Asia Pacific
Alliance of Rare
Disease Organisations

APARDO Annual Conference 2023

24 – 26 November 2023
Kuala Lumpur, Malaysia

[Post-Event Report]

By
Asia Pacific Alliance of Rare Disease Organisations Ltd. (APARDO)

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Alliance of Rare
Disease Organisations

The APARDO conference was delivered in partnership with the Malaysian Rare Disorders Society (MRDS)

NUMBER OF COUNTRIES REPRESENTED	16

NUMBER OF PARTICIPANTS PER COUNTRY	
Australia	3
Canada	1
China	5
France	2
Hong Kong	3
India	1
Indonesia	4
Malaysia	116
Singapore	9
Switzerland	1
Thailand	4
USA	1
Vietnam	2
Philippines	2
Korea	3
Nepal	2
Total	**159**

NUMBER OF PARTICIPANTS (PHYSICAL / ONLINE)	
Physical	138
Online	21

Date: November 25, 2023 (Saturday)
Time: 9AM to 4:30PM
Venue: Pullman Kuala Lumpur City Centre, Malaysia

Content	Speaker & Moderator	Summary
Welcome Remarks	Dr. Durhane Wong-Rieger (APARDO)	
Keynote Address	YBhg. Dato' Dr Azman bin Yacob (Director of Medical Development, Ministry of Health Malaysia)	Malaysia demonstrates an unwavering commitment to addressing rare diseases (RD) through initiatives like the Madani medical scheme, showcasing an efficient and patient-centred care approach for a new era of excellence. Despite budget

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		constraints, the country persistently seeks additional resources for research and development (R&D). Recognising the multifaceted challenges faced by those living with rare diseases, the commitment extends beyond healthcare to include the formation of a national committee and the establishment of a trust fund in 2022 to support R&D, treatment, and financial assistance for affected individuals. The acknowledgement of support from activists and non-governmental organisations underscores the collaborative effort. Malaysia's Ministry of Health (MOH) is pivotal in achieving universal health coverage, allocating a substantial annual budget for R&D, treatment, and diagnosis. The emphasis is on ensuring inclusivity and responsiveness within the healthcare system, with the Malaysia Trust Fund providing financial aid and access to quality healthcare. The commitment to leaving no one behind is reflected in ongoing efforts, including support groups and the annual NGO-run initiative for social and emotional assistance to people with rare diseases (PWRD). The call for continued changes, characterised by care, professionalism, and teamwork, emphasises universal health coverage as an essential element in Malaysia's healthcare system.
Opening	Ms. Nadiah Hanim Abdul Latif (MRDS)	**Topic: Empowered Rare Disease Patient Advocacy** The realisation that RD is not a straightforward path but rather a team effort, similar to raising a child in a caring community, emphasizes the journey's complexity. It emphasizes the necessity of hard work, debunking the notion that labels should define individuals, as encapsulated by the powerful message, "Your labels do not define you". Acknowledging the challenges, the narrative recognizes the loss of family members and the poignant reminder from APARDO 2023, run entirely by dedicated volunteers, urging us not to let the sacrifices be in vain. This initiative, spanning 13 countries and the 35 RD groups, hold significance in shaping the trajectory of RD in Malaysia and the broader regional context.
Session 1	Moderators: Ms. Monica Ferrie (Genetic Support Network of Victoria) Ms. Nadiah Hanim Abdul Latif (MRDS) Dr. Durhane Wong-Rieger (APARDO & Rare Disease International) Panelists: Dr. Carmencita Padilla (Philippine Society for Orphan Disorders) Dr. Kittiwan Rojnueangit	**Topic: An Ideal-but-Realistic Shared Vision of Asia-Pacific Rare Disease Network — 7 Years from Today** The discourse on rare diseases (RD) revolves around the fundamental notion that each individual is unique, necessitating precision medicine tailored to specific conditions. Anne Sophie emphasizes the urgency of investing in research and development for RD, asserting that it should be viewed as a strategic investment rather than charity. Professor Thong reflects on the 30-year history of RD in Malaysia, advocating for collaborative efforts to break down silos and achieve more impactful outcomes. Faris from Roche underscores the economic burden of RD and calls for a diversified approach beyond government reliance, stressing the need for a

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	Dr. Thong Meow Keong (Pediatrics and Clinical Genetics, University of Malaya Medical Center, Kuala Lumpur, Malaysia) Dr. Rachel Yang (China Alliance for Rare Diseases) Ms. Anne Sophie Chalandon (Sanofi) Mrs. Alexandra Heumber (RDI) Dr. Fariz Abdul Rani (Roche)	measurable return on investment. The conversation extends to regional perspectives, with insights from Thailand on the importance of investing in early diagnosis and treatment to prevent long-term costs. • The discussion also delves into the importance of shared-care pathways, regional collaboration, and the role of social media in connecting stakeholders. Prof. Thong suggests harnessing social media to educate and unite people, emphasizing the significance of psychosocial support and cultural understanding in RD treatment. The need for a global framework for RD is highlighted, with Monica stressing the importance of political will and collaboration across borders. Alex emphasizes the necessity of a unified global Asia network and the implementation of Universal Health Coverage (UHC) commitments. As the conversation progresses, the focus shifts to practical steps for collaboration, including patient support groups, clinician involvement, and government support. The call for a unified voice across the region is echoed, emphasizing a common narrative and a commitment to not leaving anyone behind. The concept of a "village" emerges, symbolizing a collaborative approach where each country plays an equal role, with metrics to measure progress and a shared message of hope and support. The overarching theme is the need for a comprehensive, interconnected strategy to address the challenges posed by rare diseases on a global scale.
Session 2	Moderators: Ms. Nadiah Hanim Abdul Latif (MRDS) Mr. Sivasangaran Kumaran Dr. Carmencita Padilla (MAHPS Professor and Chancellor, University of the Philippines Manila; Founding Chairman, Philippine Society for Orphan Disorders) Ms. Jasmine Goh Chew Yin (KK Women's and Children's Hospital Singapore) Ms. Sook Yee Yoon (Genetics Counselling Society Malaysia)	**Topic: Collaborative Approach to Shorten Diagnostic Odyssey Applying "Lessons Learned" to "Challenges" Along Diagnostic Journey** The collaborative approaches discussed by the panel highlight the importance of shared knowledge and support in the rare disease (RD) community. Siva shares insights from a patient exchange trip to Taiwan, emphasizing the value of patients exchanging information, particularly in the context of Pompe disease. Dr. Carmencita advocates for a national newborn screening program in the Philippines. showcases the success of their strategy, including the Newborn Screening Act of 2016, continuity clinics, and centers for human genetics services. Jasmine from Singapore sheds light on the services provided for genetic counseling and testing, with funding from the Temasik Foundation and collaboration with the Rare Disease Singapore Society. Mental health support for patients and parents is also a key focus, with informational leaflets accessible through QR codes. Yoon Sook Yee emphasizes the importance of supporting families through genetic, disease, and psychosocial assistance, highlighting collaborative efforts across Malaysia, Singapore, Indonesia, and Australia.

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		The challenges discussed include the need to centralize resources, designate leaders or representatives from each country, avoid burnout through a supportive network, and share both successes and disappointments. The panel underscores the necessity of persistence in current practices, resourcefulness in networking, and adaptability to the distinct journeys of newborn and adult patients. The collaboration extends to engaging with governmental and insurance entities, as seen in Singapore's efforts with adult patients funded by the government and webinars involving genetic counselors. In summary, the key takeaways include the identification of champions, collaboration through unions, comprehensive training for allied health professionals, celebration of every success, persistence as a guiding principle, and the establishment of registries and legislation, as demonstrated in the experiences of the Philippines and Taiwan. The overarching message is clear: united efforts and a holistic approach are essential to effectively address the challenges faced by the rare disease community. Strategy 1: Newborn Screening Act (2016) (RA 10747), Policy Development Strategy 2: Newborn Screening continuity Clinics Strategy 3: Centers for Human Genetics Services
Session 3	Moderator Ms. Monica Ferrie (GSNV) Mr. Will Greene (Roche) Ms. Tara Pensa (Novartis) Dr. Alison Archibald (Victorian Clinical Genetics Services) Dr. Claudia CY Chung (Hong Kong Genome Institute)	**Topic: What are the tools and technologies that improve pathways to diagnosis?** The discussions among Will Greene of Roche, Tara Pensa from Novartis Gene Therapy, Dr. Allison Archibald from the Victorian Clinical Genetics Services (VCGS), and Dr. Claudia from the Hong Kong Genome Institute provide a comprehensive overview of various initiatives and advancements in the field of rare diseases (RD). Will Greene emphasizes the importance of Newborn Screening (NBS) and its potential to lead to early intervention and treatment. He shares insights from Taiwan, the first country to implement NBS for diseases like DMD and Pompe, advocating for case study development in India, South Korea, and Vietnam to optimize NBS systems. Tara Pensa highlights the significance of advocating for NBS and introduces the RESTORE registry for Spinal Muscular Atrophy (SMA), emphasizing that good decisions are made in conversation, not isolation. Dr. Allison Archibald details the reproductive genetic carrier screening efforts in Australia, promoting choice and autonomy in family planning. The Mackenzie Mission, screening for 1300 genes associated with 750 diseases, stands out as a comprehensive and accessible initiative. Dr. Claudia from the Hong Kong Genome Institute sheds light on Whole Exome Sequencing (WES) and Whole Genome

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		Sequencing (WGS) as powerful tools for improving RD diagnosis. She underscores the impact of these technologies in shortening the time of diagnosis and reducing healthcare costs, advocating for collaborative efforts and the use of AI and big data to enhance regional diagnostic capabilities. The recap underscores the need for strong evidence-based projects, global collaboration in training and education, and a patient-centered approach in shaping RD policies. The question raised pertains to the role of AI and big data in the diagnostic journey, addressing ethical dilemmas, religious perspectives, and reproductive choices in the context of carrier reproductive screening. In essence, the collaborative efforts and advancements presented in the discussions highlight the multifaceted approaches needed to address the challenges posed by rare diseases, from early screening and diagnosis to reproductive choices and the integration of advanced technologies like AI and big data.
Session 4	Moderator: Dr. Durhane Wong-Rieger (APARDO & RDI) Ms. Nadiah Hanim Abdul Latif (MRDS) Health Professional Leads Patient Support Group Leads	**Topic: Patient Journey: Pathways to Diagnosis and Beyond (Workshop)** Objective: The Rare-Journey Roadmap aims to comprehensively map the challenges faced at key stages of life, namely newborn-childhood, teenage-adolescence, adulthood, and senior years. Additionally, the report seeks to identify national and regional opportunities to address these challenges effectively. 1) Spinal Muscular Atrophy (SMA) **Teenagers to Early Adulthood** Challenges: A) Physical Challenges and Diagnosis: Late screening and diagnosis. Physical issues such as weak bones/muscles and declining mobility. B) Psychosocial Challenges: Impact on quality of life. Emotional challenges and difficulty in education and employment. C) Continuous Care: Lack of an integrated care continuum Increased caregiver burden. D) Policy:

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		Government priorities are lacking. Economic challenges in funding Rare diseases are seen as the Health Ministry's issue alone. Solutions: • Physical therapy and wheelchair provisions. • Integrated support, counseling, and rehabilitation centers. • Training specialists for teenage SMA patients. • Promoting social enterprises by SMA individuals • Collaboration at the APAC level for patient registration • Innovative funding is needed for treatment access. 2) Phenylketonuria (PKU) **Newborn-Childhood** Challenges: • Health literacy of parents. • Access to care and treatment. • Managing dietary requirements and educating schools • Lack of resources for doctors and patients. • Understanding the condition and genetics • Achieving early diagnosis (NBS in most countries) • Reproductive implications for more children • Cost for treatment Opportunity: • Government support to enhance the quality of life. Solutions: • Advocacy for better access to genetic counseling • Early diagnosis and treatment, especially through NBS. • Multidisciplinary support for long-term care. • Addressing social impacts and reproductive implications • Education resources for doctors and patients • Planning for long term care **Adulthood - Older adults** Challenges: 1) Social • Family planning • Employment • Social life with family and friends • Stigma • Mental health: depressive disorder, intellectual development and emotional management 2) Economic • Higher burden

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		• Special foods (formula drink and supplementary food) • Regular blood test- doctor and dietician appointment 3) Physical Health • Continuous care and treatment • Chronic disease management Treatment: • FDA approved drug saproprotein - may not work for everyone with PKU • Enzyme therapy (Palynziq) Solution: • PGT • Newborn screening • Carrier test • Patient stories sharing • Psychological support workshop organized by patient organization 3) Prader-Willi Syndrome (PWS) **Newborn-Childhood** Challenges: • Early diagnosis may not be straightforward • Limited treatment • The emotional impact of worrying about baby's development • Access to expert assessment (geneticist) and to genetic testing • Diagnosis odyssey - obesity and short stature • Hyperphagia - Bariatric surgery, Ozempic and Metformin Solutions: • Teach primary physicians when to refer • Network of specialist • Educating school and childcare about dietary requirements • The government helps by expanding specialised care to other diseases. **Teenagers - Early Adulthood** Challenges: • Transition of care from child to adult • End of life conversations • Children are more involved with their own treatment pathways • Ambulant vs non ambulant • Peer support activities

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		• Intergenerational gap between seniors • Understanding the differences • Thinking about what's in life - partner, work, lifestyles, support • Body image support • Emotional , physical, mental health, and social support • No treatment • Family planning issues Opportunities: • One center for multidisciplinary care for the duration • Medicine is available in a country or put one in place • Sibling support for them to let go • To liaise with pharmca for PROS if new treatment is available • Private and public co-pay **Adulthood - older adults** Challenges: • Family planning • Sterility • Employment • Transitional period from PWS home, dietician, nurse and psychologist • Adaptation to social and cultural contexts 4) Cystic Fibrosis and Hemophilia **Newborn - Childhood** Challenges: • Symptomatic diagnosis - more accessible for hemophilia and CF, need for a NBS program • Appropriate discussion with parents is needed • Access to treatment and choice to treatment • Registration, Funding and legislation for RDD • Parents are the advocates • School adaptation and education • Insurance • Minimum data set • Preschool - kinder garden interaction with other parents, kids, professional - awareness and adaptation Opportunities: • RD National plan - policy and guidance • Expert centers • Multidisciplinary care • Sibling • consultation and family support

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		• Parents awareness and education • Support around the family • Treatment option • Home adaptation • Parents groups of exchanges • Registries Data PRO RWED **Teenagers to Early Adulthood** Opportunities • Children are more involved with their own treatment pathways **Adulthood - older adults** Challenges: • Diagnosis odyssey - comorbidities despite treatment challenges remain • Public awareness and education • Research collaboration pharmaceutical company • prioritized rare disease registry and RD framework policy • Mental health - anxiety, depression and inheritance • Frequent hospitalization, burden to caregiver Opportunities: • Genetic and reproductive counseling • Patient, financial and spiritual support • Working with medical treatment to find the source of treatment • Multidisciplinary clinic enable the coordination of therapy 5) Mayer-Rokitansky-Kuster-Hauser (MRKH) Syndrome & Amyloidosis **Teenagers - Early Adulthood** Challenges: • Monthly pain • Hearing challenge • Single kidney Treatment: • Bone breech implant • Manage lifestyle for the heart • Manage lifestyle for kidney • Hormone medication Opportunities: • To increase training for physician

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		• awareness among doctors and patients • Psycho-social support from friend and family • International peers network • Normalize the topic to create awareness • Train the young specialist to use current terms while consultation • Encourage the patient to not hide • Social media empowers **Adulthood - Older Adults** Challenges: • How disease awareness • Lack of confidence in relationships • Lack of confidence and intimacy • People assume everything is OK • Friendships with other girls • Anticipating other health complications • Stigma varies across countries • Pressure created by family relationships • Reproductive issues • Access to diagnosis • Cultural taboo cannot even be discussed • Late diagnosis, misdiagnosis or underdiagnosis • Not common in Asian • There are no specific lab test in the region • Self fund testing • Disease couse debilitating • Time to diagnose • Feelings of isolation • Cost of treatment is challenging Solutions: • Public awareness • R & D investment from government for long term treatment • Start with patients: It's not your fault • Training for health professionals • Counseling patients and families • Building data through natural history • Registries • Precision Medicine Overall Recommendations: • Emphasize early diagnosis and treatment. • Promote multidisciplinary support and integrated care. • Advocate for policy changes and increased government investment in rare diseases. • Enhance health literacy among parents and healthcare professionals.

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		• Establish innovative funding mechanisms for treatment access. • Foster collaboration at the regional level for patient registration and support.
Session 5	Moderator: Dr. Durhane Wong-Rieger (APARDO & RDI) Ms. Monica Ferrie (GSNV) Presentation: Mrs. Alexandra Heumber (RDI) Panelists: Mrs. Alexandra Heumber (RDI) Dr. Klara Yuliarti Dr. Ngu Lock Hock (Clinical Genetics Consultant Kuala Lumpur Hospital) Ms. Doreen Tan (Sanofi) Ms. Nadiah Hanim Abdul Latif (MRDS)	Feedback and Large Group Discussion Speed: How can AP Network shorten diagnostic journey with lessons learned? Scale: How can AP Network facilitate spread of shared learnings to other disease areas and countries where not yet available? Sustainability: How can AP Network address physical, knowledge, and psychological barriers among HCPs and patients; how can AP Network improve government support and pharmaceutical investment? **Rare Disease International (RDI): Building a Collaborative Ecosystem for Rare Diseases** The presentation from Rare Disease International set the stage for a strategic roadmap, with a primary focus on shaping the organization into a sustainable and agile entity. The outlined priorities for 2024 demonstrated a clear commitment to enhancing member capacities, influencing policies to foster greater equity among individuals living with rare diseases (PLWRD), and actively participating in global initiatives. A critical aspect of this strategic vision lies in the cultivation of a collaborative ecosystem. Participants underscored the significance of networking opportunities across different levels—globally, regionally, nationally, and locally. The World Health Organization (WHO) Resolution Process emerged as a pivotal avenue for policy change, with specific attention to APARDO's engagement with WHO SEARO. Furthermore, the Global Network for Rare Diseases (GNRD) was highlighted as a key platform for collaboration, with a focus on developing regional patient care pathways and addressing existing gaps. The initiative GardAccess was also spotlighted for its role in finding solutions to facilitate medicine access at both local and national levels. The local perspective, as shared by Nadiah Hanim from Malaysia, sheds light on the tangible benefits derived from RDI alliances. The exchange of knowledge among member countries was evident, underscoring the idea that collaboration leads to more impactful outcomes compared to individual efforts. The leadership role of RDI in influencing local governments and transforming aspirations into actionable initiatives was particularly emphasized. Insights from Dr. Klara Yuliarti in Indonesia highlighted progress

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		in newborn screening efforts. However, the emergence of multiple Patient Advocacy Groups (PAGs) signaled a need for a unified voice to prevent conflicting priorities and enhance engagement with the government effectively. Doreen Tan's contribution emphasized the transformative power of collaboration and networking, reinforcing the idea that a united voice is instrumental for catalyzing positive change. Moreover, the crucial role of early diagnosis in improving access to care, treatment, and knowledge for patients with rare diseases was underscored. Dr. Ngu's perspective from Malaysia further emphasized the tangible impact of collaboration with regional and global organizations. This collaboration was identified as pivotal in lobbying efforts to secure government recognition and support for rare disease community initiatives, with Patient Advocacy Groups positioned as effective entities in engaging government policymakers compared to medical professionals. In summary, the first day of the RDI session underscored a collective commitment to shaping a collaborative ecosystem, leveraging global initiatives, and influencing policies for the betterment of individuals living with rare diseases. The emphasis on a united voice, early diagnosis, and strategic alliances sets a promising tone for future endeavors.
Q&A		In the discussion on prioritizing areas for enhancing the Quality of Life (QOL) for People Living with Rare Diseases (PLWRD) through collaboration, networking, and diagnostics, various perspectives were shared. Nadiah Hanim: Nadiah emphasized the significance of newborn screening as a crucial area of focus. She suggested the creation of an 'open source' channel for sharing information and data. This extends beyond treatment statistics, encompassing day-to-day activities of the Rare Disease (RD) community worldwide, promoting a collaborative approach. Alex: Alex highlighted the importance of policy advocacy for impactful and sustainable change. Stressing that strong data is a prerequisite for effective advocacy, she emphasized the pivotal role of well-informed policies in shaping the landscape for PLWRD. Klara Yuliarti: Dr. Klara underscored the importance of inborn screening, particularly for RD conditions without direct treatment options. This approach allows patients and their families to receive supportive care. Additionally, she emphasized the role of alliances with patient organizations in minimizing the gap

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		between diagnosis and support. Data emerged as a crucial component in advocacy efforts, especially in engagements with government entities. Doreen: Doreen emphasized the need for a long-term policy on Patient Journey, specifically focusing on the psychosocial burden associated with the prolonged commitment of patients and their families in the RD community. This holistic approach aims to address not only medical aspects but also the emotional and social dimensions of the patient journey. Dr. Ngu: Dr. Ngu acknowledged the abundance of white papers on the subject of RD and stressed the importance of translating these ideas into actionable implementations. This perspective highlights the need for practical applications of theoretical concepts, ensuring that strategies outlined in research are effectively executed. In summary, while each perspective shed light on different priority areas, common themes include the importance of data, policy advocacy, and addressing the psychosocial aspects of the patient journey. The consensus is that actionable, well-informed strategies are crucial for improving the QOL of PLWRD.
Screening		**Closed Screening - Premier of UNSEEN** The movie “Unseen” of a kid with Angelman Syndrome sheds light on the challenges faced by caregivers, particularly exploring the emotional journey of a mother who, despite being physically present, often feels emotionally unseen while caring for her son with Angelman Syndrome. This poignant portrayal underscores the broader issue of caregivers' mental health, aligning with the suggested approach of empowering caregivers through potential social enterprises. In the context of the film, the caregiver's experience of feeling "unseen" serves as a metaphor for the emotional toll and the often invisible struggles faced by those who dedicate their lives to caring for individuals with disabilities. The narrative becomes a powerful illustration of the need for initiatives aimed at supporting the mental health of caregivers. The concept of a social enterprise tailored to address this specific issue becomes even more relevant. By creating a platform that not only acknowledges but actively works to uplift the mental well-being of caregivers, such a social enterprise could offer support groups, counseling services, and resources. It could serve as a community hub where caregivers can share their experiences, exchange coping strategies, and find solace in a network that truly understands their journey.

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		The film, therefore, becomes a compelling narrative that advocates for a holistic approach to caregiving—one that not only considers the medical needs of individuals with Angelman Syndrome but also recognizes and addresses the emotional and mental well-being of the unsung heroes behind their care. It reinforces the importance of initiatives, whether in the form of social enterprises or other support structures, that empower and uplift the mental health of caregivers who navigate the often challenging and emotionally taxing journey of caring for loved ones with special needs.

Date: November 26, 2023 (Sunday)
Time: 9AM to 3:30PM
Venue: Pullman Kuala Lumpur City Centre, Malaysia

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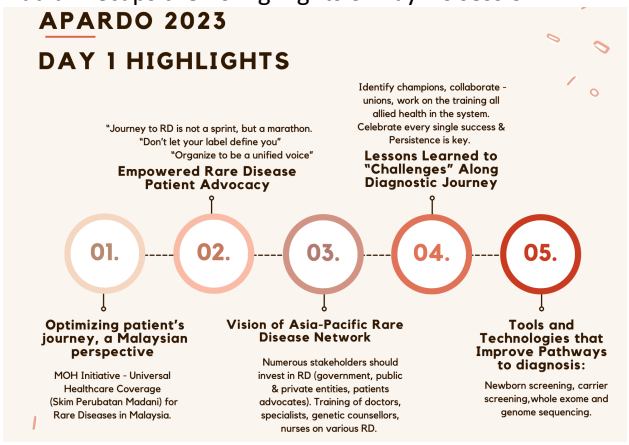
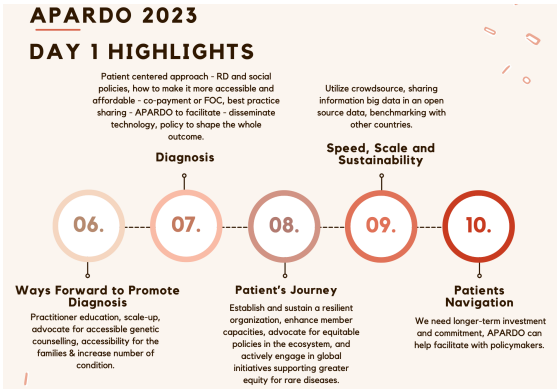
Intention Setting and Daily Meditation	Ms. Nadiah Hanim Abdul Latif (MRDS)	Nadiah recaps the 10 highlights of Day 1's session -  
Keynote Address	Dato' Dr Muhammad Radzi (Director-General of	**Advancing Rare Disease as a Key Priority in Universal Health Coverage in Malaysia & the Region**

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	Health, Ministry of Health Malaysia)	Dato' Dr. Muhammad Radzi expressed his honor to participate in the conference and shared insights into Malaysia's commitment to addressing Rare Diseases (RD). Reflecting on Malaysia's engagement with organizations like APARDO and MRDS, he highlighted the significance of early detection and diagnosis of RD, positioning Malaysia at the forefront of research, screening, and diagnosis. He emphasized the challenges posed by RD, especially in the context of the COVID-19 pandemic, underscoring the need for adaptability. Dato' Dr. Radzi conveyed the Malaysian government's commitment to supporting RD through funding and allocation, acknowledging the limitations in implementation. He reiterated the government's pledge that "No one is left behind," endorsing the importance of MRDS in society. Key Commitments and Initiatives by Malaysia: Government Support: • Specific budget allocation for RD within the government's budget. • Commitment to provide support indirectly through the Ministry of Health (MOH). • Endorsement of the "Health White Paper" in June 2023, focusing on healthcare financing as a strategy to address the burden on stakeholders Healthcare Access and Acknowledgment: • Ensuring increased access to healthcare, including RD, for all individuals. • Recognition and support for caregivers and societies associated with RD patients. Ongoing Efforts and Collaboration: • Continued budget allocation for RD management. • Improving healthcare delivery for RD, including laboratory support • Encouraging cross-sector collaboration involving all stakeholders and developing guidelines. • Addressing orphan drug access and continuous improvement National Trust Fund and Funding Strategies: • Establishment of a National Trust Fund to ensure sufficient funding for RD. • Exploring robust funding mechanisms beyond the medical fraternity, including philanthropists and entrepreneurs through Corporate Social Responsibility (CSR).

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		Integrated Inter-Ministries Approach: • Integrating RD into the broader healthcare network. • Aligning with UN initiatives to address RD challenges through multistakeholder integration. Dato' Dr. Radzi acknowledged the historical neglect of RD in mainstream healthcare and outlined key challenges, including early screening, diagnosis, scientific exploration, and the need to honor the rights of RD patients. He presented the Health White Paper of June 2023 as a crucial step in reforming the healthcare system to make it sustainable and inclusive, leaving no one behind, including RD patients and caregivers. He concluded by expressing his hopes for the conference to be impactful in taking actions that make a difference for the future of RD.
Session 1	Moderator: Ms. Monica Ferrie (GSNV) Presentations: Dr. Durhane Wong-Rieger (APARDO & RDI) Mr. Jason Colquitt (Across Healthcare) Ms. Andrea Rogers (Across Healthcare) Dr. Rachel Yang (CHARD)	**Topic: Innovative data platforms for patient powered databases and patient data platforms** **• Benefits and Risks of using real-world data & real-world evidence to optimize patient outcomes** **• Introduction to patient registries** **• MATRIX Patient Platform** **• Introduction to AI and Patient Data Application** During the conference, Dr. Durhane presented on the importance of seeking funding beyond the government to invest in Rare Diseases (RD), emphasizing the impact on treatment processes and access. She highlighted the need for evidence collection to demonstrate the worthiness of such investments, emphasizing the crucial role of providing accessible information to patients and caregivers. Dr. Durhane's presentation titled "Benefits and Risks of using real-world data for patient-centered affordable/accessible advanced therapies" delved into the role of real-world data in advancing therapies for RD. She posed critical questions about patient engagement throughout the drug life cycle, emphasizing the need to identify disease burden, understand drug efficacy, and determine long-term impacts on patients. The presentation explored the challenges and benefits of utilizing real-world data, including limited resources, heterogeneity of RD, and the importance of early collaboration among various stakeholders. Another presentation from the Vice President of MATRIX introduced the MATRIX Patient Platform, a shared platform spanning 120 countries and 82 groups to aid in the care and cure of RD. This platform facilitates the co-production of care, featuring online tools to track symptoms, visualize insights, and share data with wearables. The presentation also introduced the MATRIX database platform, showcasing its user-friendly interface, customization options, and multilingual accessibility.

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		Dr. Rachel Yang discussed the integration of IT professionals with medical personnel, exploring how Artificial Intelligence (AI) can benefit RD patients. The presentation covered the relationship between AI and humans, various data types, and the application of AI in predicting treatment response, guiding patient selection, and clinical trial design. Case studies were presented, highlighting the use of AI in precision medicine, with an emphasis on patients having ultimate control and understanding the algorithms. Monica emphasized the importance of collecting meaningful data, highlighting the responsibility of all stakeholders to provide quality input for AI systems. The idea that "scarcity makes it valuable" was noted, underscoring the significance of quality data in AI processes. In summary, the conference presentations covered a range of topics, from funding and real-world data to patient platforms and the integration of AI in the context of Rare Diseases. The common thread emphasized the importance of collaboration, data quality, and patient engagement for effective advancements in RD research and treatment.
Session 2	Moderator: Dr. Durhane Wong-Rieger (APARDO & RDI) Presentations: Mrs. Alexandra Heumber (RDI) Dr. Martin Schulz (Pfizer) Discussion/Q&A Roche Representative Ms. Julie Cini (Advocacy Beyond Borders)	**Topic: Access to Treatment – What are Rare Therapies? What are essential medicines for rare and orphan indications? What are emerging innovative therapies?** 1. Mrs. Alexandra Heumber (RDI) - WHO Essential Medicine Lists for Rare Diseases: Rare Disease International (RDI) operates in 150 countries with a focus on essential medicines for Rare Diseases (RD). Challenges include limited access, high costs, and insufficient awareness regarding RD treatments. The WHO Model List of Essential Medicines is reviewed every two years, with the next review scheduled for 2025. RDI's Global Access Working Group plays a crucial role in advocating for the inclusion of RD treatments in the WHO essential medicine list. Next steps involve RDI engaging with WHO during the review process, emphasizing the importance of including rare diseases like Thalassemia, Sickle Cell Disease, Haemophilia, Primary Immunodeficiencies (PID), Osteogenesis Imperfecta (OI), and PID diagnostic tests. 2. Dr. Martin Schulz - Gene Therapies for Rare Diseases: Dr. Schulz, representing Pfizer Rare Disease, highlighted the global prevalence of RD, with only 5% having treatments among the 10,000 known RD. Pfizer focused on gene therapy as a transformative approach, targeting the underlying cause of a disease at the cellular level.

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		He explored various aspects of gene therapy, including gene editing with CRISPR/cas9, gene regulation with siRNA and microRNA, and the use of viral vectors (rAAV, lentiviral). He then discussed the potential benefits of gene therapy for patients, caregivers, healthcare systems, and society at large. Noted the progress in clinical trials, with expectations of 13 new gene therapies by the end of 2023 and 10-20 more by 2025. Dr Martin Schulz: Gene therapy may offer greater one-time clinical benefits than those offered by conventional treatment, from transformational science to routine care - organ transplant, bone marrow and gene transfer). 3. Dr. Durhane - Reimbursement and Access Therapy Challenges: Dr. Durhane addressed challenges in reimbursement and access to therapy for RD. She then emphasized the need for patients' voices to be part of decision-making processes. She also highlighted the role of technology in providing different perspectives on RD, enabling integration with patients' expectations and a better understanding of access issues and treatment value. 4. Essential Medicines for RD - WHO Model List 2023: The WHO Model List of Essential Medicines is updated every two years, with the next update scheduled for 2025. The update considers the national disease burden and clinical need to improve access, emphasizing satisfying priority healthcare needs. The 2023 model list recognizes that the low prevalence of a disease should not be a reason to exclude medicine for its treatment, acknowledging the importance of addressing RD in essential medicines. In summary, the conference sessions discussed crucial aspects of essential medicines for RD, highlighted the potential of gene therapies, and addressed challenges in reimbursement and access. The emphasis on patient involvement, technological advancements, and the continuous efforts to update essential medicine lists for RD underscored the commitment to advancing treatments and care for individuals with rare diseases.
Session 3	Dr. Durhane Wong-Rieger (APARDO & RDI) Presentations: Ms. Alison Keetley (Janssen) Ms. Harpreet Ram	**Topic: Access to Treatment – Options for Funding What are Financing Options for Timely, Affordable Sustainable Access?** Patient-Centric Healthcare and Reimbursement:

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	(GARDaccess) Discussion/Q&A Dr. Rachel Yang (CHARD) Ms. Anne Sophie Chalandon (Sanofi) Ms. Nadiah Hanim Abdul Latif (MRDS)	Janssen Asia Pacific's mission and vision align with APARDO, recognizing the crucial role of patient input in patient-centric healthcare. The focus on reimbursement involves analyzing payer information, systematically including factors like systematic inclusion and willingness to pay. Ms. Allison emphasized the use of a framework based on a rubric for capturing patient voices, divided into "How" and "What." The "How" included aspects like patient partnership, transparency, diversity, timeliness, governance, and mechanisms. The "What" encompassed outcomes related to patient care and patient reports. Findings revealed that the process of patient partnership and transparency, measured through feedback from patient representatives and public documents, was crucial. However, patient involvement did not always lead to better patient outcomes due to economic, cultural, and social differences and limited measurement of health-related quality of life. Best practices from the Asia-Pacific (APAC) region included incorporating patient representatives, training, and providing feedback to facilitate meaningful input. Recommendations included establishing a transparent process, increasing payer accountability, collecting a broader perspective, providing guidance to decision-makers, and measuring the outcome of patient involvement on access to healthcare innovations. Patient Engagement in Healthcare Decision-Making Across AP Region: HTA and Beyond: This talk focused on how patient voices are collected and reviewed in reimbursement policy, emphasizing patient experience. Ms. Allison highlighted the importance of robust life data and patient experiences in shaping healthcare decisions. HTA session- patient input is critical in patient centric healthcare. Patient have unique experience but rely on the healthcare system, reimbursement bodies Study done analyzed, how payers gather information beyond what the company submit in the dossier, what factors support systematic inclusion and appropriate Identification of parameters capturing the patient voices- based on the HC Rubric Themed How (pt partnership, transparency to pt, diversity, methods, timeline, governance, mechanism) and What

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		A systematic literature was done 160 datas found, more publication now to recognize patients needs, ethical legal and societal impact. Bias improvement can be made to incorporate patients feedback and input, Patient involvement does not necessarily lead to better patient outcome. Hypothesis: Healthcare system with higher degree of patient of patient engagement would provide better access to medicine Best practice already happening (Taiwan, Australia, Singapore) • Establishing pt representatives as partners in value assessment and funding decision • Incorporate patient input in a timely and systematic manner • Training to ensure representative are equipped to speak on behalf of relevant patient population and provides structure feedback to facilitate meaningful input Recommendation - Transparent process to systematically collect the pt view, increase accountability of payers when utilizing pt input in decision making processes • Collect broader view that is relevant and meaningful patients • Provide guidance to decision maker as how they should weigh the qualitative patient input • Measure the outcome of the patient engagement to access to healthcare innovations Unlocking Access to RD Medicines (GARDAccess): GARDAccess, a collaboration of public and private funding, service providers, manufacturers, governments, patients' organizations, and medical experts, aims to deliver Rare Disease (RD) medicines to low and middle-income countries. The 2022 deliverables include setting guidelines and implementing programs for RD medicines. The initiative involves developing digital tools, platforms, and apps. Phase 1 is funded by Moderna, Takeda, Tanner Pharma, UCB, and Vertex in collaboration with Trent University. In summary, Ms. Allison Keetley's presentations emphasized the critical role of patient voices in healthcare decision-making, especially in reimbursement policies. The focus on transparency, collaboration, and digital tools showcases a comprehensive approach to improving access to healthcare innovations, particularly for rare diseases in low and middle-income countries. GARDACCESS session - has to be a collective effort - call to collaboration (Objective: Recognize that solving the challenges of RD access requested the collective wisdom and resources of

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		stakeholders from various sectors Project Background - Phase 1 Deliverables • Develop operational framework for an alliance of partners to address the challenges in access tiRD medicines in LMIC • Develop a set of guideline documenting best practice guidelines in developing and implementing the programs for access especially for LMIC • Develop digital transformation • Vision: Has equitable access for every RD patient
Session 4	Moderator: Ms. Nadiah Hanim Abdul Latif (MRDS) Dr. Claudia CY Chung (Hong Kong Genome Institute) Ms. Jasmine Goh Chew Yin (KK Women's and Children's Hospital Singapore) Dr. Ritu Jain (DEBRA International & APARDO) Dr. Dong Dong (Illness Challenge Foundation) Ms. Jinah Kim (Korean Organization of Rare Diseases) Jee Won Choi (AstraZeneca Korea) Ms. Shakira Jamil (Spinal Muscular Atrophy Malaysia)	**Topic: Connecting to Care** Dr. Durhane opened with her highlighting the importance of APARDO as a global conference that is working towards global resolution. The ability to pull together not only nationally, but also regionally is important. Nadiah, this session's moderator, goes on to assert that although the reality is that there is a pressing need to discuss a variety of issues, there is not enough time to sufficiently cover every facet of our lives. She then invites Dr. Claudia forward. DIscussing the socioeconomic impact of RD in Hong Kong, Dr. Claudia states that RD patients in Hong Kong have a prolonged diagnostic journey. The direct impact of RD on patients encompasses family care, care staff, care setting, community support, as well as external factors. With RD affecting 1.4 billion people globally, it is emerging as a global public health priority. Thus, the World Health Organization recognizes the importance of universal health coverage. In Hong Kong, 1 in 67 individuals has one or more rare diseases. The disparity between prevalence and cost reflects the importance of rare diseases in healthcare policies, especially considering how direct and indirect costs accounted for >61% of total RD expenditures, which are typically self-financed out of pocket. Based on the financial protection indicator, direct payments made to obtain health services should not expose people to financial hardship and not threaten living standards. Across the board, RD patients and carers had lower utility scores, indicating the need to address this issue. In her conclusion, Dr. Claudia stated that a genetic diagnosis is crucial in ending the diagnostic odyssey to improve quality of life and health outcomes. Following that, Ms. Jasmine Goh, a genetic nurse from KK Women's and Children's Hospital Singapore, touched on how it is important to be a bridge to the healthcare professionals and also a voice for the RD community. She underscored the necessity of coordinating care and advocating for patients. Despite the lack of a formal education for genetic nurses, Ms. Jasmine introduced a platform that is aimed at connecting

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		genetic nurses so that they can share best practices and guidelines as well as communicate with one another. Currently, www.gnnrd.com links 130 nurses across 30 countries globally. Session by Jasmine: • Recommendation • Adapt and adopt the other disease domain • Promote on networking between families and patient • Training of HCP in language and context • Conferences • Construct ethical framework • Translated resource • Genetic counselling • Community education and awareness Dr. Ritu Jain from DEBRA International took the stage. Her presentation began on behalf of Dr. Roy Gomez, titled "The stigma associated with genetic testing for rare diseases - causes and recommendations". Be it stereotypes or labels, stigma can impede genetic testing for RD, exacerbating the life of someone with RD. An individual living with a rare disease may be inflicted with a mark, which could be physical, metaphorical or even figurative. Stigma is typically categorized according to three distinguishing attributes: perceived, anticipated, and internalized. This may be as far-reaching as impacting the way healthcare systems exclude patients with genetic diagnosis of RD from reimbursement. In her own personal journey, Dr. Ritu opened up about how stigma even affected her own husband's perception of getting genetic testing for their daughter. She then spotlighted learnings on addressing stigma in other disease domains. To address the perceived lack of data from China, Dr. Dong Dong, from Illness Challenge Foundation, prepared her presentation with the aim of calling attention to the RD patients and communities in China. Although there is no definition for RD in China, there is a national list of recognized dieases. She goes on to breakdown the demographic distribution of people affected by RD before emphasizing the economic burden of RD. She calls for all the stakeholders to engage and collaborate in public advocacy efforts. Additionally, she brought up the financial support available from the Illness Challenge Foundation. Session by Dr Dong Dong - has a list of 120 designated rare diseases that was recently increased to 207 • In China they have 9 years of free education, but many RD patients need to leave school and job and lose opportunity to be productive • Diagnostic experience considerably shortened to within a year or 3.95 years • Many patients travel across the country to different provinces to get dx and tx

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		• More than 51% are misdiagnosed • Patient experience with tx, less than 2/3 are receiving treatment – among those who have never been treated, more than 40% claim treatment is too expensive • Huge population seek rehabilitation as treatment • Parents of the child need to work harder to have sufficient income • Many reports suffering from low QoL • The caregiver has less social support and emotional support than needed • Important to have clinical education, community empowerment, financial support, public advocacy (example: concert with all RD) Jinah Kim, of KORD, took the stage with her interpreter to shed light on the establishment called Angel Spoon. The purpose is to collect opinions and patients' needs, to shape policies on orphan drugs and for RD patients, to raise funds and develop various activities for patient welfare, to help RD patients and families to address their medical care issues, to protect their rights, to educate and to improve social welfare. There are 77 groups of RD patients in KORD, which was originally founded in 2001 and established as a corporation alliance in 2003. KORD focuses its efforts on national policy development, medical welfare services, cultural welfare services, communications, patient support, and patient shelter management. Parallel to that, Ms. Jeewon Choi, from AstraZeneca Korea, discussed patient centricity in Korea. In line with AZ's shared mission to transform the lives of rare disease patients through developing innovative medicines, the company acquired Alexion Pharmaceuticals. PAG engagement is pivotal in understanding patient needs so that companies can make use of their unique insights and co-create solutions that will ultimately help to improve outcomes. The key aspects of PAG engagement are sincerity, sustainability, and patient centricity. She goes on to showcase the work done for patients thus far, and to explore a case study on delivering patients' voices in other disease areas. Ms. Shakira Jamil of Spinal Muscular Atrophy Malaysia took to the podium. She told the story of how SMAM came to fruition, starting off as a Facebook support group before being set up as an NGO in 2017. With its vast network of patients, carers and advisory doctors, the organization provides information to the public on SMA to enhance public awareness. From educating patients on how to apply medicine, to providing them with letters for job applications and even consulting physicians and medical experts, the organization supports patients and their carers in many ways. As a carer herself, Ms. Shakira highlighted that not all hospitals understand RD. Patients have specific needs which might not be addressed if their carers are not with

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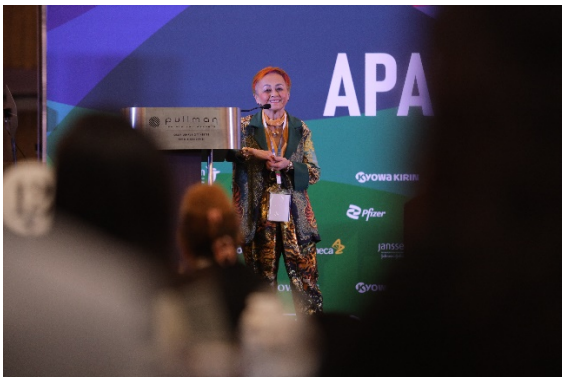
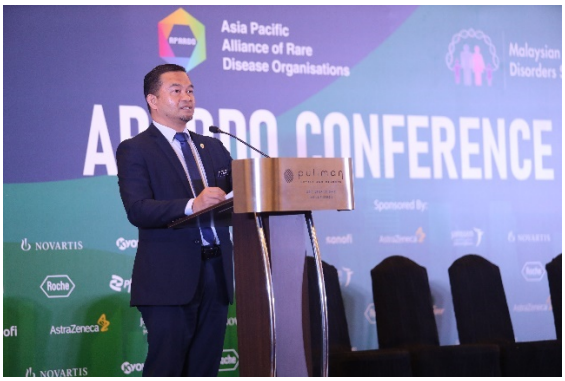
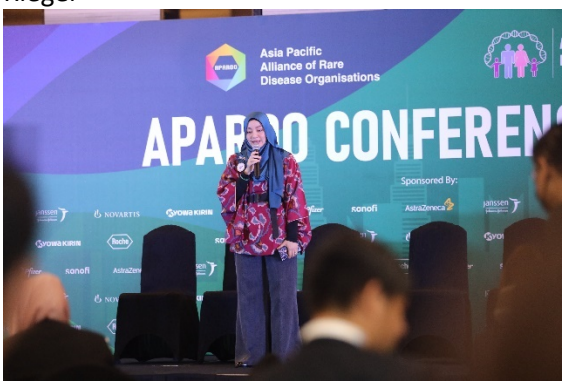
		them. Thus, there is a need to collaborate with other NGOs to achieve a shared goal that emphasizes treatment, newborn screening, and genetic testing to lead to a better quality of life. In an emotional conclusion, Ms. Shakira shines a spotlight on her own daughter, who has SMA Type 2 and was not expected to survive through adulthood. Yet, she has a strong presence in APARDO. This truly showed that a diagnosis is not a death sentence.
Closing		**Conference Summary Next Steps & Closing Remarks** Dr. Durhane closed the conference by thanking the sponsors and audience for their immense support. She stated that MRDS set a very high bar for future APARDO conferences. As the event came to an end, she emphasized that collaboration across the region is crucial to connecting on specific areas, be it diagnosis and screening or research and development.



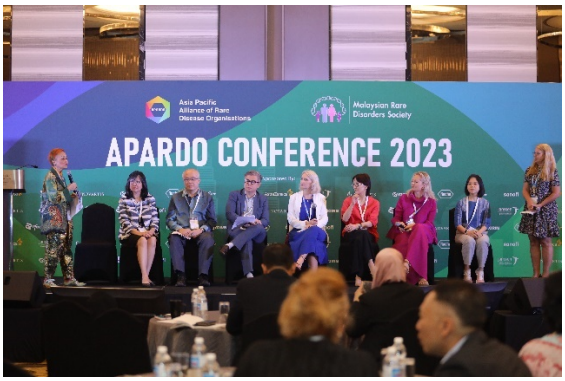
(Day 1) Welcome Remarks: Dr. Durhane Wong-Rieger



(Day 1) Keynote Address: YBhg. Dato' Dr Azman bin Yacob

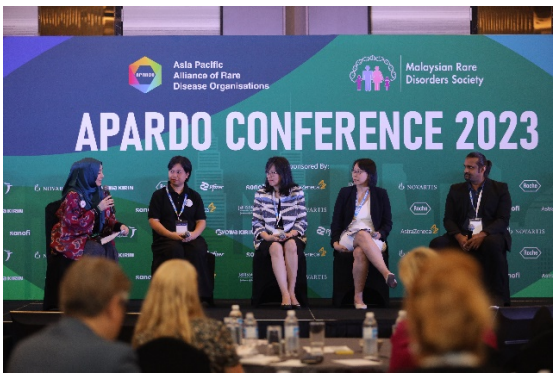


(Day 1) Opening: Ms. Nadiah Hanim Abdul Latif

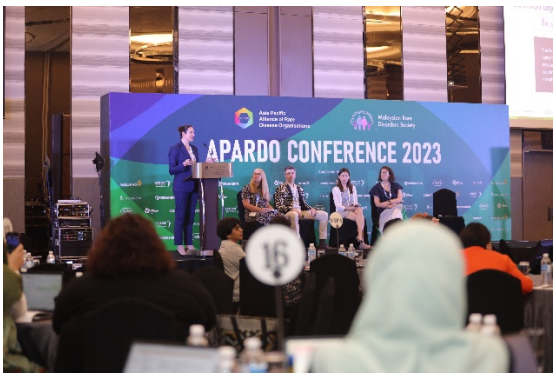


(Day 1) Session 1:
Ms. Monica Ferrie
Ms. Nadiah Hanim Abdul Latif
Dr. Durhane Wong-Rieger

Panelists:
Dr. Carmencita Padilla
Dr. Kittiwan Rojnueangit
Dr. Thong Meow Keong
Dr. Fariz Abdul Rani
Ms. Anne Sophie Chalandon
Dr. Rachel Yang
Mrs. Alexandra Heumber



(Day 1) Session 2:
Ms. Nadiah Hanim Abdul Latif



(Day 1) Session 3:
Ms. Tara Pensa

Asia Pacific
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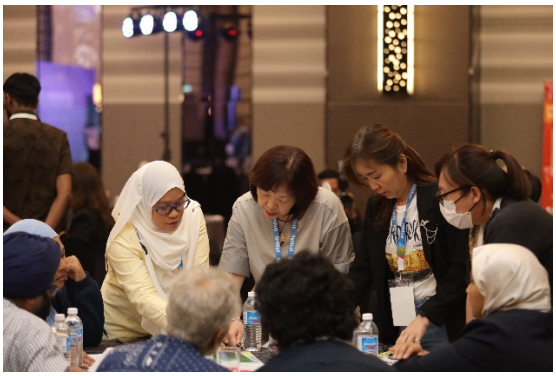
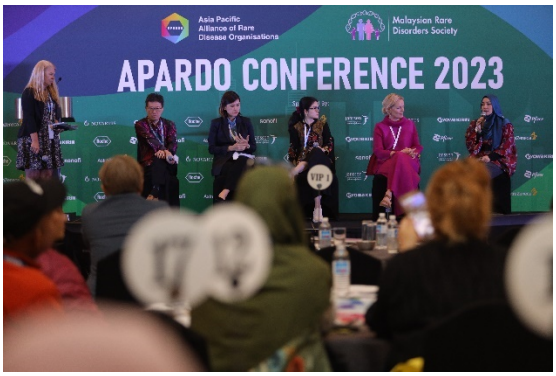
Disease Organisations

Ms. Sook Yee Yoon
Dr. Carmencita Padilla
Ms. Jasmine Goh Chew Yin
Mr. Sivasangaran Kumaran

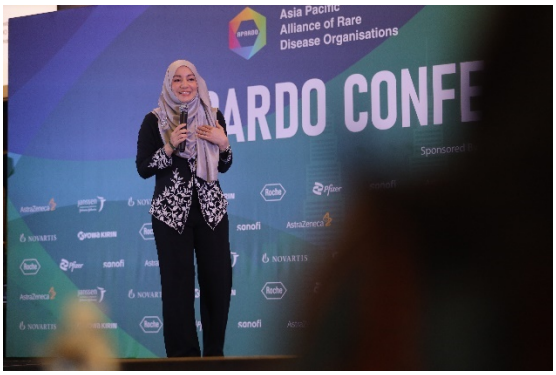
Ms. Monica Ferrie
Mr. Will Greene
Dr. Claudia CY Chung
Dr. Alison Archibald



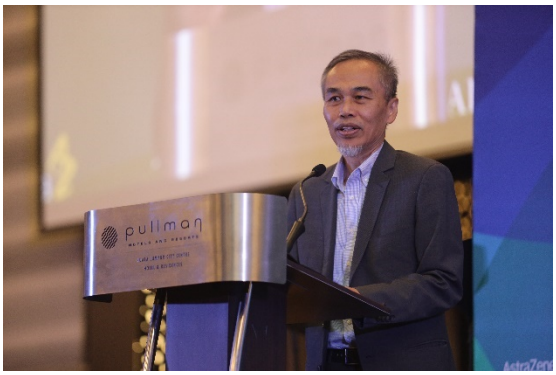
(Day 1) Session 4: Workshop



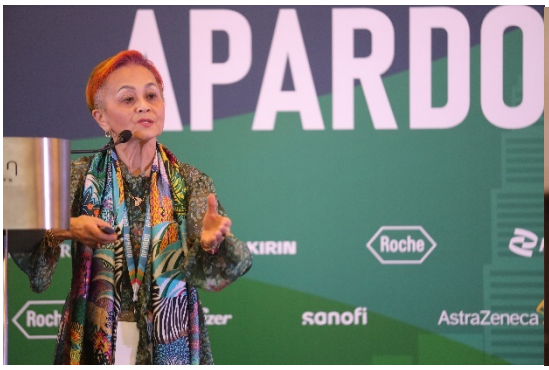
(Day 1) Session 5:
Ms. Monica Ferrie
Dr. Ngu Lock Hock
Ms. Doreen Tan
Dr. Klara Yuliarti
Mrs. Alexandra Heumber
Ms. Nadiah Hanim Abdul Latif



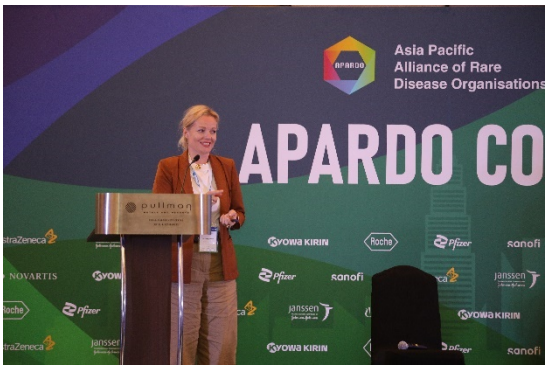
(Day 2) Intention Setting/Recap:
Ms. Nadiah Hanim Abdul Latif



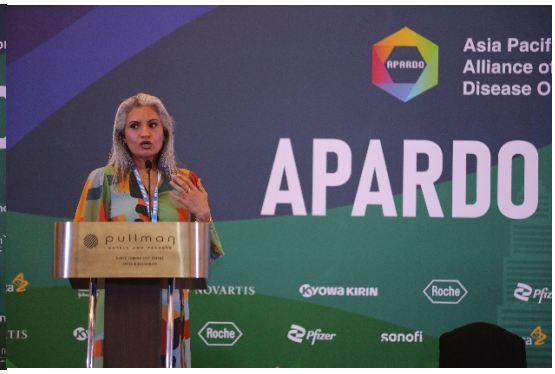
(Day 2) Keynote Address:
Dato' Dr Muhammad Radzi

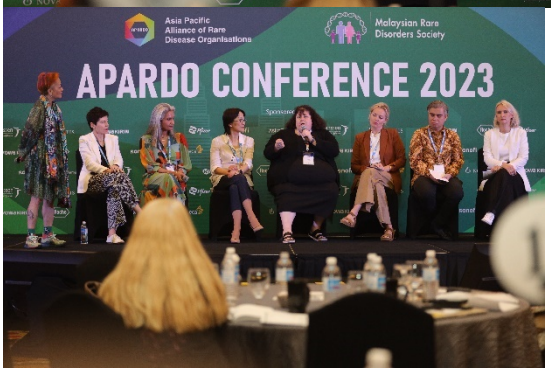


(Day 2) Session 1:
Dr. Durhane Wong-Rieger
Dr. Rachel Yang
Mr. Jason Colquitt (video presentation)
Ms. Andrea Rogers (video presentation)



(Day 2) Session 2:
Mrs. Alexandra Heumber
Dr. Martin Schulz (video presentation)

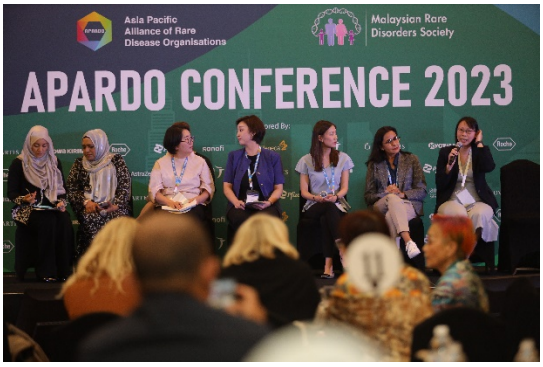




(Day 2) Session 3:
Ms. Alison Keetley, Ms. Harpreet Ram

Asia Pacific
Alliance of Rare
Disease Organisations

Panel/Q&A: Dr. Durhane Wong-Rieger, Ms. Alison Keetley, Ms. Harpreet Ram, Dr. Rachel Yang, Ms. Julie Cini, Mrs. Alexandra Heumber, Dr. Fariz Abdul Rani, Ms. Anne Sophie Chalandon



(Day 2) Session 4:
Ms. Nadiah Hanim Abdul Latif
Ms. Shakira Jamil
Ms. Jinah Kim
Ms. Jee Won Choi
Dr. Claudia CY Chung
Dr. Ritu Jain
Ms. Jasmine Goh Chew Yin
Dr. Dong Dong

Group Photo (Day 1)

Group Photo (Day 2)

Event Photos: Link Here

LIST OF 66 PARTICIPATING ORGANISATIONS

Advocacy Beyond Borders
APARDO
AstraZeneca (Korea)
AstraZeneca (Thailand)
AstraZeneca (Malaysia)
Biogen
Canadian Organization for Rare Disorders
Cawangan Komunikasi Penghakupayaan Orang Kurang Upaya (CKPOKU)
China Alliance for Rare Diseases (CHARD)

Chinese Organization for Rare Disorders (CORD)
Cornelia de Lange Syndrome
DEBRA International
Epidermolysis Bullosa Management Services
GARDaccess
Genetics Counselling Society Malaysia
Harapan OKU
Hemophilia Society of Malaysia
Hong Kong Genome Institute
Illness Challenge Foundation
Illumina
IncluCity
Indonesia Rare Disorders
Indonesian Pediatric Society / Human Genetic Research Centre (HGRC) / IMERI / Faculty of Medicine Universitas Indonesia
Institute for Medical Research, Malaysia
Janssen
Kidney Friends Association of Thailand
KK Women's and Children's Hospital Singapore
Korean Organization for Rare Diseases
Kuala Lumpur & Selangor Albinism Association
Kuala Lumpur Hospital
Kyowa Kirin Asia Pacific

Kyowa Kirin Malaysia Sdn Bhd
Loh Guan Lye Specialists Centre
Lysosomal Storage Disorders Support Society
Malaysian Rare Disorders Society
MIMS Medica
MRKH Malaysia
NakSeni
Novartis
Parents of Little People Malaysia
Penang Hospital
Persatuan Orang Kerdil Sabah (PROKESA)
Persatuan Penyakit Motor Neuro Malaysia
Persatuan Pesakit Imunodifisiensi Primer Malaysia MYPOPI
Persatuan Sindrom Apert Malaysia
Persatuan Sindrom Prader-Willi Malaysia
Persatuan Spinal Muscular Atrophy Malaysia
Pfizer Inc
Pfizer Malaysia
Philippine Society for Orphan Disorders
Psoriasis Association of Malaysia
Queen Elizabeth Hospital
Rare Disease Hong Kong

Rare Diseases International
Rare Diseases Society Nepal
RD Youth Lead
Roche
Roche Diagnostics Asia Pacific
Sanofi
Thai Rare Disease Foundation
Universiti Kebangsaan Malaysia
Universiti Putra Malaysia
University of Malaya
Victorian Clinical Genetics Services

Vietnamese Organization for Rare Diseases

WeCare Journey
Yayasan Mucopolly Sacharidosis Dan Penyakit Langka Indonesia