patients for genome sequencing in mid-2021.



even disease progression. But with limited information and scientific knowledge about rare diseases, early and accurate diagnosis seems to be an impossible dream for most rare disease patients.

Thanks to the Human Genome Project



New Hope for those with Rare ...

However, policy announcement is only the first step. There are multi-faceted issues to be addressed – (a) standardised clinical service provision; (b) more efficient laboratory services and translation of new technology to clinical use; (c) more healthcare professionals specialised in genomic medicine and enhancement of genomic literacy among general healthcare professionals; and (d) ethical, legal and social implications.

Anticipating that it will take many more years to bring the HKGP to fruition, Rare Disease Hong Kong (RDHK), as a charitable organisation representing the interests of rare disease patients and their caregivers, took the initiative to work with an institution and a renowned clinical geneticist in the private sector to start up *The Rare Disease Genome Project* (RDGP) in late 2019. The project aims to establish a database of health and genomic information of

people affected by rare diseases. This database will facilitate studies on the causes and consequences of rare diseases and will support the development of new tests and treatments. To date, about 50 families affected by facioscapulohumeral muscular dystrophy, retinitis pigmentosa, spinal cerebellar ataxia, spinal muscular atrophy, Charcot-Marie-Tooth disease, or undiagnosed disease have been, or will be, participating in the project. It is envisaged the RDGP will have collaboration and cohesion with the Government's HKGP when it is implemented.

Apart from enhancing the regulation on the storage and use of genetic data, the Government should also steer and coordinate efforts across different sectors to boost the

development of genomic medicine in Hong Kong.

It is believed that the HKGP can be sped up by promoting tripartite collaboration among the Government, the business sector and the community. Besides, clear and simple language should be used for effective communication with the public.

In summary, advances in genomics and genetics offer new hope for patients with rare diseases and with hope HK Government is able to boost the development of genomic medicine in partnership with the private and third sectors for the long-term benefit of the community.



¹ Genetic testing is a type of medical test that identifies changes in chromosomes, genes, or proteins. The results of a genetic test can confirm or rule out a suspected genetic condition or help determine a person's chance of developing or passing on a genetic disorder. More than 1,000 genetic tests are currently in use, and more are being developed. (source: <https://ghr.nlm.nih.gov/>)

² Details of the *Strategic Development of Genome Medicine in Hong Kong* can be found in the following website: https://www.fhb.gov.hk/en/press_and_publications/otherinfo/200300_genomic/index.html

³ Rare Disease Hong Kong (RDHK), formerly known as the Hong Kong Alliance for Rare Diseases, is the first patients' group in Hong Kong comprising cross-rare-disease patients and their families with the support of experts and academics in the field. RDHK aims to improve rare disease policies and services, promote public education on rare diseases, and strengthen the community's support for patients in order to ensure equal respect and protection for patients in terms of fundamental rights such as healthcare, social support, education, and daily needs.

Strategic Planning

By: Monica Ferrie



The GSNV Strategic Plan is due for renewal at the completion of 2020. Our Plan will set the direction and priorities for the next three years and guide the development of each year's annual business and work plans. We are committed to creating sustainable change and will focus our plan development on the Sustainable Development Goals, the Victorian Charter of Human Rights and Responsibilities, Australian Charter of Healthcare Rights, and other key policy documents.

In the development of our strategic plan we engage with key stakeholders across these themes.

to share their personal journeys and thoughts of this very unusual time.

Our research project, established in collaboration with Australian Genomics provides people with the opportunity to collect and share their thoughts through a variety of journaling methods.

Initial analysis shows there are some themes in the journals around day to day life, watching the impact of COVID19 change, isolation, coping and resilience. We will be using the theory around resilience to analyse the journal submissions in the coming months. We will define resilience as the dynamic ability to maintain/restore relatively stable functioning, that will ebb and flow, when confronted with stressful life events and adversity. Resilience exists on a continuum and we will all have varying coping strategies that we will need to deploy at different times.

The Genetic, Undiagnosed and Rare Disease Communities are represented on the National COVID-19 Clinical Evidence Taskforce – Consumer Panel through Monica Ferrie, Chief Executive Officer of the Genetic Support Network of Victoria.

The Australian National COVID-19 Clinical Guidelines Taskforces — Consumer Panel

There is a National Australian COVID-19 Clinical Evidence Taskforce with representation on consumer panel by Monica Ferrie, Chief Executive Officer of the Genetic Support Network of Victoria. This Taskforce brings together the peak health professional bodies across Australia and undertakes continuous evidence surveillance to

C19 Journal Research Project

The C19 Journals Project commenced in July 2020 encouraging our community

Strategic Planning ...

identify and rapidly synthesise emerging research in order to provide national, evidence-based guidelines for the clinical care of people with COVID-19. These are 'living' guidelines, updated with new research in near real-time in order to give reliable, up-to-date minute advice to clinicians providing frontline care in this unprecedented global health crisis.

Mackenzie's Mission

Mackenzie's Mission is Australia's largest reproductive carrier screening project ever undertaken, with a target of 10,000 couples and testing for more than 750 rare conditions. The Engagement Reference Group chaired by Monica Ferrie of the GSNV meets regularly for updates and governance.

While COVID-19 has had some impact, it so exciting that full-scale recruitment

is happening in four states and territories (Victoria, Western Australia, New South Wales and ACT) ahead of national expansion in 2021. Health Professionals including General Practitioner's participating in the project are receiving education and resources around conversations with couples about carrier screening and carrier screening in general.

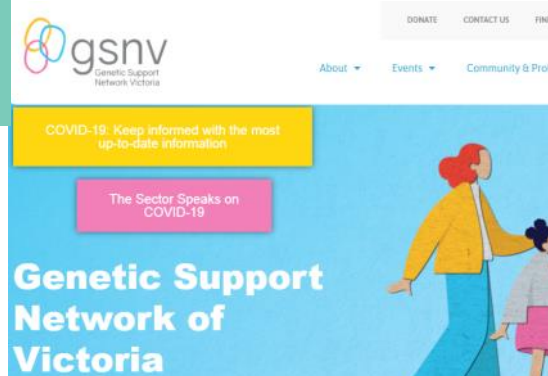
Couples can only access this study through a health professional, they cannot self-refer to the Mackenzie's Mission project. There is more information available at <https://www.mackenziesmission.org.au/>

RDNOW - A Rare Disease Project

The Genetic Support Network of Victoria was delighted to host the launch of the RDNOW Project at our 2020 Rare Disease Day.

Rare Diseases Now (RDNOW) is an exciting new initiative to deliver

genomic diagnoses and precise, personalised care to Royal Children's Hospital (RCH) families. Drawing on the research and clinical expertise at the Murdoch Children's Research Institute and Victorian Clinical Genetics Services, RDNOW will engage with non-genetics specialists within RCH to establish a pathway for undiagnosed children to have the best chance of receiving a diagnosis and to access the latest clinical trials and treatments. RDNOW will provide opportunities for families to participate in studies that will enhance our clinical and scientific knowledge of very rare conditions and increase our ability to provide great care and support to children and their families.



Healthy China Leave No One Behind

By: Illness Challenge Foundation

"Healthy China, Leave No One Behind", which is the theme of 2020 Rare Disease Symposium on Collaboration and Communication.



8772 Band (all band members are rare disease patients and their original songs are composed/written by rare disease patients and families) performed the Opening show.

Every year, the Illness Challenge Foundation (ICF) hosts the Rare Disease Symposium on Collaboration and Communication in China. Under the guidance of the China Alliance of Rare Diseases (CARD), the symposium is developed into a patient-focused joint force aiming at promoting the engagements between patient organizations and other stakeholders.

On August 22-23, 2020 Rare Disease Symposium on Collaboration and Communication was successfully held online, which includes 11 forums, an opening ceremony, and a global charity show within 2 days. There were 86 guest speakers from medical/research institutes, academia, industry, government departments, patient organizations, and international alliances who shared their insights on over 73 topics regarding orphan drug research and development, access to treatment, multi-party co-payment initiatives of healthcare expense for rare disease patients, sustainable development and global collaboration of patient organizations, and specific topics on genetic inheritance, fertility, ethics, diagnosis, treatment, and rehabilitation of some rare diseases. 8 platforms including national broadcast platforms broadcasted the symposium, and more than 100 rare disease patient organizations participated, with nearly 2 million viewers.



Mr. ZHANG Wenbao, Official of National Health Commission delivered the Opening remarks.

In the opening ceremony, government officials and representatives shared their perspectives on rare disease issues in China. The launch of Medical Aids for Rare Diseases Multi-Party Co-Payment Initiative, and roundtable discussions on Innovative Payment Model for Rare Disease Medical Insurance and Challenges and Opportunities for the Implementation of Rare Disease Policies occurred.

"Strengthening the prevention and treatment on rare diseases is a great significance for implementing the Healthy China strategy, winning the



Ms. ZHANG Shuyang, Vice Director and Secretary of the Party Committee of Peking Union Medical College Hospital, Secretary-General of China Alliance of Rare Diseases delivered a keynote speech.

battle against poverty, and decisively building a well-off society in an all-round way. The prevention and treatment on rare diseases is an important beginning to promote the in-depth development of medical reform.” Mr. ZHANG Wenbao, Official of National Health Commission stated, he continued to explain “We will continue to strengthen communication and coordination with various departments, create a good policy environment for the prevention and treatment of rare diseases, and promote the prevention and management of rare diseases in China. We also hope that all social organizations, corporations and other related aspects, actively participate in the prevention and treatment on rare diseases to promote the rapid development of it.”



Ms. WANG Yoyo, Founder and Secretary-General of Illness Challenge Foundation, Founder of China-Dolls Center for Rare Disorders, Patient with Osteogenesis Imperfecta delivered a keynote speech.

Round Table Challenges and Opportunities for the Implementation of Rare Disease Policies



Mr. WU Xiaobin, CEO of BeiGene delivered a keynote speech.

1. Medical insurance and payment model

In the past five years, China's rare disease policies have made significant improvements, especially the Catalogue of First Batch of Rare Disease released in 2018, defined 121 rare diseases for the first time in China. Several provinces and cities have issued policies to support a relatively high proportion of medical expense reimbursement for the 121 rare diseases. Policy influencers on both local and national level discussed the current situation of this



Mr. SHI Luwen, Director of International Research Center for Medical Administration Peking University delivered a keynote speech.

social security system for rare disease patients, and both the challenges and opportunities

2. Patient-focused drug development and access

Representatives across sectors shared domestic and global experience, about the current status of drug R&D for rare diseases in China, and the importance of promoting patient participation not only in the drug development process, but also from the very beginning of the process. The value of patient input when prioritizing research and drug discovery, post clinical trials and when forming legislation for access to treatments was highlighted.



Round Table: Innovative Payment Model for Rare Disease Medical Insurance

3. Sustainable development and global collaboration of patient organizations

Fundraising and Sustainability of Rare Disease Patient Organizations was discussed along with other key topics. The Illness Challenge Foundation was honored to invited Dr. Ritu Jain, President of APARDO, Dr. Durhane Wong-Rieger, Vice President of APARDO and Mr. Kin Ping Tsang, Treasurer of APARDO to share their

insights about patient-centeredness and global collaboration.. Dr. Brian Chung and Ms. Claudia Chung from Hong Kong University, along with Dr. Ritu Jain shared data and findings on an earlier survey on The Impact of COVID-19 Pandemic on Rare Disease Patient Organizations and Patients across the Asia Pacific Region, which included over 30 Chinese rare disease patient organizations.



Round Table: Innovative Payment Model for Rare Disease Medical Insurance

4. Particular topics on genetic inheritance, fertility, ethics, diagnosis, treatment, and rehabilitation of some rare diseases.

There were 5 forums focused on specific topics with Fabry China expressing concerns over access to treatments due to expense and there were in China patient discussions on treatment of LSDs, living status of patients with LSDs, potential social security model, and the development of patient organizations.

The event concluded with a Charity show called “RARE LIVE” showing the talented rare lives around the global. Rare disease patients and families around the world brought all various forms of performances, such as singing, dancing, poem reading, musical instrument playing, sand painting, and shadow play. This was the voice of the rare disease community.

The two-day event was a complete success.



Round Table: Challenges and Opportunities for the Implementation of Rare Disease Policies

Upcoming Event!

A promotional banner for the APARDO Conference 2020. The banner features a white background with a colorful geometric pattern of interconnected lines and dots in blue, orange, and green. At the top left, the APARDO logo is displayed next to the text "Asia Pacific Alliance of Rare Disease Organisations". To the right, the RARE DISEASES INTERNATIONAL logo is shown. A yellow rectangular box with the text "SAVE THE DATE" is positioned above the main title. The main title, "APARDO Conference 2020 Transforming Rare Diseases Across the Asia Pacific", is centered in a large, bold, blue font. Below the title, the dates "Wednesday, 9th & Thursday, 10th December 2020 (Virtual)" are written in a bold, orange font. Underneath the dates, the text "Time, invitation, agenda and zoom link forthcoming" is displayed in a smaller, black font. At the bottom left, the text "OUR SPONSORS" is followed by a row of logos for Takeda, Roche, AMGEN, NOVARTIS, Biogen, GILEAD, and Pfizer.

Asia Pacific Alliance of Rare Disease Organisations

RARE DISEASES INTERNATIONAL

SAVE THE DATE

APARDO Conference 2020
Transforming Rare Diseases
Across the Asia Pacific

Wednesday, 9th & Thursday, 10th December 2020 (Virtual)

Time, invitation, agenda and zoom link forthcoming

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