People Living with Rare Skin Conditions in ASEAN: Time for Action and Hope

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INTRODUCTION

The Rare Skin Conditions Society (Singapore) (RSCS) and the Asian Society of Paediatric Dermatology (ASPD) convened the region's first patient advocacy group (PAG)-led regional summit focused on rare skin conditions. This landmark event united over 50 patient advocates and clinicians from Singapore, Malaysia, Thailand, Vietnam, the Philippines, and Indonesia.

The summit, 'Beyond the Surface: 1st Regional Rare Skin Patient Support Summit' touched on the challenges, barriers and best practices in managing rare skin conditions across the region from a clinician and lived experience perspective, touching on rare conditions such as epidermolysis bullosa, congenital ichthyrosis, Netherton syndrome, and neurofibromatosis.

In Southeast Asia, although rare diseases may sound uncommon, but they have been found to affect as affect over 45 million people across the ASEAN region.[1] Most of these diseases present at birth or in childhood, and many families go through years of confusion and anxiety just for a diagnosis. This is due to a combination of a lack of medical knowledge of healthcare providers in under-served communities, and a lack of access to diagnostic tests, such as genetic testing. This also results in limited opportunities to develop care pathways, build experience and optimise standards. Even when treatment exists, it is usually scarce or too expensive for most families. Life with a rare disease can be difficult, but it does not have to stay this way.

We believe that no one should be left behind simply because they have a rare disease. Recently, the World Health Assembly (WHA) passed a landmark resolution calling on all governments to take action on rare diseases.[2] This marks a significant moment for our community and gives us hope that our needs will finally be recognised.

[1]Shafie, A.A., Chaiyakunapruk, N., Supian, A. et al. State of rare disease management in Southeast Asia. Orphanet J Rare Dis 11, 107 (2016). https://doi.org/10.1186/s13023-016-0460-9 [2]World Health Organization. (2024). EB156/15: Follow-up to the Political Declaration of the Third High-level Meeting of the General Assembly on the Prevention and Control of Noncommunicable Diseases (Rare Diseases). Available at: World Health Organization website

IDENTIFIED GAPS

Taking the opportunity to leverage on the collective experience and wisdom of the participants present, working groups were formed to discuss on the barriers, issues and recommendations facing those with rare skin conditions, exploring several themes:

1. VOICES OF INDIVIDUALS WITH RARE SKIN CONDITIONS

- **Limited collaboration and a lack of shared decision-making among patients, caregivers, and healthcare professionals hinder optimal care outcomes for rare skin conditions.** Promoting shared decision-making will empower patients, caregivers, and medical professionals to work together in developing personalised care plans. Collaboration should extend to patient support groups, medical societies, government agencies, industry partners such as pharmaceutical companies, andhealthcare institutions to enhance education, training, and research on rare skin conditions.

- **Barriers to accessibility and information-sharing restrict the ability of patients and providers to respond to emerging needs and advancements in rare skin condition care.** Expanding the use of technology,including telemedicine, online platforms, and digital media,will improve access to care, facilitate educational outreach, and support community engagement across diverse regions.

- **PLWRD and their caregivers often experience isolation and encounter significant barriers when seeking support and information within their communities.** Efforts must be made to identify the unique issues faced by patients and caregivers, and to foster a greater sense of belonging within affected communities. The formation of regional and local civil societies and patient support groups, will further strengthen community support networks and advocacy efforts.

2. PUBLIC AWARENESS AND EDUCATION

- **Low public understanding and persistent misconceptions about rare skin conditions continue to hinder effective support for patients and families.** Information should be designed and disseminated to address misconceptions, dispel fears, and be tailored to specific audiences and communities, including schools, primary or community care settings, workplaces, libraries, and online forums. Efforts to increase awareness should extend across multiple platforms to reach broader audiences by utilising innovative channels, such as providing books in school libraries, which can help address stigma and promote inclusivity.

- **Public awareness about rare skin conditions should not be confined to lived experience stories in mainstream media or educational talks among healthcare professionals in tertiary healthcare settings.** Effective communication strategies are lacking to explain diagnostic issues and therapeutic challenges of PLWRD to the public. Strategic improvement in communication about rare disease may increase awareness and advance science. Challenges faced by teams supporting the awareness of rare skin conditions in non-governmental organisations or patient groups include staffing, funding, technological resources, training and expertise.

Digital Marketing	Educational Materials	Public support
Social media platforms with active presence (e.g., Facebook, Instagram, Tiktok)	Brochures and pamphlets in hospitals, polyclinics and schools, etc to educators, counsellors, parents etc.	Lived experience sharing at public health seminars
Interactive platforms (e.g., Whatsapp Community)	Books stocked in school or public libraries	Fundraising events supported by influential personalities
Websites (supported with SEM and SEO optimisation)	Knowledge sharing through peer support programs	
	Documentaries, or media interviews	
	CMEs, CNEs etc.	

3. CAPACITY BUILDING AND SOCIAL SUPPORT

- **Significant gaps exists in the availability and integration of rare disease care within community settings, which can hinder access to necessary support.** Health services embedded within community environments play an ongoing role in advancing rare disease care through targeted legislation and program development. Such approaches can lessen the burden on people living with rare skin conditions and their caregivers, ensuring responsibility for care does not fall solely on individuals,particularly in environments where access or influence may be limited, such as social support systems in schools, workplaces, or other communal settings.

- **A lack of robust community and support networks limits access to vital peer support, advocacy, and shared learning opportunities.** Development of peer-support programs can connect experienced patients and caregivers with families of newly diagnosed rare skin patients, facilitating the exchange of practical guidance and emotional support. Strengthening collaboration among rare disease organisations, dermatological societies, and patient advocacy groups will help unify messaging and enhance the effectiveness of advocacy and lobbying efforts. In addition, organising annual conferences and workshops for patients, healthcare professionals, and policymakers will foster knowledge sharing, highlight advances in research and policy, and disseminate best practices for the management of rare skin conditions.

- **Societal stigma and lack of awareness profoundly affect the wellbeing of individuals with rare skin conditions, limiting social integration and access to support.** Reducing stigma requires coordinated public awareness initiatives, as well as engagement campaigns with schools, parents, employers and the general public. Comprehensive education efforts should be implemented to increase understanding and acceptance of rare skin conditions across societies.

- **Insufficient awareness and expertise among healthcare professionals present ongoing challenges, especially in rural and underserved areas.** Healthcare capacity must be strengthened by integrating rare skin condition education into medical school and residency curricula and providing targeted training for providers in all settings, including nurses and midwives in rural and underserved communities. These efforts will help ensure timely, high-quality care and improve outcomes for affected individuals.

4. ENSURING ACCESS TO APPROPRIATE MEDICAL CARE AND SUPPORT

- **People living with rare diseases (PLWRD) and their families face significant challenges due to delays in diagnosis, fragmented care, and insufficient support networks.** The United Nations outlined the vulnerability of this group in 2019, followed by a resolution in 2021 to address these issues. Strengthening care for rare skin conditions require coordinated support through primary care networks, community health centres, and multidisciplinary clinics. Additional help from allied health professionals and the use of telemedicine can improve day-to-day management. Enhanced access to medications and supplies, facilitated by support groups or physician recommendations, is also essential. While registries do not provide care, they offer valuable data to guide improvements in management.

- **People living with rare diseases (PLWRD) and their families struggle to navigate complex health systems, facing psychosocial strain and may result in declining trust in care.** Current resource allocation and training focus on screening and treatment, often at the expense of holistic and personalised support. Timely diagnostics and long-term management for rare skin conditions require multi-disciplinary and tailored approaches that may conflict with common healthcare norms. Effective workforce planning and specialised training are essential, given the acute shortage of rare disease experts. Achieving meaningful change relies on sustained political will and targeted organisational and regulatory action.

- **The lower prevalence of rare skin conditions limits opportunities to improve health literacy across care settings and makes it challenging to standardise cohesive approaches throughout the care journey.** Decentralising diagnosis and care is essential to address these gaps, enabling earlier detection and better access to specialised services closer to where patients live. Building capacity across all specialties and involving nurses and allied health professionals such as wound care nurses, therapists and psychologists, raises awareness of the unique needs and experiences of people living with rare skin conditions. This approach increases the number of contact points along the care pathway, facilitating longitudinal support and timely follow-ups, and ultimately leading to more consistent and effective care experiences for patients.

5. ADEQUATE FUNDING AND RESOURCES

- **A lack of adequate financial resources remains a significant barrier to effective and sustainable rare disease care.** Expand dedicated budgets and adopt early access, managed entry, and value-based reimbursement to accelerate access to high-value therapies. Capturing broader value in health technology assessments (HTA) (including caregiver time and productivity) and leveraging public–private partnerships can reduce long-term care costs, ease caregiver burden, and bolster GDP per capita. Fragmented or insufficient funding may be mitigated by integrating outpatient testing, procedures and care, medicines and mental health support into existing healthcare schemes for chronic conditions, or by working with employers and insurers to develop more inclusive insurance products. National funds should also be earmarked to accelerate the development of a national rare disease registry so that critical data can be provided to regulators to inform and optimise the allocation of resources for financial support and broader care initiatives. There should be ongoing research to find more cost-effective diagnostics and treatment modalities for these conditions.

- **The scope of care for PLWRDs is further limited by funding that is largely confined to traditional healthcare environments and contributes to a diminished quality of life for affected individuals.** Social support mechanisms frequently address only diagnosis and treatment, overlooking broader needs and creating unequal burdens for PLWRD and their caregivers. Schools, transportation, and community services typically lack sufficient resources to meet these needs, such as accommodating flexible schedules or supporting the provision of climate-controlled environments, leaving significant gaps in vital areas such as wound care and mental wellbeing, and restricting optimal support in workplaces, educational settings, and community spaces. Without structured support, financial strain, burnout, and poor mental health intensify care gaps. Embedding caregiver needs in benefits design and health technology assessment can lower system costs, improve outcomes, and protect macroeconomic productivity.

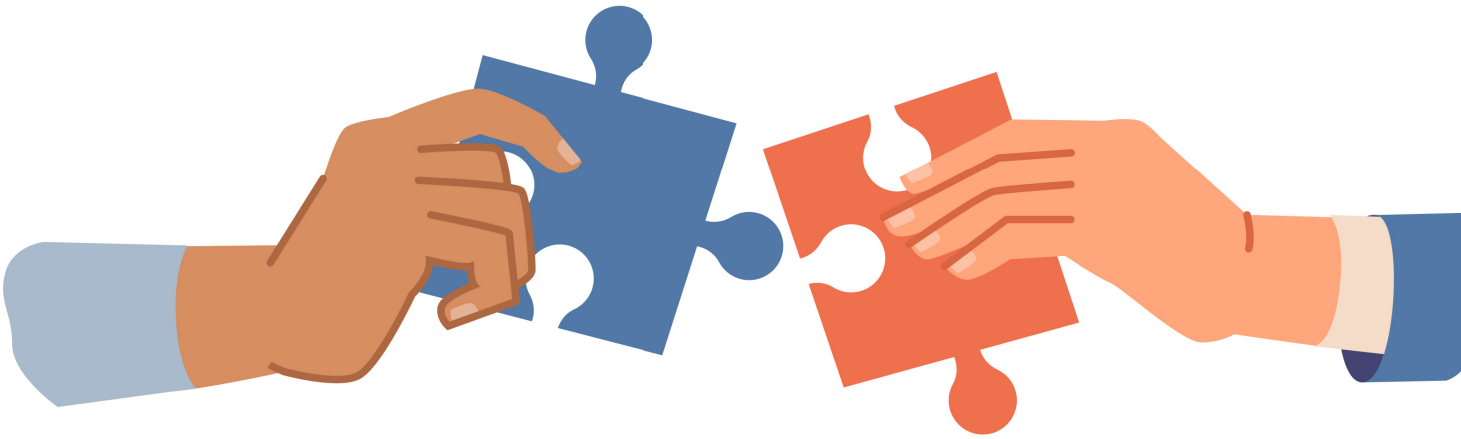
Despite the widespread recognition of corporate social responsibility (CSR) in the business sector, its application within health is underexplored.

CSR initiatives in health can emanate from both within and outside the sector, with corporations able to strengthen health systems and promote health-related activities through targeted engagement and investment. To enhance these efforts, governments must raise awareness of needs-based CSR and facilitate opportunities for corporations to better understand diverse health needs, including those associated with rare skin conditions. National endorsement and advocacy for collective support can help mobilise philanthropic resources and corporate participation, making financial contributions more accessible and impactful for those affected by rare conditions.

6. REGIONAL COLLABORATION AND NETWORKS

- **The lack of comprehensive and regionally coordinated research on rare skin conditions is particularly important for ASEAN collaboration because rare diseases often have low prevalence in each individual country, leading to limited local data and resources.** It is essential to initiate multicenter clinical studies across the region and encourage data pooling to build robust evidence and improve clinical outcomes. Increased investment in medical research on rare skin conditions will drive innovation and enhance standards of care. Equally important is regional collaborative research in the social sciences, which can illuminate key psychosocial, educational, and employment challenges, ensuring that policies are informed by the real-world experiences of affected individuals and families.

- **Rare skin conditions frequently present unique social and cultural challenges that differ across communities within ASEAN.** Governments and health authorities should establish and fund formal knowledge exchange programs for rare diseases, beginning at the community level and progressively scaling up to involve policymakers and government agencies. Such programmes should include regular meetings, virtual forums, patient- and caregiver-led workshops, and cross-sectoral exchanges. This approach will facilitate sustained dialogue, encourage the sharing of best practices and challenges, and ensure that the perspectives of affected families inform national and regional policy development.



- **The absence of dedicated national and regional societies for rare conditions, even down to specific condition types, restricts advocacy, support, and the coordination of research and resources.** The absence of dedicated national and regional societies for rare skin diseases limits advocacy, support, and coordination of research and resources. Establishing such societies, both for specific conditions at the national level and collectively across Asia, would strengthen local knowledge-sharing and enable more effective cross-border collaboration. Regulators should review and, where necessary, amend laws to enable patient-led groups to contribute fully to the wellbeing of people affected by rare diseases.

CONSENSUS STATEMENT –

EMPOWERING LIVED EXPERIENCE PERSPECTIVES

People living with rare diseases (PLWRDs) and our families are essential to driving change. When we share our stories, raise awareness, challenge stigma and advocate for better policies, we demonstrate the strength of our community. Together, our voices are powerful, and what we stand for must be included in decisions that affect us.

With the identified barriers and recommendations, we have refined these insights into six key statements forming a patient-led consensus statement. This consensus brings a lived experience perspective, aiming to inform initiatives across the region and guide stakeholders towards people-centric policy reforms. These statements, shared and discussed during the Summit, will be published and circulated to stakeholders throughout the region, supporting collaborative and inclusive approaches in rare disease policy development.

PLWRD voices matter and must be included in all decisions that affect us. Collaborative, networked care models involving patients, clinicians, experts, community, and civil society empower PLWRDs and their families to access information, peer support, and specialised care, and to advocate for their needs—ensuring that psychosocial aspects of rare disease are addressed throughout the care pathway.

Capacity building and social support across public, schools, workplaces, and healthcare settings, are needed to ensure the implementation of inclusive policies for young and working people living with rare diseases (PLWRDs) in order to strengthen the social fabric.

Adequate funding and resources should be earmarked to that no one is left behind in line with the SDG goals. Meaningful investment to ensure equitable access to better testing, treatments, and patient support will help achieve timely diagnosis, proper care, and affordable treatment for all.

Public awareness and education about living with a rare disease should include factual information, positive portrayals, and lived experiences to reduce stigma, challenge stereotypes, and build support for people living with rare diseases (PLWRDs) and their families

Ensuring access to medical care and support for PLWRDs requires decentralised, person-centred care coordinated across all levels of the health system and community. Lowering barriers to testing and treatment and providing financial and social support will help affected individuals and their families lead fulfilling lives.

Regional collaboration is needed to improve research and understanding of better tracking, innovative treatments, and novel care and financing models—such as using AI for faster diagnosis and new ways to make treatment affordable—to better bridge equity and access gaps for rare diseases.

ABOUT US

To find out more about the Rare Skin Conditions Society (Singapore) (RSCS) and the Asian Paediatric Dermatology Society (ASPD), please scan the QR codes below for further information.

Asian Society of Pediatric Dermatology

Rare Skin Conditions Society