

Unleashing future potential: Prader-Willi syndrome in Africa

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Introduction

Prader-Willi syndrome (PWS) is characterised by the absence of function of specific genes on the chromosome 15 inherited from the father. It impacts intellectual, social, and physical development. Although this syndrome is recognised to be apparent at birth, the symptoms are highly variable – not all individuals are impacted in the same way. Statistically one in 15,0000 to 25,000 babies are born with PWS and it affects races and genders equally.

Features in infancy include poor sucking/feeding and hypotonia (limp muscles). PWS is associated with delay in development and in the acquisition of abilities ranging from poor motor skills, impaired language development and mid intellectual ability. Physical impacts also include underdeveloped genitalia. A significant effect appearing from the early years is hyperphagia, known as excessive eating. There is currently no cure for PWS, but there are several ways to manage the severity of the symptoms. It is important to note, the ease of managing a condition like PWS requires awareness, resources, and great specialist expertise.

Given the complexity and lack of awareness of this rare genetic condition, the role of the International Prader-Willi Syndrome Organisation (IPWSO) worldwide is crucial. IPWSO is a non-profit membership organisation providing support for people with PWS, families, professionals and national PWS associations with contacts in over 100 countries and 45 country members. The first conference, held in 1991, brought together 22 countries and involved families, individuals with PWS and professionals building a sense of solidarity amongst stakeholders as the years progressed¹. The collaboration of professionals and families has enabled a transfer of knowledge highly valuable in diagnosis and management.

Through free diagnosis, individual grants, training programmes, social media platforms, research, and international advocacy, such as conferences and workshops, IPWSO works to ensure there is support for all who need it.

¹ International Prader-Willi Syndrome Association (2021). *IPWSO 30th Anniversary film: 1991 to 2021*. [online] www.youtube.com. Available at: <u>https://www.youtube.com/watch?v=o6_2iyD6aK8</u>

The Project: background

Brief

My project has focused on the diverse continent of Africa, with a specific focus on six countries – Algeria, Egypt, Kenya, South Africa, Senegal, and Morrocco. I have aimed to produce a report which will serve as a useful document to IPWSO in visualising the current research links as well as support for and from family groups, specialist bodies or associations engaged in working around PWS or people living with rare diseases (PLWRD) in general. The content of this report is comprised of a review of peer-reviewed articles, interviews with international contacts via email and Zoom and other information I have collated during my short internship.

Outlook

Through the course of the four weeks of my internship I aimed to both enhance and inform future valuable work for IPWSO by consolidating their current links and support in Africa. Being part of an organisation that is completely new to me, and delving into a new context with a task at hand, I aimed to maintain independence, persistence, and confidence. Although working with IPWSO for only a short period, strengthening my professional skills such as networking, verbal skills and communication are the most crucial lifelong skills I want to develop and experience.

For IPWSO this report should provide an overview of the current support links in Africa, but also an identification of the gaps in support. I hope that my discussions with contacts and members of IPWSO can then provide recommendations and a greater understanding on how to manage or fill the current gaps within the continent of Africa.

Focus

As mentioned, my project has a specific interest in Africa incorporating six countries which contrast in context. Morocco, Algeria and Egypt as North African countries, Senegal in West Africa, Kenya, and South Africa in sub-Saharan Africa, serve to highlight the great diversity in Africa, which is highly insightful to explore. The context amongst the countries is different, meaning the knowledge and expertise varies too. It is important to understand the gaps and visualise the challenges that IPWSO's contacts in these countries face and to consider the level of support they offer or receive. Meetings in the early stage of my internship expressed the need for raising the profile of PWS in Africa and expanding networks to ensure support is inclusive and meets the needs of all. Ultimately this should make my project useful to IPWSO, giving me a chance to study new material and knowledge.

The layout of my report will examine the continent of Africa at large, providing an overview of actors involved. This includes different organisations, associations, specialist bodies, civil society, and international bodies. This will be replicated with more depth when discussing the six countries I have chosen.

Key organisations

Global actors providing support in Africa:

• The United Nations (UN)

The United Nations is an intergovernmental organisation founded in 1945 made up of 193 member states. The main goals of the United Nations are to uphold global peace and security, advance the welfare of people worldwide, and foster international collaboration in pursuit of these objectives². Most recently in 2021³, the UN General Assembly formally adopted the recognition of 300 million people with rare diseases worldwide and their families. This placed the rare disease community on the agenda of the UN. Member states voted for policies supportive of rare diseases including to:

- Improve healthcare systems for universal access to high-quality and affordable care.
- Empower individuals with rare diseases, promoting rights, inclusivity, and well-being.
- Ensure access to safe and effective health products for rare diseases.
- Foster expert networks and specialized hubs for rare diseases.
- Implement national policies to support individuals with rare diseases and share best practices.
- Enhance international collaboration for rare disease research and data sharing.

• Non-Governmental Organisations

International action to promote the implementation of the UN General Assembly Resolution was promoted by rare disease patient organisations. Through campaigns by civil society partners such as the NGO committee for rare diseases, Rare



Diseases International and EURORDIS, the rare disease community launched a global challenge. Anders Olauson⁴, chair of the NGO committee stated:

"We must ensure that the rights of people living with a rare disease are respected and that we are not discriminated against because patient population numbers seem small for each rare disease. In total, there are 6,700 rare diseases forming a community of over 300 million people."

² Nations, U. (14AD). Chapter I: Purposes and Principles (Articles 1-2). [online] United Nations. Available at: <u>https://www.un.org/en/about-us/un-charter/chapter-1#:~:text=To%20achieve%20international%20co%2Doperation</u>.

³ Ostapenko, S. (2021). UN Resolution is adopted: what does it mean for Europe? [online] EURORDIS. Available at: <u>https://www.eurordis.org/un-resolution-is-adopted-what-does-it-mean-for-europe</u>

⁴ NGO Committee for Rare Diseases (2019). *News*. [online] NGO Committee for Rare Diseases. Available at: <u>https://www.ngocommitteerarediseases.org/news/#:~:text=Anders%200lauson%2C%20Chair%20of%20the</u>

And Yann Le Cam⁵, chief executive of EURORDIS declared:

"We call on the UN to adopt a resolution at the General Assembly that will formally make rare diseases a global priority..."

The campaign for the resolution lasted three years involving persistent advocacy from civil society. To ensure compliance, rare disease patient organisations around the world urged for change at local, national, and international level, framing the needs of individuals with rare diseases and their families.

Rare Diseases International (RDI)

RDI⁶ (Rare Diseases International) serves as a global advocate for rare diseases, coordinating collective initiatives to ignite international policy transformations. They facilitated the adoption of the first-ever UN Resolution on Persons Living with Rare Diseases and their Families (PLWRD), and their community also engages with the World Health Organisation (WHO).

The WHO⁷ are a specialised agency of the United Nations responsible for public health and made up of 194 member states. Collaboration between RDI and the WHO, enhances awareness of rare diseases, strengthens health systems, and encourages policy development of international and national policies for PLWRD.



 ^{5 5} NGO Committee for Rare Diseases (2019). News. [online] NGO Committee for Rare Diseases. Available at: https://www.ngocommitteerarediseases.org/news/#:~:text=Anders%20Olauson%2C%20Chair%20of%20the
 ⁶ Rare disease International (14AD). Rare Diseases International. [online] Rare Diseases International. Available at: https://www.rarediseases.org/news/#:~:text=Anders%20Olauson%2C%20Chair%20of%20the

⁷Rare Disease International (14AD). Global Collaborations Tools. [online] Rare Diseases International. Available at: <u>https://www.rarediseasesinternational.org/global-collaborations-tools/#:~:text=RDI%20working%20groups</u>

African organisations:

Although Africa may benefit from international organisations and NGOs, there are many NGOs advocating for PLWRD within the continent of Africa specifically. Below are examples of non-profit organisations which act as professional bodies aiming to voice the needs of the professionals and the patients they serve.

African Medical Association⁸ (AfMA)

AfMA was established as the unified voice of African physicians, aiming to advocate for improved health standards across the continent. Taking a proactive stance, AfMA prioritises actionoriented projects recognising the pressing healthcare challenges in Africa. It actively collaborates with the World Health Organization (WHO) and other pertinent organisations to



maximise opportunities for the benefit of African patients. They align with policies of the World Medical Association (WMA), the global umbrella organisation for physicians.

Society for the Advancement of Sciences in Africa⁹ (SASA)

SASA, the Society for the Advancement of Science in Africa, is a not-for-profit association that brings together scientists, academic institutions, research institutions, government agencies, philanthropists, and funding agencies. Its core purpose is to develop a robust scientific knowledge base in Africa and propel scientific advancements across the continent. SASA serves as a leading organisation, advocating for scientific progress in Africa and establishing connections with the global scientific community. By promoting scientific research, SASA contributes to sustainable social and economic development in Africa.

Doctors with Africa CUAMM¹⁰

Founded in 1950, the organisation advocates for the universal right to health and promotes international solidarity, justice, and peace. The focus is on improving the well-being and health of underserved communities in Africa with a sustainable development approach. They prioritise action on reproductive, maternal, new-born, child, and adolescent health; nutrition; infectious diseases; NCDs; and Universal Health Coverage (UHC).

⁸ Africa Medical Association (14AD). *AfMA - Africa Medical Association*. [online] africama.net. Available at: <u>https://africama.net/</u>

⁹International Social Council (1945). Society for the Advancement of Science in Africa (SASA). [online] International Science Council. Available at: <u>https://council.science/member/sasa-society-for-the-advancement-of-science-in-africa/#:~:text=The%20Society%20for%20the%20Advancement%20of%20Science%20in%20Africa%20is,the%20frontiers %20of%20science%20in</u>

¹⁰ Doctors with Africa CUAMM (1950). *Doctors With Africa CUAMM*. [online] Doctors with Africa CUAMM. Available at: <u>https://doctorswithafrica.org/</u>.

Rare disease and more specialised organisations:

The International Rare Diseases Research Consortium (IRDiRC)

The IRDiRC¹¹ is a global collaborative initiative launched in 2011 by the European Commission and the US National Institutes of Health to tackle rare diseases through research and accomplish the vision to enable all people living with a rare disease to receive an accurate diagnosis, care, and available therapy within one year of coming to medical attention. Today, the Consortium gathers 60 member organisations from all continents involving stakeholders from Africa.

IRDiRC, had two main goals: developing 200 new therapies and improving diagnostic capabilities for most rare diseases by 2020. Progress exceeded expectations, achieving the therapy goal in 2017 and nearing the diagnostic target. Now, IRDiRC has set new goals for 2017-2027:



- 1. Diagnose all known rare diseases within one year of medical attention and create a global diagnostic pipeline.
- 2. Approve 1,000 new therapies, prioritising diseases without treatment options.
- 3. Develop methods to assess the impact of diagnoses and therapies on rare disease patients.

This requires fundamental changes in scientific approaches and collaboration. IRDiRC members are committed to these changes and seek support for global rare disease research advancements.

African Society for Paediatric and Adolescent Endocrinology (ASPAE)

ASPAE¹² specifically addresses the unique healthcare needs and challenges faced by the endocrinology professionals working with paediatric and adolescent patients in Africa, considering factors including regional prevalence, cultural aspects, and access to healthcare resources. ASPAE is a fairly new and growing professional body. The goal is to provide specialised care and improve the health outcomes of children and adolescents with endocrine disorders across the continent. ASPAE hold annual conferences expanding information and raising awareness, which IPWSO attended in early 2023. IPWSO has had contact with Dr Suzanne Sap, President of ASPAE, noting that there is currently little information on the diagnosis or development of babies born with PWS in much of Africa. The most recent conference (their 14th) was held in Cameroon in February 2023 with the theme of: *"Diabetes and endocrine diseases in African children: the continuum of care from neonate to adulthood in the era of technological innovation."*

¹¹ International Rare Diseases Research Consortium (2011). *IRDiRC – International Rare Diseases Research Consortium*. [online] irdirc.org. Available at: <u>https://irdirc.org/</u>

¹²African Society for Paediatric and Adolescent Endocrinology (2023). ASPAED – African Society for Paediatric and Adolescent Endocrinology. [online] www.aspaed.org. Available at: <u>http://www.aspaed.org/</u>

The Union of National African Paediatric Associations (UNAPSA)

UNAPSA¹³ is the body of African paediatricians represented by National Paediatric Societies and Associations in Africa. To date they have a 38-country membership. UNAPSA is affiliated to the International Paediatric Association (IPA). IPA stated at the 76th World Health Assembly that *"universal health coverage"* and *"prevention and control of non-communicable diseases and mental health"* were items on their agenda.

African Society for Human Genetics (AfSHG)

The AfSHG¹⁴ strives to empower the African scientific community and policymakers by providing them with essential information and practical expertise to actively participate in genetics research. Additionally, they aim to raise global awareness about the valuable contributions made by African scientists in the field of genetics.

African Academy of Neurology (AFAN)

The African Academy of Neurology¹⁵ has a mission to serve as a unified platform for African neurologists by collaborating with their respective National Neurological Societies. AFAN aims to advance the field of neurology by promoting best practices and fostering training opportunities for neurologists in Africa for example through the use of Regional Training Courses (RTCs).

African International Division¹⁶ (AID)

Their purpose is to share information and promote discussion about psychiatry within the Division.

Project ECHO

The ECHO Model¹⁷ is a collaborative and sustainable learning approach that transcends disciplines. Participants join virtual sessions to present cases, exchange recommendations, and learn from each other. This continuous learning cycle fosters long-term impact. ECHO

connects specialists from diverse fields, promoting a comprehensive knowledge-





¹³ Union of National African Paediatric Societies and Associations (2022). Union of National African Paediatric Societies and Associations | UIA Yearbook Profile | Union of International Associations. [online] uia.org. Available at: https://uia.org/s/or/en/1100014190

¹⁴ African Society for Human Genetics (2003). AfSHG – African Society of Human Genetics – The African Society of Human Genetics is a forum for scientists interested in Human Genetics in Africa to meet, interact, network, and collaborate. [online] www.afshg.org. Available at: <u>https://www.afshg.org</u>

¹⁵ The African Academy of Neurology (2008). *Home*. [online] www.afaneurology.org. Available at: <u>https://www.afaneurology.org/</u>

¹⁶African International Division (2023). African international division. [online] www.rcpsych.ac.uk. Available at: <u>https://www.rcpsych.ac.uk/international/international-divisions/african-international-division</u>

¹⁷ IPWSO, P. ECHO (2003). *About Project ECHO*[®]. [online] IPWSO. Available at: <u>https://IPWSO.org/how-we-can-help/project-ECHO/about-project-</u>

<u>ECHO/?gad=1&gclid=CjwKCAjw5MOlBhBTEiwAAJ8e1hNkN2oMONIC3oolcWeFrjIvNcwnwcyl9uwcJM-zTD9NJs904Qil5xoC6nAQAvD_BwE</u>

sharing approach. In Africa, the ECHO network comprises 160+ organisations across 29 countries, facilitating programs in all 54 countries. ECHO serves as a catalyst for transforming healthcare systems in Africa, driven by passionate ECHO Champions and subject matter experts. Ministries, universities, and organisations rely on ECHO as a vital digital learning tool for a healthier and more prosperous future.

IPWSO successfully utilises Project ECHO's methodology finding real potential in fostering connections within the PWS community, enhancing comprehension of the syndrome, and exchanging information regarding care, treatments, and innovations worldwide.

N.B.: There is little empirical evidence that these professional bodies or NGOs advocating for PLWRD have much knowledge of Prader-Willi syndrome. IPWSO also does not have relationships with all of these associations, but developing links is a recommendation for enhancing awareness. My choice of these specialist bodies and research provides an identification of the relevant associations more likely to be linked or aware of PWS in their advocacy, training, and work.

Contact mapping in Africa



<u>Key:</u>

- Strong contacts in Morrocco and South Africa who are full IPWSO country members with established associations. Although Egypt is not a full member currently, they are in the process of establishing their association and connections are strong.
- Some contacts in some countries refers to some communication with IPWSO, the presence of IPWSO delegates or contacts within other NGOs there.
- Few contacts or none can be reflective of little or no connections, network, or correspondence.

The map provides a visual understanding for IPWSO of their contacts in Africa. It helps to gauge a better idea of how strong these contacts are, and where links can be made to enhance coverage of support for PWS.

Country support: in more depth

The next part of my report goes beyond discussing the continent of Africa as a whole. Through capitalising on zoom meetings, email, and my own independent research I was able to gauge a better understanding of the circumstances in particular countries. Across the continent of Africa, all countries differ in health expenditure, research specialist bodies, associations, organisations, and family support.

My analysis on each country follows this structure:

- 1) General context of country
- 2) Literature review on available research
- 3) Current networks/bodies in support of PWS
- 4) Impact of the country's IPWSO association
- 5) Challenges faced
- 6) Relations to IPWSO
- 7) Recommendations/feedback on filling and managing the support gap.

South Africa

1) Overview of South Africa

Healthcare system



In 2023/2024 the budget allocation was reduced by R4,4 billion from 2022/2023¹⁸. This decrease raised serious concerns about the healthcare system as the national Treasury has acknowledged that the health sector is underfunded. Although, South Africa's constitution guarantees access to health services for all citizens through public and private healthcare, private health in South Africa accounts for the largest share of health expenditures through out-of-pocket payments and medical schemes.

South Africa continues to face challenges that have been damaging to the quality of healthcare available including¹⁹:

- HIV/AIDS epidemic forcing the system to focus on this issue rather than funding other areas.
- Workforce shortage of medical graduates meaning inadequate provision for the population size.
- Too few psychiatrists to meet the country's mental health care needs, 80% of psychiatrists are working in the private sector a much higher proportion than is usually seen.
- Challenges for research (discussed later).

¹⁸ News, H. (2023). *Health budget allocation down R4.4-billion*. [online] Health-e News. Available at: <u>https://health-e.org.za/2023/05/09/health-budget-allocation-down-r4-4-billion/</u>

¹⁹ Maphumulo, W.T. and Bhengu, B.R. (2019). Challenges of quality improvement in the healthcare of South Africa postapartheid: A critical review. *Curationis*, [online] 42(1). doi:https://doi.org/10.4102/curationis.v42i1.1901

- Only 58% of people living in sub-Saharan Africa have access to safe water supplies.
- Noncommunicable diseases (hypertension, heart disease, diabetes) are on the rise; and injuries remain among the top causes of death in the Region.
- Variability in skill sets between rural and urban areas leading to geographical immobility for families in rural areas requiring medical attention.
- Suboptimal care levels and patient management.

2) Literature review

My independent research led me to three published scientific research papers discussing Prader-Willi syndrome. Research papers can reflect the level of expertise, knowledge, or awareness of the syndrome. It can also provide a sense of priority or importance given to PWS. Research can identify gaps, challenges, and future recommendations to understand PWS further. Hence, studying literature published, despite its challenges, can build a better picture and understanding of the context at hand.

Below are the research papers found on PubMed. The paper from Tanzania is included as a neighbouring country.

- Christianson AL, Viljoen DL, Winship WS, de la Rey M, van Rensburg EJ. Prader-Willi syndrome in South African patients--clinical and molecular diagnosis. S Afr Med J. 1998 Jun;88(6):711-4. PMID: 9687849. https://pubmed.ncbi.nlm.nih.gov/9687849/
- Segal D; Paediatric and Adolescent Endocrine and Diabetes Society of South Africa. Guideline for using growth hormone in paediatric patients in South Africa: treatment of growth hormone deficiency and other growth disorders. S Afr Med J. 2009 Mar;99(3 Pt 2):187-95. PMID: 19563097. <u>https://pubmed.ncbi.nlm.nih.gov/19563097/</u>
- Elichilia R. Shao, Lucas F. Kiyegi, Amos O. Mwasamwaja, Kajiru Kilonzo, Ben C. J. Hamel, "Respiratory Failure due to Severe Obesity and Kyphoscoliosis in a 24-Year-Old Male with Molecularly Confirmed Prader-Willi Syndrome in Tertiary Hospital in Northern Tanzania", Case Reports in Genetics, vol. 2017, Article ID 2348045, 4 pages, 2017. <u>https://doi.org/10.1155/2017/2348045</u>

Interview with Karin Clarke - Trustee of IPWSO since 2022

Through my discussions with Karin, I understand there is no research into the syndrome currently going on in South Africa. She has been in touch with Rare Diseases South Africa (RDSA) about clinical trials, but there is no current activity. She explained the difficulty in doing research where there are minimal financial incentives for families to be part of research or clinical trials. Similarly, where families must pay the expenses for their child with PWS, there is no time or motivation to join research when there are other more pressing priorities such as work.

3) Current networks

Karin has networks in place to support South African people with PWS and their families. She mentioned there is a PWS support group, referrals from rare disease associations which she has collaborated with before, and two large hospitals for diagnosis. These hospitals are situated in Pretoria and Cape Town. In Gauteng there is another hospital where diagnosis can occur as well. Karen has a close relationship with the genetic counsellors and can give information to use to determine diagnosis. There is awareness amongst professionals.

There is also good family support. Prader-Willi syndrome Support South Africa is an organisation of families and professionals dedicated to working together offering support, awareness, education, and advocacy for people living with PWS in the country (and beyond). Karin also mentioned there are three WhatsApp groups. One for parents with children under five years, one general of all ages and another including four adults with PWS. It is a valuable resource to share information and for parents to support each other. It is a place for families (including Karin) to turn to others for any questions, queries and support.

Through my own research there are some associations possibly affiliated or know about PWS:

- Paediatric Neurology and Development Association of Southern Africa²⁰
- o International Child Neurology Association²¹
- Rare Diseases South Africa²²

4) Impact of the association

Resources and services

The association deliver many types of resources and services to people with PWS and their families such as:

- A welcome pack when people first contact after diagnosis this includes guidance and positive information.
- Contacts are provided for parents to maintain a network amongst parents WhatsApp groups.
- Supplying information for dieticians, schools plus school visits.
- As PWS is so rare, using other channels such as autism networks to distribute information and resources.

²⁰ o Paediatric Neurology and Development Association of Southern Africa (2023). The Paediatric Neurology and Development Association of Southern Africa (PANDA-SA). [online] www.pandasa.org. Available at: https://www.pandasa.org/web/index.asp

https://www.pandasa.org/web/index.asp ²¹ International child neurology association (2023). ICNApedia - Home. [online] ICNApedia. Available at: https://www.icnapedia.org/icnc2024

²² Rare Disease South Africa (2023). Healthcare | Rare Diseases South Africa | Charity. [online] Rare Diseases SA. Available at:

https://www.rarediseases.co.za/?gclid=CjwKCAjw5MOlBhBTEiwAAJ8e1pr9dEGZFQBniRnsSrIq_5iSjQwEB2ihLxcRL64 Z4TI7HTpvWPmW6xoCGk4QAvD_BwE

There is also help from the government including:

- Grants families can apply to if they earn under a certain amount for under 18's small amount per month (around £10 GBP).
- Grants from the age of 18 +.
- As medical care can be expensive, keeping a record of expenses to get deducted from tax owed.



Achievement stories

Karin discussed a few inspiring stories about the care and treatment of people with PWS. For example, she mentioned that residential homes for people with PWS can successfully manage obesity. To Karin, this emphasises that there is work to be done to raise awareness that such interventions can be successful in making a difference to people's health despite there being no cure for PWS.

Other stories illustrate how it is possible to have a healthy lifestyle and live successfully without Growth Hormone Therapy; an adult, aged 40, who had no access to Growth Hormone Therapy completed 130 5k Park Runs, and another adult completed 26.



<u>Advocacy</u>

Each May, as part of the PWS global awareness campaign, the PWS support group in South Africa celebrates "Going Orange for PWS". This awareness day is well supported locally. Karin also advocated-for the approval of Growth Hormone Therapy, however this was impossible with medical insurers refusing to pay and no government support.

Main Impacts

For families there is a sense of togetherness and unity. As South Africa, and Africa as a whole, is geographically highly diverse it is hard to get together physically. Creating a community online is a practical way to get people connecting, spreading knowledge, experience and providing support for both families and professionals.

5) Challenges faced

There are many challenges in achieving the universal support for PWS in South Africa. Karin highlighted a few:

Failure of Growth Hormone Therapy (GHT) to be approved:

This limits the extent of help available to families. In South Africa the government has a duty to finance certain treatments. However, as PWS is a rare disorder it is deemed uneconomical and getting approval for PWS treatments remains difficult. It is not being pushed for by the professionals or pharmaceutical industry either. What is not made clear is that the issue for people with PWS needing GHT is not simply height but muscle strength and muscle tone. Although Karin tried to contact rare diseases organisations to help, they have not been successful so far.

Incorrect diagnosis:

People with PWS may have symptoms of autism, and misdiagnosis can occur; when children are at the age of six to seven years old, they are refereed to neurodiverse centres in hospitals and the diagnosis of PWS is not immediately recognised.

Statistics for diagnosis:



There are currently no statistics for diagnosis as geneticists do not have a national database. The association is in contact with 90 families who are part of the support group, although they are also contacted by many people who have not received a diagnosis. Diagnosis occurs when a person is referred to one of the five larger hospitals. To get a diagnosis, families must travel long distances, which is not possible for many. Paediatrics in private hospitals lacks specialist knowledge resulting in further under-diagnosis. Consequently, diagnosis at birth is unlikely. Rather it is picked up later when the child may find it difficult to keep up in school, develops hyperphagia or other physical symptoms.

Family hesitancy and culture:

When families first get diagnosed, there is great shock. The time to process the news can prevent families from reaching out. Sometimes it is the grandparents who initially reach out as the parents are scared to hear news. They consider PWS to only offer bad news, when the PWS support group can actually provide hope and positive news. There are some families who may join later but some remain isolated.

Karin explains that the lack of technology, time and inclination can all prevent families from reaching out. Cultural / societal factors are also important. Within many parts of Africa, a community brings up a child, but a child with PWS may be shunned by the wider group, even associated with witchcraft, stigma, and suspicion in some communities. This can make families hide their children and prevent them accessing diagnosis and treatment. Another cultural issue to navigate is the glamorisation of obesity especially in Sub-Saharan Africa where South Africa has the highest obesity prevalence. As food insecurity is a major issue with many impoverished communities, there is a culture for celebrating overweight people and access to food. This is problematic on many levels.

Taxation issues:

Tax relief on medical expenses is available but this policy is of little benefit to the poorer families who struggle to even afford the cost of health care.



Domino effect of lack of awareness:

Lack of awareness leads to under-diagnosis. People do not know where to access support because they have not heard the term Prader-Willi and do not know to access resources about it.

Lack of research:

Research within a country can identify specific contextual factors. This may be useful in determining how best to go about providing support, diagnosis, or treatment in a

country. With most published research into PWS coming from North America and Europe (though this is now changing) it means that the experiences and particular needs of people with PWS in African countries have not been represented in research and that treatments and procedures may not be best tailored to their needs.

6) Relations to IPWSO

Karin is on the Board of Trustees, so she has a strong relationship with IPWSO. IPWSO has helped her contact pharmaceutical companies, fund workshops and when local group chats are irresponsive, IPWSO experts can provide her the support needed. Karin has a child with PWS, but people with PWS experience a diverse range of symptoms and have different needs, so as well as relying on her personal experience and knowledge it is very useful to be able to refer to IPWSO to support the needs of families in her association.

7) How to fill the gaps in support

Recommendations to improve and widen the support include:

- Building relationships with other centres working in areas such as autism to spread knowledge of PWS in the hope that more people can be reached.
- Motivate people to complete online surveys and share their experiences.



- Growth Hormone Therapy to be pushed further and approved.
- Work together with bigger organisations (for example with rare disease bodies) to access more resources.
- Engage more volunteers and professionals to generate ideas and opportunities.
- Research conducted should involve a collaboration between the community and professional bodies within South Africa and Western institutions.

Egypt:

1) Overview of Egypt

Healthcare system:

Egypt's Minister of Finance and Head of the General Authority for the Universal Health Insurance system, Mohamed Maait, announced the government will raise healthcare spending by 15% in 2023/24²³. Egypt's rapidly growing economy has led to the health sector being seen as a priority for the country's development agenda.

The government has steadily worked towards achieving Universal Health Coverage, although for decades Egypt has been facing multiple challenges within their healthcare systems such as²⁴:

²³ Ahramonline (2023). Egypt to increase FY 2023/24 healthcare spending by 15 percent: Minister - Health - Egypt. [online] Ahram Online. Available at: <u>https://english.ahram.org.eg/NewsContent/1/1236/502283/Egypt/Health/Egypt-to-increase-FY-healthcare-spending-by-perc.aspx</u>

²⁴ Columbia University Irving Medical Centre (2021). EGYPT | Summary. [online] Columbia University's Mailman School of Public Health. Available at: <u>https://www.publichealth.columbia.edu/research/others/comparative-health-policy-library/egypt-summary#:~:text=The%20public%20health%20system%20faces.</u>

- Doctors demanding better wages, working conditions and training opportunities leading to brain drain.
- Corruption and lack of accountability.
- Inadequate health insurance.
- Geographical imbalance of skilled health care workers.
- System has increasingly been forced to respond to emerging illnesses, such as non-communicable diseases that take priority.
- Insufficient integration of services into primary healthcare.
- Widespread poverty.

2) Literature review

Unlike South Africa where I found three published research papers on PubMed, I only found one independently. This published research paper in 2019 was on Early Detection and Management of Prader-Willi Syndrome in Egyptian Patients²⁵. The first listed author of the article was Dr Hala T El-Bassyouni. She is a Professor of Clinical Genetics, Division of Human Genetics and Genome Research, National Research in Cairo. As I found minimal research, I contacted her to ask if there was any further research being conducted or published. She replied revealing three more published papers and added that there are ongoing studies yet to be published, a positive development for PWS research in Egypt.

The three other published papers are also highly scientific and technical in nature.

- Measurement of Serum Chemerin, Oxidized LDL, and Vitamin D Levels in Prader–Willi Syndrome: A Cross-Sectional Study in Paediatric Egyptian Patients Manal M. Thomas Moushira E. Zaki Eman Youness Khaled Hamed Azzah A. Khedr Phoebe M. Abd El-Massieh Sara M. Abdo Hala T. El-Bassyouni²⁶.
- DNA Damage and Neutrophil Elastase in Children with Prader-Willi Syndrome Moushira Erfan Zaki *, Eman Youness, Mohamed Gadelhak, Marwa Shehab, Safinaz El-Toukh, Doaa Soliman, Walaa Yousef and Hala El-Bassyouni²⁷.
- Behavioural problems, biochemical, and anthropometric characteristics of patients with Prader–Willi syndrome Ebtissam M. Salaha, Hala T. El-Bassyouni b, Shams Kholoussic , Marwa Shehabd and Wafaa A. Kandeele²⁸.

²⁵ El-Bassyouni, H.T., Hassan, N., Mahfouz, I., Abd-Elnaby, A.E., Mostafa, M.I. and Tosson, A.M.S. (2019). Early Detection and Management of Prader-Willi Syndrome in Egyptian Patients. *Journal of Pediatric Genetics*, [online] 8(4), pp.179–186. doi:https://doi.org/10.1055/s-0039-1695042.

²⁶ Thomas, M.M., Moushira Erfan Zaki, Youness, E.R., Hamed, K.H., Khedr, A.A., Abd, P.M., Abdo, S.M. and El-Bassyouni, H.T. (2020). Measurement of Serum Chemerin, Oxidized LDL, and Vitamin D Levels in Prader–Willi Syndrome: A Cross-Sectional Study in Paediatric Egyptian Patients. Thieme, 10(01), pp. e187–e195. doi:https://doi.org/10.1055/s-0040-1718896.

²⁷ Zaki, M.E., Youness, E., Gadelhak, M., Shehab, M., El-Toukhy, S. and Soliman, D. (2019). DNA Damage and Neutrophil Elastase in Children with Prader-Willi Syndrome. *Biomedical and Pharmacology Journal*, [online] 12(04), pp.1967–1974. Available at: <u>https://biomedpharmajournal.org/vol12no4/dna-damage-and-neutrophil-elastase-in-children-with-prader-willisyndrome</u>

²⁸ Salah, E.M., El-Bassyouni, H.T., Kholoussi, S., Shehab, M. and Kandeel, W.A. (2015). Behavioral problems, biochemical, and anthropometric characteristics of patients with Prader–Willi syndrome. *Middle East Journal of Medical Genetics*,

This research involved clinical trials and examination of case studies to develop greater expertise and knowledge of PWS.

Interview with Walaa Mohammed and Sarah Kasaby – member/adviser of PWS Egypt and the Middle East



The association is not currently undertaking any research. If there was to be research, priorities would include studies into behavioural management and therapies, all other therapies, hyperphagia, perhaps genetics and also the medical side of PWS and the possible cure.

3) Current Networks

As the PWS Association for Egypt and the Middle East is in the initial stages of formation, they do not have many connections or contacts with other organisations, medical bodies, or associations. This is a work in progress for Egypt with exciting future plans. There is a WhatsApp group chat for the mothers, building unity and a support system.

An example of a future contact they have considered is The National Research centre – the largest multidisciplinary research and development centre in Egypt dedicated to basic and applied research in key areas of interest.

Through my own independent research, I have highlighted a few bodies in Egypt that may not be aware of PWS but can learn through collaborations with the new association and/or IPWSO. This can be a way for Egypt to make contacts and build the awareness that they need for greater support as a new organisation. These organisations are:

- The Egyptian Scientific Foundation of Rare Diseases in Children Care for the Rare²⁹
- Project Hope³⁰
- Egyptian Association for International Medical Studies (EAIMS)³¹
- Sobi Rare Strength³²

4) Impact of Association

Current resources and services:



[[]online] 4(2), p.63. Available at:

https://www.academia.edu/21932217/Behavioral_problems_biochemical_and_anthropometric_characteristics_of_patients_w_ith_Prader_Willi_syndrome_

²⁹ The Egyptian Scientific Foundation of Rare Diseases in Children Care for the Rare (2023). The Egyptian Scientific Foundation Of Rare Diseases In Children Care for the Rare. [online] www.esfrd.org. Available at: <u>https://www.esfrd.org</u>
³⁰ Project Hope (2023). Learn about Project HOPE's work in Egypt. [online] Project HOPE. Available at: <u>https://www.projecthope.org/country/egypt/</u>

³¹ Egyptian Association for International Medical Studies (2007). Egyptian Association for International Medical Studies (EAIMS). [online] www.eaims.net. Available at: <u>https://www.eaims.net</u>

³² Sobi (2023). Sobi Egypt | Sobi Egypt. [online] www.sobi.com. Available at: <u>https://www.sobi.com/egypt/en</u>

To access support, individuals and communities can interact with their work through attending Zoom meetings, and access resources and translated materials on their website. They can also be in contact through their WhatsApp and Facebook group and page.

Future plans for greater awareness:

The group is very new, and they are at the planning stage rather than being able to deliver a lot of support. They have many plans to support individuals affected by PWS and their families. They would like to provide medical support and help families access the therapies they need. Currently, they are planning a conference to help educate parents and caregivers. They hope their work can be of benefit not only to Egypt, but also to other Arab-speaking countries.

Achievement Stories:

Currently, they are in the first phase of creating an organisation that will hopefully service Egypt and the Middle East. They have registered as an official organisation, agreed a name and logo and identified office premises, which is a strong starting point. Some success stories of individuals with PWS include a young boy diagnosed at age 5 who had become obese. He was able to lose the weight and is living happily now as a 13-year-old, playing different sports including boxing. In another case, a girl diagnosed earlier also managed to keep her weight under control. She is now 11 and also doing well at school. This provides a great sense of hope, that Egypt simply needs greater PWS awareness and knowledge to support families.

Main impacts:

Sarah and Walaa highlighted that there has been a great amount of awareness raised in the last two years but a lot more work still needs to be done.

5) Challenges faced

Some of the challenges in providing support for PWS include:

Lack of resources:

There is not a lot of funding to be able to raise the awareness in all the ways that they would like to. Social media is the main form of making more individuals aware of the syndrome. Egypt lacks therapy facilities and if an individual does go to therapy this is highly costly. This means families can be provided with the care and support by organisations like the association and IPWSO, but when it comes to managing the condition professionally this is difficult and costly to families. Even with the money, there is little local expertise in PWS, making management harder.

Family hesitancy:

There are cultural factors that can impact families being aware of their child's condition. There is great stigma on the child having "special needs" which some parents may see as taboo. Although Sarah and Walaa emphasise their role is to change this rhetoric and approach families by maintaining



sensitivity, there is nothing they can do when a family has particular preconceptions or

resistance to outside support or openness. For example, parents can be reluctant to share photos or videos of their child for use on social media as they do not want the community knowing,

Issues of diagnosis:

There are no statistics for diagnosis, meaning the number of people with PWS in Egypt is ambiguous – people are going undiagnosed and families and individuals with PWS are not receiving the care and support they need. Diagnosis in Egypt usually occurs between the ages of two to four years. Although this seems fairly early, it is later than in some other countries, and late enough to mean it can be more difficult for families to adapt once diagnosis occurs.

<u>Advocacy:</u>

There is currently no advocacy work on legislating for the rights and inclusion of individuals with PWS in Egypt, though the association have seen campaigns around other disorders. The lack of awareness and openness is a barrier to families receiving support.

6) Relations to IPWSO

Sarah and Walaa state: "We are grateful for the support we are getting so far from IPWSO. Without IPWSO none of this would have been able to see the light. We got granted two microgrants from IPWSO specifically to help us start the organisation and create the workshop needed. We thank IPWSO and hope to continue working together and strengthen the ties."

7) How to fill the gaps in support – raising greater awareness

The main goal Sarah and Walaa have highlighted is the need for their voice to be heard. Some recommendations they mentioned include:

- Partnering or collaborating with organisations to spread knowledge, learn and upscale diagnosis rate.
- Creation of a video, brochures, written material for doctors, health centres, schools etc.
- Continue Zoom meetings with IPWSO.
- Fund interpreters for workshops, online platforms, conferences.
- As funding is minimal, workshops can be broadcast online rather than burdening families with the costs of flights and accommodation.
- Future plans with greater funding may include a conference that is for professional bodies rather than only parents and caregivers.
- Gain a connection, e.g., an appearance/mention on a famous TV talk show well known in Egypt and try to generate awareness and fundraise.

Now that PWS Egypt is established, they need to be heard!



Morocco



1) Overview of Morocco

Healthcare system:

Morocco's healthcare budget for 2023 is set to increase to MAD 28 billion (\$2.6 billion)³³. The majority of the funds will be allocated to reforming the national healthcare system, creating jobs, and raising salaries for healthcare workers. New healthcare facilities and university hospitals are also planned. The budget includes provisions for medication supplies and the removal of taxes on select medications. Social security initiatives aim to support disadvantaged children, vulnerable families, and expand healthcare coverage. These efforts demonstrate Morocco's commitment to enhancing healthcare access and addressing the needs of vulnerable populations. Challenges in Morocco's healthcare system³⁴:

- Unequal distribution of resources
- Inadequate funding
- Healthcare worker shortages
- Rise of non-communicable diseases
- Poor education system

2) Literature review

I had difficulty in finding research papers. The only research paper I came across is:

The Diagnosis and Genetic Mechanisms of Prader-Willi Syndrome: Findings
 From a Moroccan Population Study by Mohamed Ahakoud, Hanae Daha Belghiti,
 Ayoub Nedbour, Abdelhamid Bouramtane, Sana Chaouki, Laila
 Bouguenouch and Karim Ouldim³⁵.

This study highlights the importance of early diagnosis, the underlying genetic mechanisms of PWS and the support needed for PWS requiring further research.

<u>Discussion via email with Hanae Bellahcen of PWS Morocco - full member of IPWSO</u> Hanae revealed she is also not aware of any research on the syndrome. It is not well known in Morocco nor seen as a priority for research unfortunately. She only knows of 30 people who are diagnosed, but the numbers are definitely higher. If there were to be research, she recommends it to be concentrated on hyperphagia.

3) Current networks



³³ News, J.R.-M.W. (2021). Morocco's Healthcare Budget Set at \$2.6 Billion in 2023. [online] https://www.moroccoworldnews.com/. Available at: <u>https://www.moroccoworldnews.com/2022/10/351977/moroccos-healthcare-budget-set-at-2-6-billion-in-2023</u>

³⁴ News, S.K.-M.W. (2023b). Majority of Moroccans Are Dissatisfied with Health, Education Sectors. [online] https://www.moroccoworldnews.com/. Available at: <u>https://www.moroccoworldnews.com/2023/01/353424/majority-of-moroccans-are-dissatisfied-with-health-education-sectors</u>.

³⁵ Ahakoud, M., Belghiti, H.D., Nedbour, A., Bouramtane, A., Chaouki, S., Bouguenouch, L., Ouldim, K., Ahakoud, M., Belghiti, H.D., Nedbour, A., Bouramtane, A., Chaouki, S., Bouguenouch, L. and Ouldim, K. (2023). The Diagnosis and Genetic Mechanisms of Prader-Willi Syndrome: Findings From a Moroccan Population Study. *Cureus*, [online] 15(4). doi:https://doi.org/10.7759/cureus.37866

Hanae mentioned that there is a rare disease group that has just been created with their first meeting scheduled for October. They also have a foundation for people with disabilities, which focuses on rehabilitation. This is dependent on availability in the city, so access is an issue.

There are still organisations and bodies which could be involved in PLWRD which Morocco can contact. My own research has highlighted:

- The Alliance of Rare

Diseases in Morocco³⁶ (AMRM)

- Sobi Rare strength³⁷
- Moroccan Society of Neurology³⁸
- Moroccan Association of Dynamic Psychiatry³⁹
- The Moroccan Society of Genomics and Human Genetics (SM2GH)⁴⁰

4) Impact of the association

Resource and services:

The honorary president who is a doctor generously supports the schooling of 10 children in specialised centres. Parents can reach out to the association through WhatsApp, Facebook, and can also access website resources.

Achievement stories:

The story that gives Hanae hope is that of her 27-year-old son who graduated school and continued into higher education in Canada. He was able to live alone and be autonomous. He manages his diet, does sports, socialises and works. As a mother being part of the association can provide hope to other parents.

Main impact:

The main impact was giving hope to parents who have just experienced the diagnosis and challenges of the syndrome and for them to accept the syndrome and learn how to manage it. This is their main objective.

³⁶ RESERVED, I.U. -- A.R. (2023). Orphanet: Alliance%20Maladies%20Rares%20Maroc. [online] www.orpha.net. Available at:

https://www.orpha.net/consor/www/cgibin/SupportGroup_Networks.php?lng=EN&data_id=125959&title=Alliance%20Mal adies%20Rares%20Maroc

³⁷Sobi (2023b). Sobi Morocco. [online] www.sobi.com. Available at: https://www.sobi.com/morocco/en

 ³⁸Moroccan Society of Neurology (2022). Moroccan Society of Neurology • Information - Associations • Rabat, Rabat.
 [online] www.medpages.info. Available at: https://www.medpages.info/sf/index.php?page=organisation&orgcode=375694
 ³⁹Moroccan Association of Dynamic Psychiatry (2020). Morocco – World Association for Dynamic Psychiatry WADP.
 [online] wadpinternational.com. Available at: https://wadpinternational.com/branches/morocco

⁴⁰The Moroccan Society of Genomics and Human Genetics (2023). SM2GH. [online] www.sm2gh.ma. Available at: https://www.sm2gh.ma

5) Challenges faced

<u>Low diagnosis:</u>

There are no diagnosis statistics at the moment in the country. The diagnosis is usually made according to the knowledge of paediatricians. When diagnosis occurs it usually happens late, at around age of five to six years. In 2003 there were only four samples sent to the lab for diagnostic testing.



Rights for people with PWS:

Although they try their best to raise awareness about the disease, the underdiagnosis has made Hanae believe the rights of these children cannot be guaranteed. Rights will only be established when there is greater awareness of PWS.

Poor support:

There is a lack of professional bodies and resources which are needed to care for children who are the upmost priority. The lack of connections and support from other organisations also plays a part in how far the improvements in care and support for the syndrome of PWS will go.

6) Relations to IPWSO

The association in Morocco is a full member of IPWSO, but as a fairly new member they are concerned that there is still a lot of work to be done to integrate their association into the wider organisation and to gain the support that they desperately need for the families in their community.

Want Morocco wants of IPWSO:

- Support with awareness raising activities.
- Factual and statistical information to support their work.
- The promotion and advance of scientific research and the opportunity to participate in this.

7) How to fill the gaps in support

- They need external support for funding and getting access to items including glasses, devices, and treatments.
- Greater connections with professional bodies.
- Build networks with other rare disease organisations.
- Mediate between medical professionals and families to ensure information is transferred and understood.
- Attempt to record statistics of diagnosis.
- Create a stronger relationship with IPWSO.

Algeria

1) Overview of Algeria Healthcare system: In 2022, Algeria's president Abdelmajid Tebboune signed the 2023 budget into law. A 25% increase in the budget for 2022 laid out government spending of \$98 billion⁴¹. This budget will be split amongst the public sector from healthcare, education, and social welfare. The exact amount towards healthcare is yet to be known. Despite an increase in expenditure Algeria's healthcare suffers from a range of issues such as⁴²:

- Lack of management.
- Organisational issues and bureaucracy.
 Healthcare sector reliant on imported goods.
- Cardiovascular diseases, cancer, hypertension, diabetes are on the rise.
- Ineffective licensing of generic pharmaceuticals.
- Lack of co-ordination between Ministry of Health and the Algerian patent office which leads to uncertainty for registration of health products.



- Employment issues.

2) Literature review

Unfortunately, my research has not identified any published-research papers on PWS. This may be because no research has been conducted or my lack of contacts with Algeria has been a hindrance. I had difficulty in finding research on rare diseases in general within Algeria but was able to find some on obesity. I do not have enough information to conclude the reason for the lack of research available.

3) Current networks

IPWSO has contacts with Algeria developed through the ASPAE conference. Algeria has contacts with the African Society of Paediatric and Adolescent Endocrinology and their next conference will be held there.

Some possible bodies which may be useful:

- Algerian Society of Paediatric Ophthalmology and Strabismus⁴³
- Algerian Society of Neurology and Clinical Neurophysiology⁴⁴
- Arab-German young academy of science and humanities⁴⁵
- Sobi rare strength⁴⁶



⁴¹ Zine Labidine Ghebouli (2023). Algeria's 2023 budget: President Tebboune's make-or-break first-term project. [online] Middle East Institute. Available at: <u>https://www.mei.edu/publications/algerias-2023-budget-president-tebbounes-make-or-break-first-term-project</u>

⁴²Canada, G.A. (2022). Algeria health sector market profile. [online] GAC. Available at: <u>https://www.tradecommissioner.gc.ca/algeria-algeria-market-reports-etudes-de-marches/0006431.aspx?lang=eng.</u>

⁴³ Communications, A. and swdadmin (2019). Algerian Ophthalmology Society. [online] WSPOS | World Society of Paediatric Ophthalmology and Strabismus. Available at: <u>https://www.wspos.org/algeria/</u>

⁴⁴ Algerian Society of Neurology and Clinical Neurophysiology (1975). *Member Societies – PAUNS*. [online] pauns.org/en/member-societies. Available at: <u>https://pauns.org/en/member-societies/</u>

⁴⁵ Arab-German young academy of science and humanities (2023). Agya-Website. [online] Arab-German Young Academy. Available at: <u>https://agya.info/</u>

⁴⁶ Sobi (2023). Sobi Algeria. [online] www.sobi.com. Available at: <u>https://www.sobi.com/algeria/</u>

- Alliance Algérienne contre les Maladies Rares⁴⁷

4) Impact of association – no current PWS association

Resources and services:

IPWSO has some local contacts.

The next conference hosted by ASPAE will hopefully take place in 2024 in Algiers.

Diagnostic testing:

Since 2003, there were 35 samples sent to the IPWSO lab for diagnostic testing from Algeria. This is a fairly large number in contrast to other countries. This shows that there is some medical awareness of the signs and symptoms and that there are a number of diagnosed people in the country, though access to diagnosis locally is clearly an issue.

5) Challenges faced

Lack of support:

There is no organised PWS group, just individuals and professionals.

Lack of research:

As far as I can gather, there is not any research happening within the field of PWS or rare diseases in general.

6) Relations to IPWSO

Algeria has contacts with IPWSO. There are professional contacts and a family who has contacted a staff member at IPWSO. However, there is no association IPWSO is aware of and contact is fairly sporadic.

7) How to fill the gaps in support

Through looking at previous documents where Algeria has contacted IPWSO, they seem to largely ask for more support. They also wanted help arranging an awareness day.

There are some recommendations that can help expand support locally including:

- Developing connections with other organisations, professionals, or medical centres.
- Promoting research to develop expertise of PWS.
- Workshops and conferences to raise awareness.

⁴⁷ Alliance Algérienne contre les Maladies Rares (2023). *Alliance Algérienne contre les Maladies Rares*. [online] Rare Disease Day 2023. Available at: <u>https://www.rarediseaseday.org/friends/alliance-algerienne-contre-les-maladies-rares/#:~:text=About%20Alliance%20Alg%C3%A9rienne%20contre%20les</u>

Kenya:

1) Overview of Kenya

Healthcare system:

As a repercussion of the recession and the Covid-19 pandemic, the Kenyan budget is geared towards bringing the economy into recovery. Although there are increases to social spending it is still a downwards trend from 2019 given the burden of debt⁴⁸. In Kenya where over 43% of people live in poverty, health challenges include⁴⁹:

- High maternal and child mortality
- High burden of infectious diseases HIV, Tuberculosis, Malaria -
- Supply chain setbacks facilities are understaffed.
- Poor healthcare financing limits innovation and technology

2) Literature review

There is no research available on PWS in Kenya. There are many active organisations who may have more information on research.

3) Current networks

IPWSO has connections with two people in particular, one being Esther Maina a geneticist and IPWSO advisor, and Dr Paul Laigong a member of the board of ASPAE and an IPWSO delegate. Kenya is well connected with professional networks and Rare Disorders Kenya is very active. However, there is no PWS association and little information about levels of awareness.

There are other active organisations who can be of support in raising awareness for PWS:

- Kenya Paediatric Association ⁵⁰ _
- Rare Disorders Kenya (RDK)⁵¹ _
- Kenya Psychiatrist Association (KPA)⁵²
- African Child Neurology Association⁵³
- Kenya Association of Physicians⁵⁴ -
- Non-Communicable disease Alliance Kenya⁵⁵

⁴⁸ World Bank (2022). Kenya Economic Update (KEU). [online] World Bank. Available at: https://www.worldbank.org/en/country/kenya/publication/kenya-economic-update-keu.

⁴⁹ Awiti, J.O. (2014). Poverty and health care demand in Kenya. BMC Health Services Research, 14(1). doi: https://doi.org/10.1186/s12913-014-0560-y

⁵⁰ Kenya Paediatric Association (2023). KPA | Kenya Paediatric Association. [online] kenyapaediatric.org. Available at: https://kenyapaediatric.org

⁵¹ Rare Disease Kenya (2023). RDK | Rare Disorders Kenya. [online] RDK | Rare Disorders Kenya. Available at: https://rarediseasekenya.org/#:~:text=Rare%20Disorders%20Kenya%20(RDK)%20is 52 Kenya Psychiatrist Association (2023). Kenya Psychiatrist Association (KPA). [online] kenyapsychiatrist.org. Available at:

https://kenyapsychiatrist.org

⁵³ African Child Neurology Association (2023). ICNApedia - ACNA. [online] ICNApedia. Available at: https://www.icnapedia.org/my-groups/60-acna

⁵⁴ Kenya Association of Physicians (2021). Home - Kenya Association of Physicians. [online] kapkenya.org. Available at: https://kapkenya.org

⁵⁵Non-Communicable disease Alliance Kenya (2019). NCD Alliance Kenya. [online] NCD Alliance. Available at: https://ncdalliance.org/ncd-alliance-kenya

There are also five national hospitals in Kenya⁵⁶:

- Moi Teaching and Referral Hospital.
- National Spinal Injury Referral Hospital.
- Kenyatta National Hospital.
- Mathare National Teaching and Referral Hospital.
- Kenyatta University Teaching and Referral Hospital.

4) Impact of association – no current PWS association

Professional links:

Our contacts have knowledge of PWS but there is little knowledge of how many people may be diagnosed with the syndrome locally.

Due to my limited timescale and as with Algeria, I was not able to connect with an individual to obtain information on the support or care available to people with PWS. However, as there are some contacts within the country to IPWSO, there is some awareness of PWS and the need to support families and increase the spread of knowledge.

5) Challenges faced

Diagnostic testing:

We are not aware of any statistics. IPWSO's diagnosis service has been used on a couple of occasions.

6) Relations to IPWSO

There are some good contacts for IPWSO within Kenya and there is potential to develop these further, however it would be good to also be in contact with families.

7) How to fill the gaps in support

Some possible recommendations to increase coverage of support for PWS includes:

- Clear route and expertise around diagnosis within Kenya rather than internationally.
- Collaborations with medical centres, organisations, and associations to enhance spread of knowledge and gain new contacts.
- Promote scientific research.
- Strengthen current contacts through regular contact.
- Organisation of workshops to increase awareness.



⁵⁶ Wikipedia Contributors (2019). *Healthcare in Kenya*. [online] Wikipedia. Available at: <u>https://en.wikipedia.org/wiki/Healthcare_in_Kenya</u>.

Senegal

1) Overview of Senegal

Healthcare system:

Senegal in 2022, approved the 2023 financing bill focusing on four main issues - one being healthcare⁵⁷. The Covid-19 pandemic has since been contained, but the strength of the health system is in some doubt.

As with many countries in Africa, Senegal faces challenges to their healthcare system including⁵⁸:

- Major human resources failure leading to shortage of health workers, which is more prominent in remote and rural areas.
- Failure to retain health workers in remote areas.
- Limited availability of safe, quality medicines to treat patients.
- Prominent disease of Malaria.
- High rates of infant malnutrition.

2) Literature review

There are no recent available papers on PWS in Senegal.

3) Current networks

My limited contact with Senegal has made my understanding of the current networks unclear. I am aware there is a good contact from ASPAE 2023.

Some other organisations that may be involved or contacted for further support on PWS:

- Centre for Genomic regulation⁵⁹
- Imed Hospitales⁶⁰
- Senegalese Neurology Society ⁶¹
- Nestle Nutrition Institute in Africa⁶²



⁵⁷ Mehlmann-Wicks, J. (2022). COVID-19: Impact of the pandemic on healthcare delivery. [online] The British Medical Association is the trade union and professional body for doctors in the UK. Available at: <u>https://www.bma.org.uk/advice-and-support/covid-19/what-the-bma-is-doing/covid-19-impact-of-the-pandemic-on-healthcare-delivery</u>.

⁶⁰ Hospitales, I. (2023). *IMED Valencia Hospital diagnosis and treat a rare disease on a Senegalese child* | *Group IMED Hospitales: Private Hospitals in Alicante, Murcia y Valencia.* [online] www.imedhospitales.com. Available at: <u>https://www.imedhospitales.com/en/actualidad/imed-valencia-hospital-diagnosis-and-treat-a-rare-disease-on-a-senegalese-child</u>

⁵⁸ Paul, E., Ndiaye, Y., Sall, F.L., Fecher, F. and Porignon, D. (2020). An assessment of the core capacities of the Senegalese health system to deliver Universal Health Coverage. Health Policy Open, [online] 1, p.100012. doi:https://doi.org/10.1016/j.hpopen.2020.100012

⁵⁹ Centre for Genomic regulation (2023). CRG - Centre for Genomic Regulation | EU-LIFE. [online] eu-life.eu. Available at: <u>https://eu-life.eu/about/members/crg-centre-genomic-regulation</u>

<u>child</u> ⁶¹ Senegalese Neurology Society (2023). *WFN Member Societies: Pan-African Region*. [online] wfneurology.org. Available at: <u>https://wfneurology.org/wfn-membership/member-societies/wfn-member-societies-pan-african-region</u>

⁶² Nestle Nutrition Institute in Africa (2017). Senegal: Congress of Senegalese Paediatric Society. [online] nnia.nestlenutrition-institute.org. Available at: <u>https://nnia.nestlenutrition-institute.org/news/article/2017/10/20/senegal-</u> <u>congress-of-senegalese-peadiatric-society</u>

- Senegalese Society of Human Genetics⁶³

4) Impact of association – no current PWS association N/A

5) Challenges faced

N/A

6) Relations to IPWSO

There has been some contact with IPWSO, as made through ASPAE.

7) How to fill the gaps in support

With limited information I can only suggest similar suggestions as in the previous sections.

⁶³ Senegalese Society of Human Genetics (2023). Senegal – AfSHG – African Society of Human Genetics. [online] www.afshg.org/about/member-societies/senegal. Available at: <u>https://www.afshg.org/about/member-societies/senegal</u>

Conclusion

The aim of my project was to examine research and current support links across the continent of Africa and then explore this in depth for six chosen countries. Some of the organisations I identified were focused on broader topics than PWS, such as rare diseases or medical specialities. This is because PWS is so rare and without significant numbers of people diagnosed it is important to tap into these broader networks. My identification of various associations, organisations, civil society, specialist bodies and families within each country becomes valuable in understanding the current networks, but also the gaps that need to be filled over time through solutions such as collaborating with these other organisations and actors.

Africa is a huge, diverse, and continent, so generalisations tend to be unhelpful. It was therefore important to consider the status of PWS in various specific countries to understand the national contexts. There are different networks, healthcare challenges, research, and awareness in each country I have explored. This clearly has an impact on the level of support families are receiving and how exactly countries can take action to enhance awareness of this rare disease.

My research has emphasised a few key recommendations to fill the gaps in support which will be of relevance across these countries. There is some work that IPWSO can do alone, but much needs to be done in partnership with the national associations where present, or to support the development of embryonic groups. For example, to:

- Promote and prioritise scientific research into PWS developing greater local expertise and encouraging the spread of information from guidance, management, diagnosis etc, as well as encouraging research to take place within these countries.
- Encourage the local collection of information about numbers of diagnoses and people living with PWS, which ultimately can be used by any developing PWS groups.
- Capitalise on new and relevant platforms other than Facebook and LinkedIn such as TikTok where short, educational, and engaging videos can be created generating greater awareness.
- The creation of tailored materials such as brochures or posters for professionals or families.
- Collaboration and connections to be made between medical centres, NGOs, families, and other stakeholders engaged in rare diseases.
- Support existing and developing PWS associations to become sustainable and ensure that other organisations know where to find PWS resources.



Reflections

This next part of my report is a discussion of my project journey, joining as a "Research Officer" for IPWSO for four weeks. In the early stages of my project, I scoped out the project creating the criteria of what I wanted to achieve. Throughout my time I was able to speak to international contacts abroad via Zoom and email, join a session of the ECHO programme, staff meetings, and had regular communication with my supervisor. My discussions were built on questions that could enhance my project and provide answers the internet could not. This was successful to me as I was able to speak to two countries via zoom – Egypt and South Africa, an email discussion with Morocco, engage in a new contact in Egypt (Dr Hala T El-Bassyouni) and hear from a range of staff members and advisers providing their input and feedback for me to look into.

As much as I gained a great amount from speaking to a variation of people, I was unable to have any meaningful contact with Kenya, Senegal, and Algeria. Though IPWSO does have contacts in these countries, the timescale of my project made it difficult to set up meetings with these busy professionals. This reinforces this important message that, IPWSO as a rather small but international organisation will find connections are at times minimal and difficult to reinforce. Also, that PWS is one rare disease amongst very many other diseases and health conditions. Professionals in particular will have other interests and priorities.

With only two African countries as full members, and Egypt in its early stages, other countries have IPWSO delegates, and they may be engaged to various degrees depending on their circumstances. To address this limitation, it is crucial for IPWSO to cultivate stronger relationships within individual countries and involve international actors who can advocate for PWS under the PLWRD umbrella term. The contact map is also a clear illustration of the limited contacts, that as an intern, I encountered challenges with.

Aside from these challenges, there is a lot I can take away from this experience. This was my first remote internship; I have learnt not only about the organisation's missions and values but about Prader-Willi syndrome itself.

I was interested in the role of technology in diagnosis, treatment and awareness and asked about this in interviews. For countries within Africa the lack of funding for healthcare means that technological advances may take longer to be adopted or may not be adopted at all. However, technology is being used for communication, particular the mobile phone, which can therefore be used to access information and raise awareness of conditions. For Western countries technological innovations are continually upgrading the healthcare systems from diagnosis to treatment. This was eye-opening for me as living in the UK all my life, healthcare is free at the point of use and highly accessible, yet this is clearly not the case in many countries.

It has been most enjoyable writing a report that has been built on my own conversations, research and most importantly my interest. My project has been very

flexible and based on what I am intrigued in making this so much more meaningful to me. I will always thank IPWSO, for taking me on board and giving me such an amazing opportunity.