

# PUTTING PWS ON THE MAP: Navigating the world of rare diseases

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## Abbreviations

AFRO - Africa Region

ALIBER - Iberoamerican Alliance for Rare Diseases

APARDO - Asian Pacific Alliance of Rare Disease Organisations

CoNGO - Conference of NGOs in Consultative Relationship with the United Nations

ECOSOC - UN Economic and Social Council

EMRO - Eastern Mediterranean Region

EU - European Union

EURO - European Region

ICORD - International Conference on Rare Diseases and Orphan Drugs

IRDIRC - International Rare Disease Consortium

MoH - Ministry of Health

MoU - Memorandum of Understanding

NGO - Non-governmental organisation

NORD - National Organization for Rare Disorders

PAHO - Region of the Americas

PWSA USA - Prader-Willi Syndrome Association USA

RAB - Rainbow Across Borders

RDI - Rare Diseases International

RONARD - The Romanian National Alliance for Rare Diseases

RPWSA - Romania Prader-Willi Syndrome Association

SEARO - South-East Asian Region

SDGs - Sustainable Development Goals

UHC - Universal Health Coverage

UN - United Nations

WHO - World Health Organization

WHO CGN4RD - WHO Collaborative Global Network for Rare Diseases

WPRO - Western Pacific Region

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## Introduction

This report has been researched and written by Tegan Elliott, an undergraduate student in International Relations at the London School of Economics (LSE). The International Prader-Willi Syndrome Organisation (IPWSO) has been delighted to be part of the LSE's internship scheme for charities which has enabled the production of this report.

IPWSO set Tegan the task to map the international and regional organisations and non-governmental organisations that are relevant to the work of IPWSO and our role as a non-profit membership organisation representing national Prader-Willi syndrome (PWS) Associations and the needs of people with PWS and their families globally.

Prader-Willi syndrome (PWS) is a complex genetic disorder characterised by low muscle tone and failure to thrive at birth, and later evidence of short stature, intellectual disabilities, behavioural and psychiatric challenges, and the development of a chronic feeling of hunger that results in over-eating and in severe obesity and associated health problems.

Multiple studies have shown that between 1 in 15,000 to 30,000 people in the populations studied have PWS, which occurs equally across all races and both sexes, although life-expectancy and population prevalence globally may differ depending on the availability of an early diagnosis and of information, treatments, services and supports.

How can IPWSO make a difference at a global level and which of the possible different identities that people with PWS might relate to should we focus on? People with PWS can variously be seen as a group with a rare disorder, with a specific genetic cause of obesity, with a syndrome associated with endocrine abnormalities, or as people with developmental disabilities.

Whilst each person with PWS is an individual living within the context of their family and their culture and country they are also people with complex needs that if not met lead to considerably increased morbidity and mortality and reduced life-expectancy.

The lens through which we have primarily focused our attention in this report is 'PWS as a rare disorder' but these other identities are also important. The task for IPWSO will be to respond to this report and seek ways of increasing our presence at an international level in order to improve the lives of people with PWS and their families globally.

Tony Holland,

September 2020

## Rare Diseases at a Glance

1. The definition of rare disease varies between countries and many countries do not have a definition.<sup>1</sup>
2. In the EU and Canada, a rare disease is defined as any condition affecting 5 in 10,000 people or fewer.<sup>2</sup>
3. There are over 300 million people living with one or more of over 6,000 identified rare diseases worldwide.<sup>3</sup>
4. Rare diseases impact more people than Cancer and Aids combined.<sup>4</sup>
5. Rare diseases currently affect 3.5-5.9% of the global population.<sup>5</sup>
6. 72% of rare diseases are genetic and the others result from infections, allergies, environmental causes or are degenerative and proliferative<sup>6</sup>.
7. 70% of those genetic rare diseases start in childhood.<sup>7</sup>
8. Only 5% of rare diseases have treatments.<sup>8</sup>
9. Patients with rare diseases are frequently misdiagnosed or undiagnosed.<sup>9</sup>
10. Rare neurological diseases (RNDs,) including neurodevelopmental syndromes make up a significant proportion of the total of rare diseases (over 7% of the total.<sup>10</sup>)

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<sup>1</sup> 'Navigating rare neurological diseases: meeting the challenge for policy makers, patients, and healthcare professionals' Economist Intelligence Unit, (2020)

<sup>2</sup> Ibid

<sup>3</sup> 'What Is a Rare Disease?' <https://www.rare diseaseday.org/article/what-is-a-rare-disease>

<sup>4</sup> 'RARE Facts,' <https://globalgenes.org/rare-facts/>

<sup>5</sup> 'What Is a Rare Disease?' <https://www.rare diseaseday.org/article/what-is-a-rare-disease>

<sup>6</sup> Ibid

<sup>7</sup> Ibid

<sup>8</sup> 'NORD fact sheet,' <https://rare diseases.org/get-involved/educate/raise-awareness/nord-fact-sheet-cover/>

<sup>9</sup> Ibid

<sup>10</sup> 'Navigating rare neurological diseases,' Economist Intelligence Unit

# Rare Diseases at the Global Level

## Why is a Global Effort Needed?

A rare disease affects only a small percentage of the population, yet collectively rare diseases are not rare, affecting over 300 million people worldwide.<sup>11</sup> Rare Diseases International (RDI) highlights that ‘rare disease patients face common challenges derived from the rarity of their conditions.’<sup>12</sup> These common challenges must be tackled internationally, the World Health Organization (WHO) states that ‘while a domestic focus is not particularly problematic for many health issues, for rare diseases it is disastrous.’<sup>13</sup> This is because due to the low prevalence of rare diseases individually, no country or region has the resources to tackle rare diseases alone.<sup>14</sup>

Moreover, disparities in healthcare systems and rare disease management between countries and regions is a significant problem. Rare Diseases International states that, ‘addressing rare diseases on an international level is critical to reduce health inequalities between populations worldwide and ensure that people living with a rare disease have access to the same resources as any other population.’<sup>15</sup> Thus, a global effort on rare diseases is vital not only to make progress on diagnosis, treatments and care but also to ensure those living with a rare disease do not suffer from the disparities in healthcare systems between countries.

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<sup>11</sup> D. Julkowska et al., ‘The Importance of International Collaboration for Rare Diseases Research: A European Perspective,’ *Gene Therapy*, Volume 24,9 (2017): 562-571

<sup>12</sup> ‘Rare Diseases International Vision, Mission, Objectives’ <https://www.rarediseasesinternational.org/vision-mission-objectives/>

<sup>13</sup> ‘Coming together to combat rare diseases,’ *Bulletin of the World Health Organisation*, Volume 90, Number 6 (2012) <https://www.who.int/bulletin/volumes/90/6/12-020612/en/>

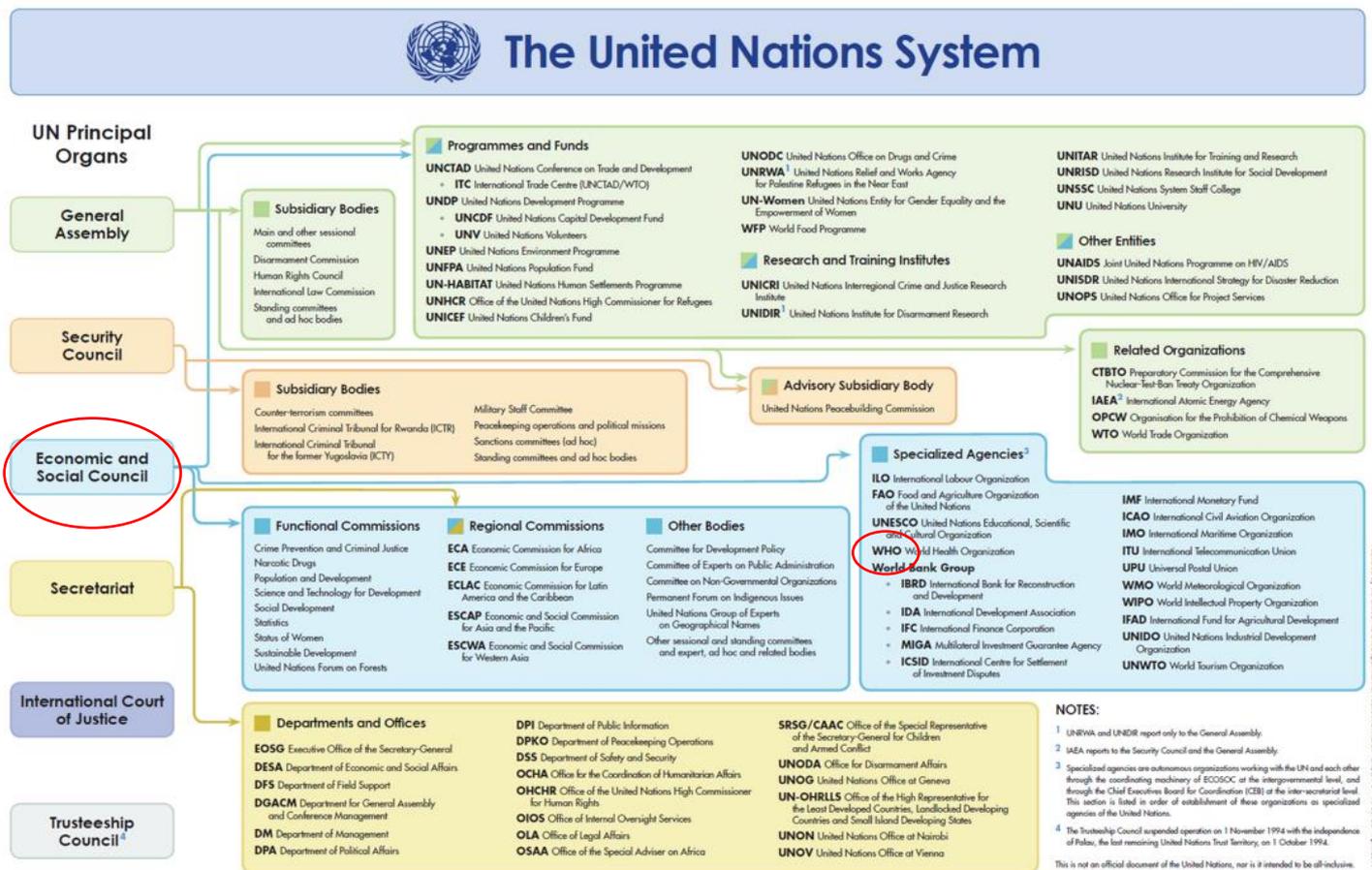
<sup>14</sup> Julkowska, A European Perspective, 562-571

<sup>15</sup> ‘Rare Disease International,’ <https://www.rarediseasesinternational.org/vision-mission-objectives/>

# Key Global Players

## Key Intergovernmental Organisations

Figure 1



## The United Nations (UN)

The UN is an international organisation of 193 member states. As listed on its website, it takes action on issues including peace and security, climate change, sustainable development, human rights, disarmament, terrorism, humanitarian and health emergencies, gender equality, governance, food production, and more.<sup>16</sup> The UN General Assembly adopted the Sustainable Development Goals (SDGs) in 2015 and set targets for 2030 (see figure 2.) The NGO Committee for Rare Diseases highlights that ‘there is a clear alignment between a number of these goals and the fight to tackle rare diseases.’<sup>17</sup> The UN is thus a highly important body for impacting the rare disease world as it takes meaningful actions to achieve its goals that can align with the global effort on tackling rare diseases. With regards to global health its overarching goal is Universal Health Coverage (UHC,) that is ‘all individuals and communities receive the health services they need without suffering financial hardship.’<sup>18</sup>

<sup>16</sup> ‘About the UN,’ <https://www.un.org/en/about-un/>

<sup>17</sup> ‘NGO Committee for Rare Diseases, Common Goals,’ <https://www.ngocommitteerarediseases.org/common-goals/>

<sup>18</sup> ‘WHO UHC’ [https://www.who.int/news-room/fact-sheets/detail/universal-health-coverage-\(uhc\)](https://www.who.int/news-room/fact-sheets/detail/universal-health-coverage-(uhc))

The UN's efforts in global health are led by the World Health Organization, a specialized agency of the UN. In May 2019, the UN Office of the High Commissioner for Human Rights (OHCHR) made reference to persons living with a rare disease in an annual report to the UN Economic and Social Council (ECOSOC.<sup>19</sup>) The report highlighted the use of a human rights framework to contribute to the conceptualisation and implementation of Universal Health Coverage (UHC.) Following this, UN member states included rare diseases in a political declaration on UHC in September 2019.<sup>20</sup>



Figure 2

*The Economic and Social Council (ECOSOC)*

ECOSOC is an organ of the UN and it advances the three dimensions of sustainable development: economic, social and environmental. It takes the leading role in achieving a balanced integration of these three dimensions. It links UN entities that are dedicated to sustainable development, providing coordination and guidance. It also fosters UN partnerships with the rest of the world such as policymakers, foundations and academics. The ECOSOC annual High-Level Segment comprises a High-Level Political Forum focusing on political leadership and sustainable development; and a Development Cooperation Forum focusing on trends and progress in development cooperation. ECOSOC has other sessions such as a Youth Forum and Humanitarian Affairs Segment.<sup>21</sup>

The NGO committee for rare diseases states, ‘ECOSOC stands out as the one forum where a new global debate on rare diseases and their numerous economic and social dimensions must take place, and the one hub through which to create the opportunity to enable other UN

<sup>19</sup> ‘UN Member States Include Rare Diseases in Political Declaration on Universal Health Coverage,’ NGO Committee for Rare Diseases, <https://www.ngocommitteerareddiseases.org/news/>

<sup>20</sup> Ibid

<sup>21</sup> ‘About Us, ECOSOC,’ <https://www.un.org/ecosoc/en/about-us>

bodies and agencies of relevance to understand how rare diseases relate to their own priorities and initiatives.’<sup>22</sup> The UN High-Level Meeting on UHC in 2019, which resulted in rare diseases being included in a political declaration, had the two multi-stakeholder panels within the ECOSOC chamber.

### The World Health Organization (WHO)

The World Health Organization (WHO) is a specialised agency of the UN. WHO’s primary role is to ‘direct and coordinate international health within the United Nations.’<sup>23</sup> The decision-making body of WHO is the World Health Assembly, which is held annually in Geneva, Switzerland and it is structured around a specific health agenda. One of WHO’s top priorities is ensuring Universal Health Coverage (UHC,) including access to diagnosis and treatment for people with rare diseases.<sup>24</sup> Rare diseases are increasingly becoming a priority of WHO, a statement from WHO’s Director General, Dr Tedros Adhanoms Ghebreyesus on Rare Disease Day in February 2018 said:

*“The vision of the Sustainable Development Goals is a world in which no one is left behind, including people who suffer from rare diseases. Just because a disease affects a small number of people does not make it irrelevant or less important than diseases that affect millions”<sup>25</sup>*

The World Health Organization is a highly important body to influence due to the impact of its work on health issues around the world. Following a global immunisation campaign led by WHO, Smallpox was eradicated in 1980. Its work surrounding malaria led to an approximate 60% drop in deaths from the disease between 2000-2015. In 2011, WHO launched a world report on disability for policy makers, professionals, service providers and advocates to use as a key resource.<sup>26</sup>

### Influencing the UN and WHO

As a small rare disease organisation, the most effective way to influence the UN and WHO and thus international health policy is through being a part of the larger rare disease organisations that have created a relationship between them and these intergovernmental bodies. In an interview with Yann Le Cam, on the council of Rare Diseases International (RDI), he explained that influencing these bodies is very hard. Five years ago, RDI could not get an appointment at the UN. This highlights the importance of a small organisation such as IPWSO using its voice within a bigger organisation that has been able to establish a

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<sup>22</sup> ‘Why ECOSOC?’ <https://www.ngocommitteerarediseases.org/why-ecosoc/>

<sup>23</sup> ‘About WHO,’ <https://www.who.int/about>

<sup>24</sup> ‘RDI UN Advocacy’ <https://www.rarediseasesinternational.org/un-advocacy/>

<sup>25</sup> ‘RDI Working with the WHO’ <https://www.rarediseasesinternational.org/working-with-the-who/>

<sup>26</sup> ‘About WHO’ <https://www.who.int/about>

relationship with WHO. It is also important to note that with the current pandemic it is going to be even harder for rare disease organisations to have an influential voice. Getting rare diseases to be included in the political declaration on UHC and other important actions were achieved through the advocacy steps taken by EURORDIS, Rare Diseases International and the NGO Committee for Rare Diseases. This includes meetings with UN officials and the Director-General of WHO. These influential organisations also release joint statements calling for important actions to be taken.<sup>27</sup> Moreover, Yann Le Cam highlighted the great importance of positioning rare diseases within the SDGs in order to ensure the UN listens. The individual work of these rare disease organisations will be discussed further in the next section.

### **Key Rare Disease Organisations, Alliances, Committees and Societies**

#### **The NGO Committee for Rare Diseases**

Founded by EURORDIS, The NGO Committee for Rare Diseases is established under the umbrella of the Conference of NGOs in Consultative Relationship with the United Nations (CoNGO.) CoNGO brings NGO expertise to the United Nations, facilitating the participation of NGOs in the UN system.<sup>28</sup> The NGO Committee's main purpose as listed on its website is 'to bring visibility and understanding about rare diseases to the UN, a platform where they have received little attention until now.'<sup>29</sup> This is in line with the UN's Sustainable Development Goals (SDGs) as it ensures that no individual with a rare disease is left behind.<sup>30</sup>

#### ***Recommendation 1***

*IPWSO is currently not a member of the NGO Committee for Rare Diseases. It should look into becoming an observer member due to the impact this NGO Committee has had on international policy such as the inclusion of rare diseases in the political declaration on UHC in September 2019. International federations have been very active for the NGO Committee for Rare Diseases in the past. For example, the Secretary-General from the International Federation for Spina Bifida and Hydrocephalus gave a presentation on promoting prevention and early interventions at an informal side event hosted by RDI and the NGO Committee for Rare Diseases at the 72nd World Health Assembly in 2019.*

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<sup>27</sup> 'UN Member States Include Rare Diseases in Political declaration on Universal Health Coverage,' <https://www.rarediseasesinternational.org/un-member-states-include-rare-diseases-in-political-declaration-on-universal-health-coverage/>

<sup>28</sup> 'CoNGO' <http://ngocongo.org/>

<sup>29</sup> 'NGO Committee for Rare Diseases Our Actions,' <https://www.ngocommitteerarediseases.org/our-actions/>

<sup>30</sup> 'NGO Committee for Rare Diseases Common Goals' <https://www.ngocommitteerarediseases.org/common-goals/>

## Rare Diseases International (RDI)

As described on its website, RDI is the ‘global alliance of people living with a rare disease of all nationalities across all rare diseases.’<sup>31</sup> RDI’s mission is ‘to be a strong common voice on behalf of rare disease patients around the world, to advocate for rare diseases as an international public health priority and to represent its members and enhance their capacities.’<sup>32</sup> RDI does not currently have the resources for capacity building but focuses on an ecosystem of rare diseases and advocacy.

RDI’s aims:<sup>33</sup>

- Unite, expand and reinforce the rare disease movement of patient organisations and patient advocates
- Put rare diseases on the agenda of international organisations and multilateral institutions such as the United Nations, ECOSOC and the WHO, and on the national agenda of every country around the world
- Strengthen rare disease patient groups’ capacity to act at local, national, regional and global levels and to interact with other rare disease groups

In an interview with Yann Le Cam, the driving force behind the initiation of RDI in 2009 and a member of RDI’s council, he spoke about RDI’s most important advocacy steps.

Positioning rare diseases within the SDGs was very important, when wanting to influence WHO as it is important to come to them with activities that enable them to achieve their goals. Moreover, having something concrete to show these bodies is very important, such as publishing regular reports on a region or bringing together different key players. Yann Le Cam is aiming for an eventual UN resolution on rare diseases.

RDI and WHO have signed an agreement to collaborate under the Memorandum of Understanding (MoU) which comprises two main activities. The first activity focuses on ‘harmonising the way rare diseases are defined internationally.’<sup>34</sup> In an interview with Hlawulani Mkhabela, the Outreach and Engagement Manager at RDI, Hlawulani highlighted that this will be an important process and right now the focus is on developing a framework to get to a definition. For example, it is important that the definition is not based on Western understanding. The definition process must be multi stakeholder and so IPWSO may be contacted for this. The second set of activities focuses on ‘laying the groundwork for the development of a global network of centres of excellence for rare diseases’<sup>35</sup> (WHO CGN4RD.)

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<sup>31</sup> ‘RDI, who are we?’ <https://www.rarediseasesinternational.org/what-is-rdi/>

<sup>32</sup> ‘RDI, Vision, Mission and Objectives,’ <https://www.rarediseasesinternational.org/vision-mission-objectives/>

<sup>33</sup> Ibid

<sup>34</sup> ‘RDI and WHO: towards a global network of centres of excellence for rare diseases’ <https://www.rarediseasesinternational.org/rdi-signs-memorandum-of-understanding-with-the-world-health-organization/>

<sup>35</sup> Ibid

## **WHO CGN4RD:<sup>36</sup>**

The Rare Disease Global Hub will help those living with a rare disease around the world locate centres and expertise in order to access holistic, affordable and highly specialised healthcare for diagnosis, care and prevention of co-morbidities.

### *How these centres will work<sup>37</sup>*

- Members of the Rare Disease Global Hub will be internationally recognised university medical centres that provide healthcare, research, education and technology.
- Centres and expertise will be grouped by therapeutic areas and according to the six WHO regions to ensure no patient or disease area is left behind.
- The GCN4RD acts locally by enabling access to and coordination of high-quality diagnosis, care and treatment as well as supporting the development of local health systems' capacity and competency in rare diseases.
- The GCN4RD thinks globally by creating a framework for collaboration with other global rare disease hubs and international multi-stakeholder rare disease initiatives.

### *How can IPWSO have a voice in RDI?*

#### ***Recommendation 2***

*From interviews with Hlawulani Mkhabela and Yann Le Cam, the biggest emphasis was put on engagement. Engagement in RDI's projects, events, and papers will enable IPWSO to have a voice on an international stage and shape that voice, allowing what is most significant to IPWSO to be heard. RDI has a Facebook Workplace, a virtual space for members to connect and exchange information, this is currently being revamped over the summer and IPWSO should join once this is completed*

Moreover, Rare Diseases International has an Advocacy Committee which as listed on the RDI website is responsible for: 'developing and recommending priorities, initiatives and policies for the Organisation; for preparing position papers or statements or declaration, as well as to review any position, statement, declaration of petition considered for endorsement by the Organisation.'<sup>38</sup> The advocacy committee accepts patient advocates and is open to all members.

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<sup>36</sup> 'WHO Collaborative Global Network 4 Rare Diseases' <https://www.rarediseasesinternational.org/wp-content/uploads/2020/04/RDI-Members-Webinar-WHOCGN4RD-27.04.2020-Final.pdf>

<sup>37</sup> Ibid

<sup>38</sup> 'RDI Committees and Working Groups,' <https://www.rarediseasesinternational.org/working-groups/>

### **Recommendation 3**

*Hlawulani mentioned that if someone from IPWSO wished to join they can make a formal request which is then considered by the RDI council. This could be a very effective way to influence international health policy. It is recommended that IPWSO makes a formal application for membership of the Advocacy Committee. This might be an appropriate role for the CEO of IPWSO.*

With regards to the global centres of excellence for rare diseases, RDI coordinates the project and ensures the needs of the rare disease community are reflected at each stage of the project. IPWSO has been involved in one focus group as part of the EURO Region and there will be regional touch points roughly every two months in which IPWSO will be involved. The hubs will need to be jointly endorsed by a country's health minister and its national rare disease alliance.

### **Recommendation 4**

*It is vital that IPWSO is engaged with national rare disease alliances and encourages its member organisations to build a strong relationship with them. A close relationship with national rare disease alliances allows IPWSO or national PWS associations to have an even greater voice in this project. Additionally, a key part of these hubs will be 'advocate members' representing an international or national population. In order for IPWSO to be an influential advocate member it should be as international and connected to as many people living with PWS as possible.*

The official requirements for advocate members are:<sup>39</sup>

1. Recognised competency in advocates
2. Endorsement by patient organisation board
3. Assessment by independent assessors

IPWSO's voice will be as an international federation involved in a disease specific area and its role as an advocate member will be:<sup>40</sup>

1. Experts by experience in rare diseases
2. Support vulnerable and isolated populations to access care
3. Understand the needs of the rare disease community
4. Capacity building and promotion of health literacy

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<sup>39</sup> 'Towards a WHO Collaborative Global Network for Rare Diseases: Needs Assessment and Concept development,' <https://www.rarediseasesinternational.org/wp-content/uploads/2020/05/4.-WHOCGN4RD-19.05.2020-v3.pdf>

<sup>40</sup> Ibid

## 5. Bridge between the Network and the community

### International Rare Disease Research Consortium (IRDiRC)

As described on their website, the IRDiRC: ‘unites national and international governmental and non-profit funding bodies, companies (including pharmaceutical and biotech enterprises), umbrella patient advocacy organizations, and scientific researchers to promote international collaboration and advance rare diseases research worldwide. Importantly, the coverage of the Consortium is global and involves stakeholders from Africa, Asia, Australia, North America, and Europe.’<sup>41</sup>

As IPWSO does not represent broad patients’ interests for all rare diseases it cannot be a member of IRDiRC. Therefore, IPWSO cannot directly influence the IRDiRC but through having a close relationship with rare disease alliances and international organisations it can be indirectly influential.

### Global Genes

As described on its website Global Genes is a ‘non-profit organization that connects, empowers and inspires the rare disease community, with the ultimate goal of eliminating the burdens of rare disease for patients and families everywhere. They provide hope for the more than 400 million people affected by over 8,000 rare diseases around the globe by helping patients find and build communities, gain access to information and resources, connect to researchers, clinicians, industry, government and other stakeholders, share data and experiences, and stand up, stand out and become effective advocates on their own behalf. As an organization supporting over 675 rare disease foundations, through their RARE Foundation Alliance, and the millions of rare disease patients globally, they provide a platform for the rare disease community to share best practices, develop tools and create larger common data sets that facilitate rare disease research, development and advocacy. ‘Global Genes does a lot of work in ensuring materials are culturally sensitive and also works on making resources accessible to people all around the world.

James O’Brien the Vice President of IPWSO, represents IPWSO on the Global Genes RARE Global Advocacy Leadership Council and so this is how IPWSO can influence the work of Global Genes. Hira Chowdhary, Senior Program Manager of the Global Genes RARE Global Advocacy Leadership Council, stressed that Global Genes continues to build relationships with international federations and organizations to strengthen the network of resources and information to support patients and families, as well as learning from one another about better ways to support the rare disease community.

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<sup>41</sup> ‘IRDiRC, About,’ <https://irdirc.org/about-us/>

## International Conference on Rare Diseases and Orphan Drugs (ICORD)

As described on its website ICORD is: ‘An International Society for all individuals actively involved in rare diseases and / or orphan drugs, including health care, research, academic, industry, patient organizations, regulatory authorities, health authorities, and public policy professionals.’<sup>42</sup>

There is an annual ICORD meeting in which individuals active in rare diseases and/or orphan drugs are brought together, this includes patient groups. In 2019 at the annual ICORD meeting there were many important speakers from all over the world, including Yann Le Cam, Chief Executive Officer at EURORDIS. At this event there was a Panel with families, patients and organisations but the program was predominantly scientific.<sup>43</sup>

To be a member of ICORD costs €50 annually. The application form can be found on their website. This could be useful for IPWSO to join in terms of getting access to new and innovative information on rare diseases however it is not overly useful in terms influencing the global scene.

### **1.3 Key Global Rare Disease Platforms**

#### Social Media

Social Media is an important way to connect with those living with a rare disease and their families. In a white paper published by Evidera, Merinopoulou and Cox (2019) highlight that ‘the need to connect and find support, especially across rare disease communities has led to an increasing number of people with rare diseases and caregivers turning to social media platforms for valuable insight on their disease.’<sup>44</sup> The authors list RareConnect, Facebook groups, publicly accessible disease specific discussion boards and closed-access online communities such as Inspire and Smart Patients as available health-care specific platforms.<sup>45</sup> Overall this paper highlights the extremely positive impact social media can have on rare disease research including getting unsolicited views not subject to potential bias that may be seen in surveys or interviews. Social media is also a good way of disseminating information about rare diseases and care.<sup>46</sup>

#### ***Recommendation 5***

*IPWSO should continue using social media platforms to connect with people and share information. It should continuously check for any new platforms in which people may be*

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<sup>42</sup> ‘International Conference on Rare Diseases and Orphan Drugs’ <https://rarediseases.org/event/international-conference-on-rare-diseases-orphan-drugs-icord/>

<sup>43</sup> ‘ICORD 2019,’ <https://www.icord2019.org/>

<sup>44</sup> Evie Merinopoulo., Andrew Cox., ‘How Social Media can be Used to Understand People with Rare Diseases,’ Evidera, (2019): pg1

<sup>45</sup> Ibid, pg2

<sup>46</sup> Ibid pg3-4

*discussing Prader-Willi syndrome and join current ones. It should use social media for many different tasks such as discovering unmet needs, opinions, side effects of treatments, and people living with PWS in locations where they would normally be hard to find.*<sup>47</sup>

### RareConnect

As stated on its website, RareConnect is:<sup>48</sup> ‘A safe, easy to use platform where rare disease patients, families and patient organizations can develop online communities and conversations across continents and languages. RareConnect partners with the world's leading rare disease patient groups to offer global online communities allowing people to connect around issues which affect them while living with a rare disease.’<sup>49</sup>

On EURORDIS’ connect with others page, RareConnect is listed<sup>50</sup> and appears on many rare disease alliance and organisation’s sites. There are international federations on the RareConnect platform that are part of communities for the particular rare disease, for example the International Niemann-Pick Disease Alliance is part of the Niemann-Pick Disease Type C community.

### **Recommendation 6**

*There is no community for Prader-Willi syndrome on RareConnect currently and this is something IPWSO should look into in order to be as connected and representative as possible. Moreover, in the general discussion on the RareConnect Platform, IPWSO can have access to news and studies on rare diseases that might aid it in its work. For example, there is a recent post on the platform regarding Parents’ experience of caring for children with rare neurodevelopmental disorders.*

### The Mighty

As described on its website, The Mighty is<sup>51</sup>: ‘a safe, supportive community for people facing health challenges and the people who care for them.’ On this platform people can have access to health stories, communities and conversations. The Mighty has a rare disease section<sup>52</sup> with 191k members and 257k likes. This might be a good platform to find people with Prader-Willi syndrome or gain a better understanding of the rare disease community generally. There is a Prader-Willi syndrome section on this page<sup>53</sup> that has 277 members and 26.3k likes.

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<sup>47</sup> Ibid

<sup>48</sup> ‘RareConnect,’ <https://www.rareconnect.org/en>

<sup>49</sup>

<sup>50</sup> ‘Connect with others,’ <https://www.eurordis.org/get-involved-patient-groups-connect>

<sup>51</sup> ‘The Mighty’ <https://themighty.com/>

<sup>52</sup> ‘The Mighty, Rare Disease,’ <https://themighty.com/topic/rare-disease/>

<sup>53</sup> ‘The Mighty, Prader-Willi Syndrome,’ <https://themighty.com/topic/prader-willi-syndrome/>

## **1.4 Key Global Rare Disease Campaigns and Movements**

### *Rare Disease Day*

Rare Disease Day takes place on the last day of February each year with the main objective being to raise awareness of rare diseases. Though primarily it is the general public that is targeted, awareness is also raised amongst policy public authorities, industry representatives, researchers and health professionals.<sup>54</sup> Rare disease organisations all over the world hold events offline and online for Rare Disease Day, in 2020 thousands of events took place over 100 countries. For example, in South Africa there was a Denim Walk by Night fundraiser. The official hashtag for Rare Disease Day also trended in 13 countries and across 5 continents.<sup>55</sup>

### **Recommendation 7**

*IPWSO should ensure that it celebrates Rare Disease Day on all its social media platforms and could organise an online seminar, panel or fundraiser for the day. Moreover, as an umbrella organisation, IPWSO should encourage its member organisations to celebrate Rare Disease Day and join events organised by national alliances wherever possible. Being a big part of Rare Disease Day is another way of being a key part of the international sphere and the common global voice.*

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<sup>54</sup> ‘What is Rare Disease Day?’ <https://www.rarediseaseday.org/article/what-is-rare-disease-day>

<sup>55</sup> ‘Rare Disease Day News,’ <https://www.rarediseaseday.org/news>

## **Section 2 - Regions and Key Regional Players in the Rare Diseases World**

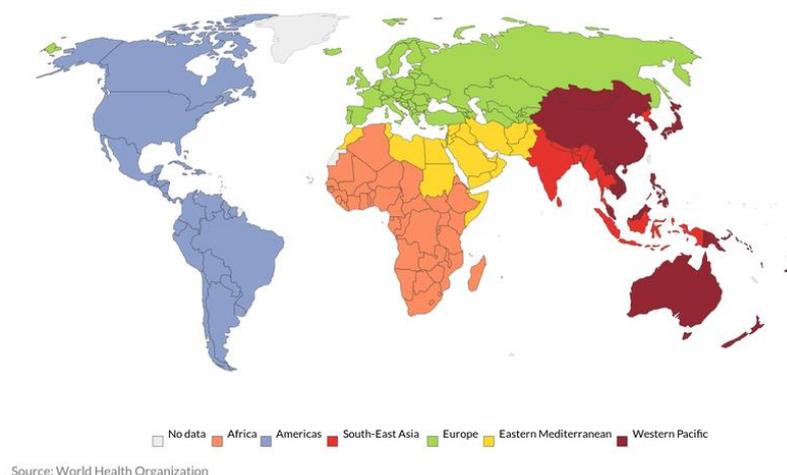
### **2.1 Overview**

#### *What are the Regions?*

For the regional part of this research project, the World Health Organization's regional groupings were used as it is the most significant intergovernmental body with regards to global health policy and these regions are being used in its partnership with Rare Diseases International. A key part of WHO's governance and practice is regionalised and the regions have a significant amount of autonomy over their leadership, budget and priorities.<sup>56</sup> The regions are based on geographical terms but are not synonymous with geographical areas.<sup>57</sup> The 194 WHO members are currently in 6 regions:

- The Americas (35 countries)
- Europe (53 countries)
- Eastern Mediterranean (21 countries)
- South-East Asia (11 countries)
- Western Pacific (27 countries)
- Africa (47 countries)

*Figure 3 - The WHO Regions*



It is important to look at different regions as there are challenges and opportunities unique to certain regions with regards to rare diseases. These will be discussed in this section. Moreover, many regions have regional bodies and rare disease organisations that should be identified in order to get the full picture of the rare disease world.

<sup>56</sup> Elizabeth Free et al, 'At the Roots of the World Health Organization's Challenges: Politics and Regionalization, Am J Public Health, Volume 106, No.11, (2016): pg1912-1917

<sup>57</sup> 'WHO Working with the Regions,' <https://www.who.int/chp/about/regions/en/>

What is important to note, however, is that the WHO regions may not always be appropriate when thinking about policies and actions as there are still large health disparities and socio-cultural differences within the WHO regions. Hlawulani Mkhabela, Outreach and Engagement Manager at RDI, stated that with the current project with WHO, there has been overlapping observed between regions and some countries see themselves in a different region than the WHO regions. Moreover, Hlawulani highlighted that the WHO distinctions will sometimes be too rigid for some projects and movements and the rare disease movement in general does not need to fit into this structure. In many of the papers consulted for this research, the countries in each of the regions varied. For example, in an Orphanet Journal on Southeast Asia there was a mixture of countries from the WHO Western Pacific Region and South-East Asia Region.<sup>58</sup>

### Note on case studies

The case studies included in this report are intended to give a snapshot of conditions in particular regions, however, they are based on the opinions of individuals and may not be representative of the region as a whole.

## **2.2 Africa Region (AFRO)**



### Background

Within nations and as a continent, rare diseases are increasingly recognised as a public health priority in Africa.<sup>59</sup> Following the 11th International Conference on Rare Diseases (ICORD) in 2016, the Africa-Rare initiative was launched, with an emphasis on the importance of international efforts in addressing the impact of rare diseases on global health.<sup>60</sup> At this conference, representatives of the International Rare Disease Research Consortium (IRDiRC) were present and following this the IRDiRC increased its attention on the region and

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<sup>58</sup> Asrul Akmal Shafie et al, State of Rare Disease Management in Southeast Asia, Orphanet Journal of Rare Diseases, (2016)

<sup>59</sup> Gareth S. Baynam et al., 'A call for global action for rare diseases in Africa,' Nature Genetics, Volume 52, (2020), pg21

<sup>60</sup> Ibid

indigenous populations worldwide.<sup>61</sup> However, it is the most vulnerable continent due to a number of issues such as limited financial stability and other serious health problems.<sup>62</sup>

### Challenges in Addressing Rare Diseases in the AFRO Region

Some of the challenges faced in Africa with regards to rare diseases include:

- Urban-rural disparities.
- Lack of recognition of and information on rare diseases including no registry.<sup>63</sup>
- Sustainability of patient organisations.<sup>64</sup>
- Limited funds and resources due to high levels of communicable diseases and socio-economic issues.<sup>65</sup>
- Socio-cultural factors unique to the African context.<sup>66</sup>
- Limited genomic data.<sup>67</sup>

### Case study: Kenya

These are the findings from an interview with Roselyn Odero and Christine Mutena co-founders of Rare Disorders Kenya:

1. Biggest challenge with regards to rare diseases in Kenya:
  - Awareness amongst doctors, policymakers and general public.
  - Urban-rural disparities.
  - Costs.
2. Influence of those representing people with rare diseases on policy making:
  - Very low.
  - Contact with ministry of health before Covid-19 but it has now returned to how it was before, Covid-19 has had a huge impact on progress.
3. Priority given by government:
  - Very low.
  - Government starts with communicable diseases and then when they get to non-communicable diseases the focus is on cancer or diabetes, so rare diseases are not visible.
4. Socio-cultural factors unique to the African context:
  - Belief that God is punishing a family with a child with a rare disease leading people to not seek treatment but for example pray more.
  - Use of witch doctors.
  - Families getting ostracised by the community.

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<sup>61</sup> Ibid

<sup>62</sup> 'Africa-Rare,' <http://africa-rare.org/>

<sup>63</sup> Ibid

<sup>64</sup> Banyam et al, A call for global action, pg23

<sup>65</sup> 'Africa-Rare' <http://africa-rare.org/>

<sup>66</sup> 'Rare Disorders Kenya' <http://rarediseasekenya.org/>

<sup>67</sup> Banyam et al, a call for global action, pg22

- These issues are more prevalent in rural settings but occur in urban settings too.
5. Awareness amongst public:
    - Gets better each year.
    - Lots of engagement with Rare Disease Day, in 2018 there was media coverage and the Minister of Health was involved.
  6. Which organisations have been helpful for Rare Disorders Kenya:
    - National - Non-Communicable Disease Alliance Kenya (NCDAK): NCD organisation in Kenya. There is considerable attention around NCDs in Kenya at present.
    - Regional - Africa-Rare.
    - International - Rare Diseases International (RDI).
  7. Clinical and diagnosis services in Kenya:
    - Referral system takes a long time.
    - Odyssey of GPs taking a family round and round when it is not a GP issue.
    - ‘Luck of the draw’, some people may get referred to a specialist.
    - People in urban centres have a higher level of income so there is a better chance of being treated there.
    - National Health Insurance Fund for public hospital treatment but it is much harder to get the right doctors and medications - to go private you need to fundraise.
  8. Awareness of anyone with PWS:
    - Searching for people living with PWS.
    - Before Roselyn Odero met Tony Holland, she had never heard of PWS.

Regional Rare Disease Organisations:

Africa-Rare

As listed on its website, Africa-Rare is: ‘A joint initiative of African partners for the advancement of recognition and treatment for Rare Diseases in Africa, we aim to bring all umbrella rare disease patient organisations in every country and all regional and international networks for every rare disease into one global community to speak with one voice. We are building the rare disease patient movement of tomorrow.’<sup>68</sup>

It is a South African initiative. International federations can apply to be members. In an interview with Yann Le Cam who sits on RDI’s council, he said that for the moment it comprises predominantly English-speaking countries and there has been difficulty getting French-speaking countries on board. The social media for the organisation has been inactive in the last year.

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<sup>68</sup> ‘Africa-Rare,’ <http://africa-rare.org/>

### **Recommendation 8**

*IPWSO should try to get in touch with this organisation and share resources on Prader-Willi syndrome to be distributed and increase the chances of someone with Prader-Willi syndrome being identified in the region. Moreover, IPWSO could provide advice and guidance on policies and actions.*

### The AFRO region and IPWSO

#### Which countries in the AFRO Region are connected to IPWSO?

Algeria, Angola, Benin, Botswana, Burkina Faso, Burundi, Cameroon, Cape Verde, Central African Republic, Chad, Comoros, Ivory Coast, Democratic Republic of the Congo, Equatorial Guinea, Eritrea, Eswatini, Ethiopia, Gabon, Gambia, Ghana, Guinea, Guinea-Bissau, Kenya, Lesotho, Liberia, Madagascar, Malawi, Mali, Mauritania, Mauritius, Mozambique, Namibia, Niger, Nigeria, Republic of the Congo, Rwanda, São Tomé and Príncipe, Senegal, Seychelles, Sierra Leone, South Africa, Togo, Uganda, Tanzania, Zambia, Zimbabwe.

1/47 countries in the AFRO Region are full members of IPWSO

6/47 countries in the AFRO Region are associate members of IPWSO

7/47 countries in the AFRO Region are connected to IPWSO

## **2.3 Western Pacific Region (WPRO)**



### Background

Some of the countries in this region are advanced in terms of their rare disease management. For example, in an article in an Orphanet Journal,<sup>69</sup> Australia, South Korea and Taiwan have been used as examples of countries with the best practice. In 2018, the Asia-Pacific Economic Cooperation (APEC), which has many members in the WPRO Region, announced

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<sup>69</sup> Asrul Akmal Shafie et al, State of Rare Disease Management in Southeast Asia, Orphanet Journal of Rare Diseases, pg 1

the launch of a Rare Disease Action Plan providing APEC Economies with a rare disease policy framework. However, there are many countries in this region that are not in APEC. WHO highlights that in this region there are great variations in the strength of the health systems and in the economies of the different countries.<sup>70</sup> This is evident in the rare disease management disparities between countries. For example, countries such as Australia and South Korea are far more advanced in their rare disease management than countries such as Laos or Mongolia.

### Challenges in Addressing Rare Diseases in the WPRO Region

Some of the challenges faced in the WPRO Region with regards to rare diseases include:

- Large health and rare disease management disparities between countries in this region.<sup>71</sup>
- Urban-Rural disparities.
- Funding in many countries is a central challenge.<sup>72</sup>
- Misdiagnosis or failure to diagnose in many countries.<sup>73</sup>
- A lack of national rare disease plans across the region.<sup>74</sup>

### Case Study: China

These are the findings from an interview with Dr. Sun Qi from the Chinese Organisation for Rare Disorders (CORD):

1. Biggest challenge with regards to rare diseases in China:
  - Policy on rare disease - everything relies on policy.
  - Diagnosis and medication availability.
2. Socio-cultural factors that pose a challenge to the work of CORD and progress on rare diseases:
  - Some people are shy about seeking help
  - People believe there is something wrong with their heritage.
  - Huge Urban-rural disparities, Rare disease healthcare resources are mostly concentrated in Metropolitans (Beijing, Shanghai and Guangzhou etc.)
3. The level of priority of rare diseases given by the Chinese government:
  - The level of priority is improving.
  - CORD ranked healthcare resources in different provinces to hopefully push the government to improve their policies on rare diseases.
4. Actions taken by CORD that have been successful in improving the quality of life for people living with a rare disease in China:

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<sup>70</sup> 'WHO where we work,' <https://www.who.int/westernpacific/about/where-we-work>

<sup>71</sup> Ibid

<sup>72</sup> Asurl Akmal, Southeast Asia, pg2

<sup>73</sup> Ibid, pg2

<sup>74</sup> Neil Khosla., Rodolfo Valdez., 'A compilation of national plans, policies, and government actions for rare diseases in 23 countries, Intractable and Rare Diseases Research, (2018) pg218

- Empowerment of different patient organisations including helping them become legally established organisations.
  - Education.
  - Advocacy events.
5. Organisations CORD works with:
- Regional - Asian Pacific Alliance of Rare Disease Organisations (APARDO.)
  - International - EURORDIS, RDI.
6. Clinical and diagnosis services in China:
- Good infrastructure for rare disease diagnosis and treatment.
  - Very good in Eastern China.
7. Situation for people with PWS in China:
- Independent PWS organisation that has a healthcare training centre.
  - This organisation is very active and distributes lots of information to paediatric specialists and raises awareness.
  - It is established as a legal organisation.

### Regional Rare Disease Organisations

#### *Asia Pacific Alliance of Rare Disease Organisations (APARDO)*

President: Dr. Ritu Jain

From its website, APARDO is: ‘patient advocate leaders from across the Asia Pacific Region representing rare diseases and rare cancers bound together with the goals of not only providing a forum for sharing experiences and learning but also increasing rare disease patients’ voice and addressing priority issues.’<sup>75</sup>

The APARDO summit in 2019 was supported by Rare Diseases International and had 51 attendees from 17 different countries.<sup>76</sup> APARDO also has a Three-Year Action Plan to strive towards its strategic objectives, this plan can be found on the organisation’s website.<sup>77</sup> It must be noted that many of the countries in this Region are not part of this alliance.

#### ***Recommendation 9***

*IPWSO should get in touch with this alliance in order to establish a relationship and share resources on PWS.*

#### *Rainbow Across Borders (RAB)*

From its website: ‘RAB aims to improve the quality of life of, and give dignity to, patients and their families by chronic or life-threatening illnesses. RAB is Asia’s first regional support group alliance within the Asia-Pacific Region. RAB aims to empower patient support

<sup>75</sup> ‘APARDO, about,’ <https://www.apardo.org/about>

<sup>76</sup> ‘APARDO, 2019’ <https://www.apardo.org/2019speakers>

<sup>77</sup> APARDO, strategic objectives and 3-year action plan,’ <https://www.apardo.org/3-year-action-plan>

organisations through appropriate programmes, services and training while facilitating learning and experience among the RAB affiliates in our network<sup>78</sup>,

## The WPRO Region and IPWSO

*Which countries in the WPRO Region are connected to IPWSO?*

Australia, Cambodia, China, Cook Islands, Fiji, Japan, Kiribati, Laos, Malaysia, Marshall Islands, Micronesia, Mongolia, Nauru, New Zealand, Niue, Palau, Papua New Guinea, Philippines, South Korea, Samoa, Singapore, Solomon Islands, Taiwan, Tonga, Tuvalu, Vanuatu, Vietnam

5/27 countries in the WPRO Region are full members of IPWSO

5/27 countries in the WPRO Region are associate members of IPWSO

10/27 countries in the WPRO Region are connected to IPWSO

## **2.4. European Region (EURO)**



### Background

This region is generally quite advanced with regards to rare disease management, with the EU in particular as a regional body that has a coherent approach to rare diseases<sup>79</sup>. However, like the other WHO regions, there are inequalities in the performance of health systems both within the same country and between countries across the European Region<sup>80</sup>. Europe has the most substantial rare disease regional body, EURORDIS, bringing together 929 rare disease patient organisations from 72 countries.<sup>81</sup>

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<sup>78</sup> 'Join the RAB network,' [http://rabasia.org/affiliate\\_registration](http://rabasia.org/affiliate_registration)

<sup>79</sup> Neil Khosla, 'A compilation,' pg217

<sup>80</sup> 'Health, prosperous lives for all: the European Health Equity Status Report,' <https://www.euro.who.int/en/publications/abstracts/health-equity-status-report-2019>

<sup>81</sup> 'Eurordis,' <https://www.eurordis.org/>

## Challenges in Addressing Rare Diseases in the EURO Region

- Inequalities within nations and between nations.<sup>82</sup>
- Urban-rural disparities.
- Long diagnosis times or misdiagnosis.
- Growing costs of orphan medications due to growing number of these medications.<sup>83</sup>
- Limited dedicated funding for rare disease research.<sup>84</sup>
- Scattered expertise due to scarcity of people living with each rare disease.<sup>85</sup>

### Case study: Romania

Findings from an interview with Dorica Dan, chair of Prader-Willi Association Romania (RPWSA), President of the Romanian Association for Rare Cancers and The Romanian National Alliance for Rare Diseases (RONARD) and a member of the EURORDIS Board of Directors:

1. Biggest challenge with regards to rare diseases in Romania?
  - Half of the population live in rural spaces and so a lot of people living with a rare disease have no access to services because services don't exist in these spaces.
  - In the rural spaces there is no one to coordinate the care and rate of diagnosis is low.
2. Socio-cultural factors posing a challenge to the work of RPWA and RONARD in Romania:
  - Those living with a rare disease are not always proactively engaged in their needs and rights; this is an issue for advocacy and rare disease diagnosis.
3. The level of priority for rare diseases in Romania given by the government and its funding:
  - In 2008, the Romanian government declared rare diseases a priority and a plan for rare diseases was signed with the Ministry of Health.
  - There have been several further steps since then.
  - However, funding is only allocated to a few objectives and others have to be funded by other stakeholders and EU funding.
4. Most successful methods in achieving the successes on rare diseases seen in Romania:

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<sup>82</sup> 'European Health Equity Status Report,' <https://www.euro.who.int/en/publications/abstracts/health-equity-status-report-2019>

<sup>83</sup> 'EURORDIS position paper,' <https://www.eurordis.org/publication/orphan-drugs-rising-challenge-ensure-better-future-30-million-patients-europe>

<sup>84</sup> 'Research and Policy actions,' <https://www.eurordis.org/research-policy-actions>

<sup>85</sup> Charlotte Rodwell., Segolene Ayme., Rare Disease Policies to improve care for patients in Europe, BBA, Volume 1852, Issue 10, (2015,) pg2329

- Most important factor is to have a national alliance with a common voice.
  - Cooperation with professionals for a stronger voice, such as the National Alliance for Human Genetics.
  - Getting journalists involved.
  - ‘Alone we can’t do anything.’
5. RPWA’s connections and relationships with non-rare disease organisations:
- Very important to be aligned and work with disability bodies for example, because the majority of those with a rare disease have disabilities.
  - The NoRo centre has built a national network as part of the European Reference Network.
6. Challenges in childhood and adult life of the management of access to food for those living with PWS:
- Huge challenge that is not approached at a national level.
  - Even in adult life there is a need for supervision all the time.
  - A lot of work to do in terms of raising this challenge at the national and international level.

## Regional Rare Disease Organisations

### EURORDIS

As described on its website, EURORDIS is: ‘A non-governmental patient-driven alliance of patient organisations representing 929 rare disease patient organisations in 72 countries. We are the voice of 30 million people affected by rare diseases throughout Europe. EURORDIS seeks to improve the quality of life of people living with rare diseases in Europe through advocacy at the European level, support for research and medicines development, facilitating networking amongst patient groups, raising awareness, and many other actions designed to reduce the impact of rare diseases on the lives of patients and family. EURORDIS is attracting an increasing number of patient organisations outside of Europe and is gaining traction at promoting rare diseases at the international level.’<sup>86</sup>

EURORDIS is highly involved in advocacy and policy change at a regional and international level. For example, in March 2019 EURORDIS and RDI met with the Director-General of WHO, Dr Tedros, to discuss the inclusion of rare diseases in UHC. In September 2019 rare diseases were included in a UN political declaration on UHC.<sup>87</sup> EURORDIS has contributed to and advises European Union policies on rare diseases. Moreover, EURORDIS also contributes to national processes.<sup>88</sup>

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<sup>86</sup> ‘EURORDIS, what we do,’ <https://www.eurordis.org/what-we-do>

<sup>87</sup> ‘Victory as rare diseases included in UN political Declaration on UHC,’ <https://www.eurordis.org/content/victory-rare-diseases-included-un-political-declaration-uhc>

<sup>88</sup> Rare Disease Policy,’ <https://www.eurordis.org/eu-rare-disease-policy>

### **Recommendation 10**

*IPWSO is not currently a member of EURORDIS but many international federations and PWS national alliances are. IPWSO should consider becoming a member of EURORDIS. Being a member comes with many benefits including informing and shaping European policies on rare diseases. IPWSO meets the criteria to apply and the membership fees are based on an organisation's national budget.*

### The EURO Region and IPWSO

*Which countries in the EURO Region are connected to IPWSO?*

Albania, Andorra, Armenia, Austria, Azerbaijan, Belarus, Belgium, Bosnia and Herzegovina, Bulgaria, Croatia, Cyprus, Czech Republic, Denmark, Estonia, Finland, France, Georgia, Germany, Greece, Hungary, Iceland Ireland, Israel, Italy, Kazakhstan, Kyrgyzstan, Latvia, Lithuania, Luxembourg, Malta, Monaco, Montenegro, Netherlands, North Macedonia, Norway, Poland, Portugal, Moldova, Romania, Russia San Marino, Serbia, Slovakia Slovenia, Spain, Sweden, Switzerland, Tajikistan, Turkey, Turkmenistan, Ukraine, United Kingdom, Uzbekistan.

25/53 countries in the EURO Region are full members of IPWSO

18/53 countries in the EURO Region are associate members of IPWSO

43/53 countries in the EURO Region are connected to IPWSO

## **2.5 South-East Asian Region (SEARO)**



### Background

Generally, good progress has been made on rare disease management within the South-East Asian countries though there are still large challenges.<sup>89</sup> Notably, there is a growing

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<sup>89</sup> Asrul Akmal Shafie et al, 'Southeast Asia,' pg1

awareness of rare diseases due to the development of patient support and advocacy.<sup>90</sup> However as in other regions, there are wide variations across the SEARO Region.<sup>91</sup> For example, Thailand has both a rare disease organisation and a Prader-Willi syndrome organisation whereas Bhutan has no rare disease organisation or PWS group.

### Challenges in Addressing Rare Diseases in the SEARO Region

- Disparities between countries.<sup>92</sup>
- Issues with basic healthcare systems.<sup>93</sup>
- Funding in many countries is a central challenge.<sup>94</sup>
- Lack of patient registries.<sup>95</sup>
- Misdiagnosis or failure to diagnose.<sup>96</sup>
- A lack of clinical expertise.<sup>97</sup>

### Case study: Thailand

These are the findings from an Orphanet Journal of Rare Diseases, '*State of rare disease management in Southeast Asia*<sup>98</sup>'

1. Biggest challenge with regards to rare diseases in Thailand:<sup>99</sup>
  - A lack of clinical rare disease expertise.
  - Urban-rural disparities, most of the geneticists are located in major cities.
2. Level of awareness and advocacy:<sup>100</sup>
  - Increasing awareness.
  - Thailand has a patient support group (and a PWS organisation).
3. Priority given by government:<sup>101</sup>
  - No formal governance structure for rare disease.
  - Limited government funding for rare disease treatment and no government funding for rare disease research.
  - However, Thailand does have very low out-of-pocket expenditure for healthcare in the region and has had success in its mechanism for rare disease funding.
4. Socio-cultural factors:

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<sup>90</sup> Ibid, pg2

<sup>91</sup> Ibid, pg3

<sup>92</sup> Ibid, pg2

<sup>93</sup> Ibid

<sup>94</sup> Ibid

<sup>95</sup> Ibid, pg6

<sup>96</sup> Ibid, pg2

<sup>97</sup> Ibid pg6

<sup>98</sup> Ibid, pg1-8

<sup>99</sup> Ibid

<sup>100</sup> Ibid, pg5

<sup>101</sup> Ibid, pg6-8

- Limited understanding and medicinal support mean patients often feel socially and psychologically isolated.<sup>102</sup>
5. Clinical and diagnosis services:<sup>103</sup>
    - Many cases result in initial misdiagnosis or failure to diagnose.
    - Thailand screens 100% of its new-born babies which enables early intervention but there are still a limited number of specialists.
    - A lack of clinical rare disease expertise, in 2016 there were only 22 geneticists to serve a population of 67 million people.
  6. Situation for people with Prader-Willi:
    - There is a Prader-Willi group, PWS Thailand which is a full member of IPWSO.
    - From the associations Facebook page, there is clinical testing for PWS available in Thailand.

### Regional Rare Disease Organisations

#### *Asia Pacific Alliance of Rare Disease Organisations (APARDO)*

President: Dr. Ritu Jain

From its website, APARDO is: ‘patient advocate leaders from across the Asia Pacific Region representing rare diseases and rare cancers bound together with the goals of not only providing a forum for sharing experiences and learning but also increasing rare disease patients’ voice and addressing priority issues.<sup>104</sup>’

The APARDO summit in 2019 was supported by Rare Disease International and had 51 attendees from 17 different countries<sup>105</sup>. APARDO also has a Three-Year Action Plan to strive towards its strategic objectives, this plan can be found on the organisation’s website.<sup>106</sup> It must be noted that many of the countries in this region are not part of this alliance.

#### ***Recommendation 11***

*IPWSO should get in touch with this alliance in order to build a relationship and share resources on PWS.*

#### *Rainbow Across Borders (RAB)*

From its website:

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<sup>102</sup> Ibid, pg2

<sup>103</sup> Ibid pg2-6

<sup>104</sup> ‘APARDO, about,’ <https://www.apardo.org/about>

<sup>105</sup> ‘APARDO, 2019’ <https://www.apardo.org/2019speakers>

<sup>106</sup> APARDO, strategic objectives and 3-year action plan,’ <https://www.apardo.org/3-year-action-plan>

‘RAB aims to improve the quality of life of, and give dignity to, patients and their families by chronic or life-threatening illnesses. RAB is Asia’s first regional support group alliance within the Asia-Pacific Region. RAB aims to empower patient support organisations through appropriate programmes, services and training while facilitating learning and experience among the RAB affiliates in our network<sup>107</sup>’

### The SEARO region and IPWSO

Bangladesh, Bhutan, North Korea, India, Indonesia, Maldives, Myanmar, Nepal, Sri Lanka, Thailand, Timor-Leste.

2/11 countries in the SEARO Region are full members of IPWSO

3/11 countries in the SEARO Region are associate members of IPWSO

5/11 countries in the SEARO Region are connected to IPWSO

## **2.6 Eastern Mediterranean Region (EMRO)**



### Background

The Middle East and North Africa Region (MENA), which covers many of the countries in the EMRO Region, has one of the highest rates of rare diseases globally.<sup>108</sup> IQVIA estimates that there are close to 3 million patients with rare disease in the MENA Region.<sup>109</sup> Rare diseases in the Middle East receive very little attention amongst policy makers and other

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<sup>107</sup> ‘Join the RAB network,’ [http://rabasia.org/affiliate\\_registration](http://rabasia.org/affiliate_registration)

<sup>108</sup> ‘Orphan Disease in the Middle East and African Region,’ IQVIA, (2018,) <https://www.iqvia.com/locations/middle-east-and-africa/library/white-papers/orphan-disease-in-the-middle-east-and-africa-region>

<sup>109</sup> Ibid

stakeholders despite this high prevalence of people with rare disease.<sup>110</sup> However, there has been increased attention on orphan diseases by large Pharma companies who are looking to commercialise new drugs in the Middle East markets.<sup>111</sup>

### Challenges in Addressing Rare Diseases in the EMRO Region

- Low priority of rare diseases given by governments and key stakeholders.
- Low awareness amongst patients and doctors.
- Inadequate infrastructure for diagnosis and treatment of rare diseases.<sup>112</sup>
- Lack of access to new innovative drugs for the treatment of rare disease conditions.<sup>113</sup>
- Diseases with higher public health implications take precedence.<sup>114</sup>
- Socio-cultural factors such as issues of shame and social stigma.

### Case study: Bahrain

Findings from an interview with Dr. Cristina Skrypnik, Consultant Medical Geneticist, Assistant Professor from the Department of Molecular Medicine, Al Jawhara Center for Molecular Medicine, Genetics and Inherited Disorders, Rare Disorders Day team, Bahrain.

1. Biggest challenge with regards to rare diseases in Bahrain:
  - There are no patient organisations like the ones seen at the European level but some small groups initiated by patients.
  - There are barriers to the creation rare disease alliances.
  - No rare disease registries available at national level.
2. Socio-cultural factors that pose a challenge to the progress on rare diseases:
  - There is a general lack of understanding of rare diseases, which means that people are unable to speak freely about them.
  - There can be different extremes when people find out their child has a rare disorder: some people believe it can be fixed with one pill whereas others believe nothing can be done for improvement.
  - Difficulties with integration, serious issue with shame and social stigma.
3. Level of priority of rare diseases given by the government:
  - There is a level of priority there as the Ministry of Health (MoH) in Bahrain is engaged in the rare disease movement and various disorders have available specific treatments.
  - Not only does the MoH in Bahrain strongly support the rare disease movement but now it is opening the Bahrain Genome Project.
  - There is still a lack of registries, which is something that is now being advocated for.

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<sup>110</sup> Ibid

<sup>111</sup> Ibid, pg5

<sup>112</sup> Ibid, pg7

<sup>113</sup> Ibid, pg3

<sup>114</sup> Ibid, pg4

4. Influence of those representing people with rare diseases on policymaking:
  - Good level of influence.
  - Government knows and works with the academics in the rare disease field.
  - There is a relationship with Government ministers.
  - Over the past ten years things have improved greatly there is a change in mentality and awareness.
5. Level of awareness of rare diseases in Bahrain:
  - Growing awareness and changing mentality.
  - 2013 was the first rare disease national symposium that successfully gathered together the university, genetic centre, governmental hospital and the Ministry of Health, promoting the cause of rare diseases
  - Dr. Cristina Skrypnyk and her team in the university and genetic centre is very active in raising awareness of rare diseases in Bahrain. Campaigns with multiple events for Rare Disease Day are organised every year, for professionals and general public. The university clinic has an advertising screen that also promotes the genetic clinic.
6. Prader-Willi syndrome in Bahrain:
  - Dr. Cristina Skrypnyk has had no patients diagnosed with PWS through the genetic clinic so far but is convinced that there are patients with PWS in Bahrain and may have been patients in the government hospital. Clinical aspects and pathology of PWS are well covered for medical students during their training.
  - If someone believes a member of their family has PWS or a medical doctor suspects this diagnosis, testing for this is available in Bahrain.
7. Clinical services and diagnosis:
  - Good infrastructure for rare disease diagnosis and treatment.
  - People are able to get two or three opinions if wanted.
8. Regional and international organisations that the Al Jawhara Centre works with:
  - Academic institutions.
  - Research and testing genetic laboratories
  - EURORDIS.
  - European School of Medical Genetics

### Regional Rare Disease Organisations

There is currently no regional rare disease organisations in the EMRO Region.  
There is the Arab Organization for Persons with Disabilities.<sup>115</sup>

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<sup>115</sup> 'AODP,' <http://www.aodp-lb.net/>

## The EMRO Region and IPWSO

Afghanistan, Bahrain, Djibouti, Egypt, Iran, Iraq, Jordan, Kuwait, Lebanon, Libya, Morocco, Oman, Pakistan, Palestine, Qatar, Saudi Arabia, Somalia, Sudan, Syria, Tunisia, United Arab Emirates, Yemen.

0/21 countries in the EMRO Region are full members of IPWSO

15/21 countries in the EMRO Region are associate members of IPWSO

15/21 countries are connected to IPWSO

## **2.7 Region of the Americas (PAHO)**



### Background

As written in WHO PAHO's Strategic Plan for 2020-2025, 'Significant inequities in health still persist between and within most countries, with worse health outcomes for populations living in conditions of vulnerability.'<sup>116</sup> This problem of inequity and populations living in conditions of vulnerability can be observed with rare disease management. What is interesting to note is that rare diseases were not mentioned in this document, signalling that rare diseases may be a very low priority in the region. When comparing rare disease management and awareness in the US or parts of Latin America with the Caribbean Region, the difference is stark. Many countries in the Caribbean area for example have no rare disease alliance or organisation at all.

### Challenges in Addressing in Rare Diseases in the PAHO Region

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<sup>116</sup> 'Strategic Plan of the Pan American Health Organization 2020-2025: Equity at the Heart of Health,' PAHO, (2020,)

[https://iris.paho.org/bitstream/handle/10665.2/52473/9789275173619\\_eng.pdf?sequence=1&isAllowed=y](https://iris.paho.org/bitstream/handle/10665.2/52473/9789275173619_eng.pdf?sequence=1&isAllowed=y)

- Significant disparities in health outcomes across groups with less social and economic power such as women and girls, people living in poverty, Afro-descendant and Roma populations, and refugees and migrants.<sup>117</sup>
- Lack of policies that aim to improve health equity.<sup>118</sup>
- Rare diseases not a priority, not mentioned in the PAHO strategic plan for 2020-2025.
- Lack of information and low levels of discussion in parts of the region.<sup>119</sup>
- Lack of registries in many parts of the region.

### Case study: Chile

Findings from an Orphanet Journal of Rare Diseases, '*Rare diseases in Chile: Challenges and recommendations in universal health coverage context*'<sup>120</sup> and an interview with Dr Juan Francisco Cabello, Director of the Diagnostic Centre at the University of Chile.

1. Biggest challenges with regards to rare disease management:<sup>121</sup>
  - Lack of institutions e.g. patient and physician associations.
  - Access to treatment and care.
  - Lack of a registry.
2. Socio-economic and socio-cultural factors posing a challenge to rare disease patients:
  - There are marked differences in access and health outcomes between the private and public systems.<sup>122</sup>
  - Inequities between Chile and its Latin American neighbours such as Bolivia, Chile is much more advanced.
3. Level of priority of rare diseases given by the Chilean government:
  - Level of priority changes with the government.
  - Current government's health minister has been engaged with rare diseases.
  - Chile has the Ricarte Soto law (RSL) aimed at providing funding for the care of patients with rare disease and is assigned a grant of 200 billion pesos for a period of four years.<sup>123</sup>
  - The Ricarte Soto law however is not rare disease legislation, it does not solve the issue of long diagnosis or treatment, only the high cost of medications.

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<sup>117</sup> Ibid, pg9

<sup>118</sup> Ibid

<sup>119</sup> Angela P Chaves, Migdalia Denis, 'Rare Diseases in Latin America: Challenges and opportunities for the equitable attention and proposal of Patients Organizations,' ISPOR, (2017,) <https://press.ispor.org/LatinAmerica/2017/09/rare-diseases-in-latin-america-challenges-and-opportunities-for-the-equitable-attention-and-proposal-of-patients-organizations/>

<sup>120</sup> Gonzalo Encina et al, 'Rare Diseases in Chile: Challenges and recommendations in universal health coverage context,' Orphanet Journal of Rare Diseases, (2019), pg1-8.

<sup>121</sup> Ibid, pg2-5

<sup>122</sup> Ibid, pg2

<sup>123</sup> Neil Khosla, 'a compilation,' pg219

4. Level of awareness and advocacy<sup>124</sup>:
  - Awareness and advocacy are good in Chile.
  - There is the Federation for Rare Diseases (FECHER) and the Federation of Uncommon Diseases (FENPOF), these are the main advocacy groups.
  - FECHER actively contributed to the development of RSL.
  - Both organisations increase awareness of rare diseases and attempt to influence policymakers in the Ministry of Health and Congress.
  - There is a need for more awareness of rare diseases amongst medical students both at undergraduate and postgraduate level.
5. Clinical and diagnosis services<sup>125</sup>:
  - 33 practicing clinical geneticists in Chile, one third less than the recommended ratio.
  - Lack of healthcare professionals who play an important role in addressing psychosocial dimensions of rare disease diagnoses.
6. Prader-Willi Syndrome resources:
  - There is a Prader-Willi Association in Chile that is a member of IPWSO.

The need for a regional and international effort was stressed by Dr. Cabello in this interview, he stated that he had not had contact with ALIBER.

Case study: US

Findings from interviews with Mary Dunkle, Senior Advisor at the National Organisation for Rare Diseases (NORD) and Paige Rivard, CEO of PWSA USA.

1. Biggest challenges with regards to rare diseases:
  - Late diagnosis and misdiagnosis still occur.
  - Very hard to get researchers interested in rare diseases
2. Socio-cultural challenges:
  - Religious factors.
  - Disparities in access to healthcare including rural-urban disparities and racial disparities.
  - Bullying in schools and a general feeling of being alone.
  - PWS specifically - food is everywhere and it is difficult to get people to understand the need to control food access to those with PWS such as grandparents.
3. Clinical and diagnosis services:
  - Big difference depending on where you live, urban-rural disparities.
  - People may not have the financial means to travel to special centres.
4. PWSA USA's connections and relationships with non-rare disease organisations:
  - The connections and relationships with rare disease and non-rare disease organisations are equally as important

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<sup>124</sup> Gonzalo Encina, 'Rare Diseases in Chile,' pg4-5

<sup>125</sup> Ibid, pg3

5. Level of awareness for PWS and rare diseases in general in the USA:
  - Awareness is growing.
  - PWSA Chapters and volunteers – educating families, medical students, doctors, and the general public.
6. Level of priority given by the government for rare diseases such as PWS:
  - That is where NORD is important - it does an important job of advocating for rare diseases.
  - NORD has an office in Washington and frequently meets with members of Congress.
  - Long way to go before rare diseases are a priority.
7. PWSA USA's most successful actions with regards to public policy and influencing the US government:
  - In the past not a huge amount of advocacy on a government level.
  - Trying to advocate for other rare disease communities too as it can be impactful for the PWS community.
8. Key ways a PWS association can make a practical difference to people's lives:
  - Support the families.
  - Advocacy / awareness.
  - Research.

## **Regional Rare Disease Organisations**

### *Iberoamerican Alliance for Rare Diseases (ALIBER) - Latin America*

As described on its website, ALIBER is: ‘A network of 19 organizations of patients with rare diseases, present in 16 countries in Latin America, coordinating actions to strengthen the associative movement, give visibility to the FER and represent people with rare diseases in Latin America before local, regional bodies, national and international, creating a permanent joint space to share knowledge, experiences and best practices in social, health, educational and collaborative work areas.’<sup>126</sup>

### ***Recommendation 12***

*IPWSO should engage with this umbrella organisation, including sharing resources and encouraging the PWS associations in Latin America to connect with this organisation if they haven't already. For example, Prader Willi Uruguay (ACSPW,) is a partner of ALIBER.*

### *NORD and CORD (North America)*

The National Organization for Rare Disorders (NORD) and Canadian Organization for Rare Disorders (CORD) are national organisations rather than regional ones, however, they cover

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<sup>126</sup> ‘ALIBER,’ <https://aliber.org/web/>

the North America Region, so it is important to highlight them. Moreover, they are both well connected internationally.

NORD is, ‘a patient advocacy organization dedicated to individuals with rare diseases and the organizations that serve them. NORD, along with its more than 300 patient organization members, is committed to the identification, treatment, and cure of rare disorders through programs of education, advocacy, research, and patient services.<sup>127</sup>’ Rare-disease specific international organisations can join NORD at platinum level as, ‘International Members.’

### ***Recommendation 13***

*Joining NORD as an international member costs a flat fee of 50 USD. This is recommended as there are some really important benefits from membership. In an email from Allie Crafton from NORD, she stated that NORD would love to support the work of IPWSO by sharing their resources, connections and opportunities. Some of the benefits include financial scholarships, capacity building and support for advocacy.*

CORD is, ‘Canada’s national network for organizations representing all those with rare disorders. CORD provides a strong common voice to advocate for health policy and a healthcare system that works for those with rare disorders. CORD works with governments, researchers, clinicians and industry to promote research, diagnosis, treatment and services for people with any rare disorders in Canada.’<sup>128</sup>

### **IPWSO and the PAHO Region**

Antigua and Barbuda, Argentina, Bahamas, Barbados, Belize, Bolivia, Brazil, Canada, Chile, Colombia, Costa Rica, Cuba, Dominica, Dominican Republic, Ecuador, El Salvador, Grenada, Guatemala, Guyana, Haiti, Honduras, Jamaica, Mexico, Nicaragua, Panama, Paraguay, Peru, Saint Kitts and Nevis, Saint Lucia, Saint Vincent and the Grenadines, Suriname, Trinidad and Tobago, United States, Uruguay, Venezuela.

7/35 countries in the PAHO Region are full members of IPWSO

15/35 countries in the PAHO Region are associate members of IPWSO

22/35 countries in PAHO Region are connected to IPWSO

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<sup>127</sup> ‘NORD,’ <https://rarediseases.org/>

<sup>128</sup> ‘About CORD,’ <https://www.raredisorders.ca/about-cord/>

## **Section 3 – Identity and Non-Rare Disease Organisations**

### **3.1 Overview**

A person with Prader-Willi syndrome will share part of their identity with other people living with PWS but they will also identify with other groups based on different aspects of their life, particular challenges and interests. This is also true for families with a child with PWS. The identity of someone with PWS can sit in the rare diseases field, endocrine disorders, obesity disorders, neurodevelopmental syndromes and intellectual disabilities. It is important to identify the communities and groups that specialise in some of these shared characteristics and challenges in order to get a better understanding of these specific issues.

### **3.2 Interviews**

When carrying out interviews with various specialists and advocates in the field of rare diseases and Prader-Willi syndrome, the need and benefit of connecting with non-rare disease communities and organisations was stressed multiple times. Hlawulani Mkhabela, the Outreach and Engagement Manager at RDI, stated that it was incredibly important for the rare disease community to collaborate with groups with a similar context; rare diseases should not be seen as aside from everything else and advocacy should not be done alone. Hlawulani noted that RDI has made an effort to align the rare disease movement with the human rights movement and disability movement. Dorica Dan, president of RONARD and chair of RPWA also noted the importance of aligning and working with disability bodies as the majority of people living with a rare disease have disabilities; this is something Dorica has done with the NoRo Centre. Paige Rivard, CEO of PWSA USA also noted that PWSA USA has worked with the Munro-Meyer Institute, a developmental disability institute, highlighting that any information they can share is always useful.

#### ***Recommendation 14***

*IPWSO is a small organisation and so there is the concern that it could stretch itself too thin if it tries to align with all the different movements related to Prader-Willi syndrome.*

*Hlawulani noted that strategy is important, don't align with all movements but pick the ones most closely tied with the work and aims of IPWSO.*

### **3.3 Organisations and Alliances**

#### **Down Syndrome International**

Down Syndrome International is an interesting organisation to look at as a model as it aligns with the disability movement. As stated on its website: 'Much of the advocacy work we do is very similar to that of other disabled people's organisations and as members of the International Disability Alliance, we collaborate closely with our disability community

partners at national and regional levels and within the United Nations system.<sup>129</sup> Down Syndrome International also works in partnership with Inclusion International to develop international guidelines on the inclusive participation of people with intellectual disabilities in the running and work of organisations.<sup>130</sup>

### International Disability Alliance (IDA)

The International Disability Alliance based at the United Nations, empowers organisations of persons with disabilities and works at country level promote the rights of persons with disabilities. IDA states that it represents the estimated one billion people worldwide living with disabilities.<sup>131</sup>

#### ***Recommendation 15***

*Though IPWSO cannot be a member of IDA, it would be really interesting to get in touch with them and have a conversation about people with PWS all over the world living with disabilities, the organisation may offer insight, resources or support.*

### Inclusion International

As described on their website, Inclusion International is, ‘The international network of people with intellectual disabilities and their families advocating for the human rights of people with intellectual disabilities worldwide.’<sup>132</sup> IPWSO cannot join Inclusion International as a full member due to it not being a national organisation, however it can become an affiliate member. Being part of Inclusion International has a number of benefits including access to and sharing of resources and information in an issue-based network.

### International Alliance of Patient Organizations (IAPO)

IPWSO is a member of this organisation which is a ‘unique global alliance representing patients of all nations across all disease areas and promoting patient-centred healthcare across the world.’<sup>133</sup> This organisation works on policy and advocacy, capacity building and cross-sector alliances and collaborative working.

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<sup>129</sup> ‘Who we work with,’ <https://www.ds-int.org/who-we-work-with>

<sup>130</sup> ‘Inclusive participation,’ <https://www.ds-int.org/inclusive-participation>

<sup>131</sup> ‘IDA, Who We Are,’ <http://www.internationaldisabilityalliance.org/about>

<sup>132</sup> ‘Inclusion International,’ <https://inclusion-international.org/who-we-are/>

<sup>133</sup> ‘International Alliance of Patients’ Organisations,’ <https://www.iapo.org.uk/>

### **Recommendation 16**

*As a member, IPWSO should be as engaged as possible in the work this organisation does in order to be part of the wider global community of patients. For example, IAPO holds Patient Solidarity Day every December, IPWSO should ensure it publicises this on social media and the website. Moreover, IAPO invites members to High Level meetings to contribute to its discussions with the World Health Organization, if IPWSO builds a good relationship with IAPO it may be able to influence general global and regional health policy.<sup>134</sup>*

### **International Association for the Scientific Study of Intellectual and Developmental Disabilities (IASSIDD)**

As described on their website, IASSIDD ‘promotes the scientific study of intellectual disabilities and related developmental disabilities and of conditions of persons with these disabilities and their families. IASSIDD defines Intellectual Disability as a significant intellectual deficit present from birth, or which originates at an early age or during the developmental period.<sup>135</sup> Membership of IASSIDD provides opportunities to meet and exchange ideas with people having similar interests in other parts of the world and to take part in Special Interest Research Groups.’<sup>136</sup>

### **Recommendation 17**

*IPWSO can join as an individual member, ‘a person or organisation who subscribes to the aims and objectives of IASSIDD,’ for 190 USD. If this is something in IPWSO’s budget it would be very useful in terms of accessing and advancing research on intellectual disabilities.*

### **Endocrine Society**

As described on their website, the Endocrine Society is a global community 18,000 strong energised by the promise of unravelling the mysteries of hormone disorders to care for patients and cure disease. They are devoted to advancing hormone research, excellence in the clinical practice of endocrinology, broadening understanding of the critical role hormones play in health, and advocating on behalf of the global endocrinology community.<sup>137</sup> Part of being a member includes access to Community Connect, Endocrine Society’s exclusive

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<sup>134</sup> ‘Benefits,’ <https://www.iapo.org.uk/why-become-member>

<sup>135</sup> ‘IASSIDD, who we are and what we do’ <https://iassidd2020.com/about-iassidd/>

<sup>136</sup> ‘Membership’ <https://www.iassidd.org/membership/>

<sup>137</sup> Endocrine Society, ‘who we are’ <https://www.endocrine.org/about-us>

online community where you can network, talk to experts, join a conversation with people who share interests, and find what you need throughout your journey with your Society.<sup>138</sup>

***Recommendation 18***

*Members of Endocrine Society are endocrine researchers and practitioners. IPWSO as an organisation can't join but someone within IPWSO who meets the requirements can. This would be very interesting to look into due to their activities and reach,*

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<sup>138</sup> 'Community Connect,' <https://www.endocrine.org/membership/community-connect>

## **Section 4 - Summary**

### **4.1 Summary of recommendations:**

#### **Become as international as possible to achieve two overarching goals:**

1. To reach as many people with PWS as possible in order to improve their lives.
2. Have influence in RDI and the global sphere generally. The more connected IPWSO is to the world, the stronger its voice and influence. RDI and other organisations will view IPWSO as a key organisation to speak and work with, this will enable IPWSO to influence global policies.

How to become as international as possible?

- Engage in international activities and organisations including the NGO committee for Rare Diseases, RDI and EURORDIS.
- Consider joining NORD.
- Reach out to a national rare disease alliance where there is no PWS organisation or alliance - this is key. National rare disease alliances will be very influential in the future, for example in RDI's work with WHO.
- Encourage national PWS associations to form strong relationships with their national rare disease alliance or regional body.
- Where there is no rare disease alliance, reach out to a national disability alliance, they may know someone who might have or has Prader-Willi syndrome.
- Use social media and rare disease platforms such as RareConnect to connect with people and share key resources.

#### **Become as culturally sensitive as possible to those living with PWS in different parts of the world:**

- Develop papers on the variations in experiences of those living with PWS in different countries to better understand this issue.
- Develop resources to help those families in different countries deal with the unique challenges they are facing, such as shame and exclusion from the community.
- Try and keep in regular contact with someone from each region, and countries within that region in order to stay up to date on changing situations and challenges.

#### **Consider ways to overcome issues of inequality in rare disease management between countries:**

- One issue that came up over and over again was inequities between countries and regions.
- IPWSO could develop a paper on this specifically about the challenges people with PWS face in areas with very little to no rare disease infrastructure and awareness.

- Align with the UN's Sustainable Development Goals and add this to the website - here is an example: <https://www.ngocommitteerarediseases.org/common-goals/>
- Advocate for the development of rare disease plans and organisations in countries where this does not exist.

**Link with organisations and groups that are part of the PWS identity:**

- This is key - voices are stronger and more impactful together.
- Connect with disability charities and alliances.
- Look at forums, communities and papers linked with the identity of someone living with PWS.

## **4.2 Resources**

Countries with no PWS association/organisation – who to get in touch with:

### AFRO Region

Algeria - Algeria National Alliance against Rare Diseases (no website)

Botswana - Bordis (no website) <https://www.rarediseaseday.org/association/532>

Ivory Coast - Aux Pas Du Coeur <https://www.facebook.com/pg/auxpasducoeurhibm/about/>

Ghana - Rare Disease Ghana initiative <https://www.rarediseaseghana.org/> b

Kenya - <http://rarediseasekenya.org/>

Lesotho - RARE Diseases Lesotho Associations - <https://www.facebook.com/RDLAnGo/>

Rwanda - Centre-Alliance - <https://www.rarediseaseday.org/association/1040>

South Africa - Rare Disease South Africa <https://rarediseases.co.za/>

Tanzania - Ali Kimara's Rare Disease Foundation - <https://www.facebook.com/alikimarasfoundation/>

Burkina Faso and Guinea – FITIMA <https://fitima.org/contexte-handicap/>

Mauritius – Brave Girls Association <https://www.rarediseaseday.org/event/mauritius/194>

Namibia – National Federation of People with Disabilities Namibia - <http://safod.net/safod-content/cid/93/>

Nigeria – The Borgen Project - <https://borgenproject.org/disabilities-in-nigeria/>

Uganda – National Union of Disabled Persons Uganda

Zambia – Zambia Federation of Disability Organisations, <https://www.zafod.net/>

Zimbabwe – Rare Disorders Trust - <http://raredisorderstrust.co.zw/>

Congo - Lenire Asbl - <https://www.facebook.com/LENIREasbl>

Eritrea - National Association of Intellectual/Developmental Disability in Eritrea (NAIDDE) - <https://www.facebook.com/pages/category/Community/National-Association-of-Intellectual-Developmental-Disability-of-Eritrea-112649872750275/>

Ethiopia - Ethiopian National Disability Network - <https://endanethiopia.org/>

Liberia - My Hearts Appeal - <https://myheartsappeal.org/>

Nigeria -Joint National Association for Persons with Disability - <http://www.jonapwd.org/>

Tunisia - AMED - <https://amedtunisie.com/>

Uganda – EmbraceKulture (<https://www.embracekulture.org/>)

Angel's Centre for Children with Special Needs (<https://angelscentre.org/>)

National Union of Disabled Persons of Uganda(<https://nudipu.org/about-us/>)

### WPRO Region

Philippines - Philippine Society for Orphan Disorders <http://www.psod.org.ph/>

South Korea - South Korea Organisation for Rare

Diseases <https://www.rarediseaseday.org/association/135>

Singapore - Rare Disorders Society <https://www.rdss.org.sg/>

Cook Islands - Cook Islands National Disability Council <https://cincd13.wordpress.com/about/>

Fiji - Fiji Disabled Peoples Federation <https://fdpf.org/>

Laos - Lao Disabled People's Association (LDPA) <https://www.facebook.com/LaosDisabledPeople/>

Marshall Islands Disabled Persons Organisation (no website)

Mongolia - Mongolian National Federation of Disabled People's Organizations

Nauru - The Nauru Disabled People's Association (NDPA)

<https://sites.google.com/site/naurudpa/who-we-are>

Papua New Guinea - PNG Assembly of Disabled Persons (ADP)

<https://www.disabledpeoplesinternational.org/NationalPapuaNewGuinea>

Samoa - Nuanua O le Alofa - <https://www.facebook.com/NOLASamoa/>

Solomon Islands - People with Disability Solomon Islands - <https://www.facebook.com/People-with-Disability-Solomon-Islands-630711223738555/>

Tonga - Tonga mental health and disabilities association - <https://www.facebook.com/TMHDA/>

Tuvalu - Tuvalu Disabled Persons Organisation (No website)

### EURO Region

Albania - Rare Diseases Association, <http://rda-al.com/en/>

Anorra - Andorra Association of Rare Diseases - <https://ammaandorra.wixsite.com/amma>

Azerbaijan - Sanofi in Azerbaijan <https://ammaandorra.wixsite.com/amma>

Belarussian Organization of Patients with MPS and Other Rare Genetic Disorders -

<https://www.eurordis.org/content/belarussian-organization-patients-mps-and-other-rare-genetic-disorders-0>

Bosnia and Herzegovina - Alliance for Rare Disease of the Republic Srpska - <http://savezzarijetke.rs/>

Bulgaria - National Alliance of People with Rare Diseases (NAPRD)- <https://rare-bg.com/>

Croatia - Rare Diseases Croatia - <http://www.rijetke-bolesti.hr/rare-diseases-croatia/>

Georgia - Georgian Foundation for Genetic and Rare Diseases -

<http://resourcerepository.org/organization/view/1051>

Iceland - Einstok Born - <https://www.einstokborn.is/is/english>

Kazakhstan - Patients with Cancer and Rare Diseases Support Association (No website)

Latvia - Latvian Alliance for Rare Diseases, Rare Disease Association - <https://retasslimibas.lv/>

Lithuania - Rare Disease Lithuania - <http://retosligos.lt/>

Luxembourg - Maladies Rares Luxembourg - <https://www.alan.lu/home/home/rare-diseases/en/>

Malta - National Alliance for Rare Disease Support Malta - <https://rarediseasesmalta.com/>

Montenegro - National Organisation for Rare Diseases -

<https://www.rarediseaseday.org/association/859>

North Macedonia - National Alliance for Rare Diseases of North Macedonia -

<https://www.rarediseaseday.org/association/357>

Portugal - Aliança Portuguesa De Associações Das Doenças Raras

(<http://www.aliancadoencasraras.org/>)

Federação Portuguesa De Doenças Raras (<https://fedra.pt/>)

Russia - Russian Association of Rare Diseases (<http://www.rare-diseases.ru/>)

Serbia - National Organisation of Rare Diseases Serbia (<http://www.norbs.rs/>)

Slovenia - Viljem Julijan Association for Children with Rare Diseases (no website)

Turkey - Nadir Hastalıklar AGI (<https://www.nadirhastaliklaragi.org.tr/duyurular>)

Ukraine - NGO 'Rare diseases of Ukraine' (<https://www.rarediseaseday.org/association/10>)

### SEARO Region

Indonesia - Indonesia Rare Disorders

- [https://www.facebook.com/IdRareDisorders/about/?ref=page\\_internal](https://www.facebook.com/IdRareDisorders/about/?ref=page_internal)

Sri Lanka - Rare Disease Forum - Sri Lanka College of Paediatricians - <https://slcp.lk/rare-disease-forum/>, Kosala Dullewe Foundation for Children with Special Needs-

<https://kosaladullewafoundation.org/>, SOULINK LANKA - <https://www.facebook.com/soulinklk/>

Bhutan - Ability Bhutan Society <http://absbhutan.org/>

## EMRO Region

Iran - Rare Disease Foundation of IRAN (RADOIR) - <http://radoir.org/en/>

Pakistan - Bin Adam Foundation (all rare diseases but especially Ataxia) - [http://www.binadam.net/about\\_us.htm](http://www.binadam.net/about_us.htm)

UAE - United Arab Emirates Rare Disease Society - <http://uaerds.ae/ar/>

Bahrain – Al Jawhara Centre - <https://www.agu.edu.bh/en/Academics/CMMS/College-Related-Centers/Pages/The-HH-Al-Jawhara-Centre-for-Molecular-Medicine-and-Inherited-Disorders.aspx>

Egypt - Egyptian Society of Human Genetics - <http://www.nshg-society.eg.net/>

Lebanon - Rare Disease Day hosted by Lebanese Association for Neuromuscular Diseases - <http://www.landforhope.org/>

Libya - Rare Disease Day, Stronger Together hosted by Tripoli Children's Hospital -

Morocco – Rare Disease Day hosted by Espor Vml Maroc

<https://www.rarediseaseday.org/association/372>

Oman – Association of Early Intervention for Children with Disability - <http://www.aei.org.om/>

Pakistan – Bin Adam Foundation - <http://www.binadam.net/>

Saudi Arabia - Rare Disease Day organised by WERATHAH - <http://www.werathah.com/>

Tunisia - Rare Disease Day hosted by

[http://www.pasteur.tn/index.php?option=com\\_content&view=category&layout=blog&id=41&Itemid=147&lang=fr&limitstart=70](http://www.pasteur.tn/index.php?option=com_content&view=category&layout=blog&id=41&Itemid=147&lang=fr&limitstart=70)

Qatar – Hamad Medical Cooperation - <https://www.hamad.qa/EN/news/2019/February/Pages/HMC-and-Heidelberg-to-Establish-National-Center-for-Rare-Disease.aspx>

## PAHO Region

Costa Rica - Enfermedades Raras Costa Rica- <https://www.facebook.com/EnfermedadesRarasCR/>

Dominican Republic - Fundacion Maria Laura - <http://fundacionmarialaura.com/>

Guatemala - Association Para Todos - <http://www.asociacionparatodos.org/>

Panama - Ayoudas Panama - <https://www.panamaparaninos.com/servicios/Fundaciones-y-ONGs-para-ninos/Ayoudas-Panama>

Peru - Federacion Peruana de Enfermedades Raras -

<https://www.facebook.com/enfermedadesrarasperu/>

Uruguay - ATUERU - <https://www.facebook.com/ATUERU>

Venezuela - Federacion Venezolana de Enfermedades Poco Frecuentes -

<https://www.facebook.com/enfermedadesrarasvenezuela/>

Bolivia – Instituto De Genetica - [https://www.umsa.bo/institutos-umsa/-](https://www.umsa.bo/institutos-umsa/-/asset_publisher/1pMoOCPnVT2d/content/instituto-de-genetica)

[/asset\\_publisher/1pMoOCPnVT2d/content/instituto-de-genetica](https://www.umsa.bo/institutos-umsa/-/asset_publisher/1pMoOCPnVT2d/content/instituto-de-genetica)

Ecuador - Rare Disease Day hosted at Hospital De Especialidades Carlos Andrade Marin -

<http://hcam.iess.gob.ec/>

Paraguay – Rare Disease Day hosted by Asociacion Paraguay De Pacientes Con Enfermadades

Lisosomales - <https://www.facebook.com/ASOCIACION-PARAGUAYA-DE-PACIENTES-CON-ENFERMEDADES-LISOSOMALES-275910139110910/about/>

## **4.3 Recommended Papers**

### **Rare Diseases at the Global Level**

Julkowska, D et al. “The importance of international collaboration for rare diseases research: a European perspective.” *Gene therapy* vol. 24,9 (2017)

Merinopoulo, E., Cox, A. ‘How Social Media can be Used to Understand People With Rare Diseases,’ Evidera, (2019) pg1-4

### **Regions - General**

Khosla, N, Valdez, R. ‘A compilation of national plans, policies, and government actions for rare diseases in 23 countries, Intractable and Rare Diseases Research, Volume 7,4, (2018)

### **SEARO Region**

Akmal, A, S et al. ‘State of Rare Disease Management in Southeast Asia, Orphanet Journal of Rare Diseases, (2016)

### **AFRO Region**

Baynam et al, ‘A call for global action for rare diseases in Africa,’ *Nature Genetics*, Volume 55 (2020)

### **WPRO Region**

Akmal, A, S et al. ‘State of Rare Disease Management in Southeast Asia, Orphanet Journal of Rare Diseases, (2016)

### **EMRO Region**

‘Orphan Disease in the Middle East and African Region,’ IQVIA, (2018.)

### **PAHO Region**

Chaves, P, A., ‘Rare Diseases in Latin America: Challenges and Opportunities for the equitable attention and proposal of Patients Organizations,’ ISPOR, (2017.)

### **EURO Region**

Julkowska, D et al. “The importance of international collaboration for rare diseases research: a European perspective.” *Gene therapy* vol. 24,9 (2017)

## **Identity**

Morel, T., Cano, S.J. Measuring what matters to rare disease patients – reflections on the work by the IRDiRC taskforce on patient-centered outcome measures. *Orphanet J Rare Dis* 12, 171 (2017)

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